COMPLETE GENE LIST WES NIJMEGEN DG 2.15 (4209 genes)

Releasedate: 31-01-2019

Gene	*GenePanel*	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
A2M	HEMOSTATIC/THROMBOTIC DISORDERS	119.2	99.5	98.5	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
A2ML1	HEREDITARY CANCER	130.8	100	99.7	No OMIM phenotype Noonan-like syndrome (Vissers et al. 2015) Noonan syndrome (van Trier (2015) Int J Pediatr Otorhinolaryngol, epub) Otitis media, susceptibility to (Santos-Cortez (2015) Nat Genet 47,917)
A4GALT	MENDELIOME	140.8	100	100	NOR polyagglutination syndrome, 111400 [Blood group, P1Pk system, p phenotype], 111400 [Blood group, P1Pk system, P(2) phenotype], 111400
AAAS	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	106.4	100	99.7	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	SKIN DISORDERS MENDELIOME	151.8	100	98.3	Keratoderma, palmoplantar, punctate type IA, 148600
AARS	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.3	100	99.6	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	126.2	100	99.3	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	128.8	99.6	97.4	Hyperlysinemia, 238700 Saccharopinuria, 268700
ABAT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	92.7	100	99.5	GABA-transaminase deficiency, 613163

ABCA1	MENDELIOME PRECONCEPTION SCREENING	123.6	100	99.5	HDL deficiency, type 2, 604091 Tangier disease, 205400 {Coronary artery disease in familial hypercholesterolemia, protection against}, 143890
ABCA12	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	140	99.6	97.8	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCA3	MENDELIOME PRECONCEPTION SCREENING	124	99.9	99.3	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.1	100	99.5	Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Stargardt disease 1, 248200 {Macular degeneration, age-related, 2}, 153800
ABCA5	MENDELIOME	59	93.1	79.6	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB10	IRON DISORDERS	75.2	75.6	67.1	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014)).
ABCB11	MENDELIOME PRECONCEPTION SCREENING	158.2	99.9	99.1	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	MENDELIOME PRECONCEPTION SCREENING	129.8	99.9	97.9	Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347 Gallbladder disease 1, 600803
ABCB6	SKIN DISORDERS MENDELIOME	127.2	100	99.6	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 [Blood group, Langereis system], 111600
ABCB7	MOVEMENT DISORDERS BONE MARROW FAILURE IRON DISORDERS MENDELIOME	131.5	99.9	98.4	Anemia, sideroblastic, with ataxia, 301310
ABCC2	MENDELIOME PRECONCEPTION SCREENING	135.8	100	100	Dubin-Johnson syndrome, 237500
ABCC6	VISION DISORDERS SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	116.4	93.6	92.6	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	EPILEPSY INTELLECTUAL DISABILITY	146.6	100	99.9	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176

	MENDELIOME PRECONCEPTION SCREENING				Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCC9	SKIN DISORDERS HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME RITME	157.9	99.9	99.2	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	76	74.7	68	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	METABOLIC DISORDERS	186.3	100	99.8	No OMIM phenotype
ABCD3	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.7	95.2	89.5	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	BONE MARROW FAILURE METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.6	99.9	98.3	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	145.2	100	99.2	Sitosterolemia, 210250
ABCG8	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.4	99.2	96.6	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ABHD12	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	107	97.3	88	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	209.6	100	99.9	Chanarin-Dorfman syndrome, 275630

	PRECONCEPTION SCREENING				
ABL1	MENDELIOME	140.2	100	99.9	Congenital heart defects and skeletal malformations syndrome, 617602 Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 0
ACACA	METABOLIC DISORDERS	135.6	98.2	97.5	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	141.5	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	135.2	98.4	95.7	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.3	98.8	95.6	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.9	99.3	97.6	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	119.1	99.5	95.6	2-methylbutyrylglycinuria, 610006
ACADVL	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	118.8	98.7	95.1	VLCAD deficiency, 201475
ACAN	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	121.6	91.6	85	?Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800
ACAT1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.7	99.2	94.6	Alpha-methylacetoacetic aciduria, 203750
ACAT2	METABOLIC DISORDERS	159	100	100	?ACAT2 deficiency, 614055
ACBD5	VISION DISORDERS BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS	145.3	97.8	96	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACD	BONE MARROW FAILURE	135.2	100	98.2	?Dyskeratosis congenita, autosomal dominant 6, 616553

	SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER				?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	120.7	99.5	97.4	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase], 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 {SARS, progression of}, 0 {Stroke, hemorrhagic}, 614519
ACER3	MENDELIOME PRECONCEPTION SCREENING	105.8	99.9	97.5	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	129.3	95.8	91.8	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	155.3	100	100	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	METABOLIC DISORDERS MENDELIOME	136.2	100	99.8	Bile acid synthesis defect, congenital, 6, 617308
ACP4	CRANIOFACIAL ANOMALIES MENDELIOME	64.5	90.2	82.7	Amelogenesis imperfecta, type IJ, 617297
ACP5	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	196.2	100	99.9	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128.8	99.9	99.3	Combined malonic and methylmalonic aciduria, 614265
ACSL4	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	104.7	97.5	91.8	Mental retardation, X-linked 63, 300387
ACSL6	MENDELIOME	118.5	99.8	98.4	Myelodysplastic syndrome, 0 Myelogenous leukemia, acute, 0

ACTA1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	99.7	99.2	95.3	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ACTA2	SKIN DISORDERS HEART PANEL MENDELIOME	137.6	100	99.8	Aortic , familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	MOVEMENT DISORDERS SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTC1	CARDIO CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	164.1	100	99.6	Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Left ventricular noncompaction 4, 613424
ACTG1	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	149.4	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ACTG2	MENDELIOME	133.3	99.7	97.6	Visceral myopathy, 155310
ACTL6A	INTELLECTUAL DISABILITY	126.3	99.5	96.8	No OMIM phenotype Marom R. et al., Hum Mutat. 2017 Oct 38(10:1365-1371)
ACTN1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	143.6	100	99.9	Bleeding disorder, platelet-type, 15, 615193
ACTN2	CARDIO HEART PANEL MENDELIOME	156.3	100	100	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158
ACTN4	RENAL DISORDERS MENDELIOME	143.7	99.5	97.7	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	CONGENITAL HEART DISEASE HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	165.1	100	100	Fibrodysplasia ossificans progressiva, 135100

ACVR1B	MENDELIOME	160.3	96.3	95.2	Pancreatic cancer, somatic, 0
ACVR2B	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	140.5	97.1	94.7	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	122.7	99.9	98	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	132.8	99.9	98.3	Aminoacylase 1 deficiency, 609924
ADA	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING SCID	113	98.9	97.3	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	101.4	99.9	99.1	?Sneddon syndrome, 182410 Polyarteritis nodosa, childhood-onset, 615688
ADAM10	SKIN DISORDERS MENDELIOME	123.6	94.6	92	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM17	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	139.4	97.6	93.8	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	140.5	99.9	98.6	?Epileptic encephalopathy, early infantile, 61, 617933
ADAM9	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	146.3	98.6	94.1	Cone-rod dystrophy 9, 612775
ADAMTS10	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	107.8	99.9	98.7	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS	99.9	96.3	91.7	Thrombotic thrombocytopenic purpura, familial, 274150

	MENDELIOME PRECONCEPTION SCREENING				
ADAMTS17	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	117.1	88.9	86.7	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.5	99.9	98.9	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.4	98.5	96.6	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	SKIN DISORDERS MENDELIOME	161	100	99.9	?Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTS9	CILIO RENAL DISORDERS	141.4	97.9	96.7	Nephronophthisis
ADAMTSL2	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	112.2	96.5	91	Geleophysic dysplasia 1, 231050
ADAMTSL4	 VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	90.6	99.9	98.8	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	MOVEMENT DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125	100	99.8	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADAT3	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	82.1	99.5	97.3	Mental retardation, autosomal recessive 36, 615286
ADCK5	METABOLIC DISORDERS	106.7	99.8	97.9	No OMIM phenotype
ADCY1	HEARING IMPAIRMENT MENDELIOME	145	94.7	93.3	?Deafness, autosomal recessive 44, 610154
ADCY10	RENAL DISORDERS	149.4	100	99.9	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADCY3	HH	130.4	99.8	98.4	{Obesity, susceptibility to, BMIQ19}, 617885
ADCY5	MOVEMENT DISORDERS HEART PANEL	129.2	92.3	89.1	Dyskinesia, familial, with facial myokymia, 606703

	METABOLIC DISORDERS MENDELIOME				
ADCY6	MENDELIOME	172.1	100	100	?Lethal congenital contracture syndrome 8, 616287
ADD3	MENDELIOME PRECONCEPTION SCREENING	169.7	100	99.7	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	MENDELIOME	160.5	96.7	96.1	Vibratory urticaria, 125630
ADGRG1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	149.7	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG2	MENDELIOME	96.5	97	92.3	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	MENDELIOME PRECONCEPTION SCREENING	147.6	99.8	98	Lethal congenital contracture syndrome 9, 616503
ADGRV1	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	140.3	99.5	97	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADIPOQ	MENDELIOME	139.5	100	100	Adiponectin deficiency, 612556
ADIPOR1	VISION DISORDERS	111.5	100	98.8	No OMIM phenotype syndromic retinitis pigmentosa (Xy (2016) Hum Mutat 37(3):246-249
ADK	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	100.4	99.5	96.1	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	INTELLECTUAL DISABILITY MENDELIOME	242.8	100	100	Helsmoortel-van der Aa syndrome, 615873
ADPRHL2	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	163.7	100	99.9	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRA2B	MENDELIOME	199.9	100	100	Epilepsy, myoclonic, familial adult, 2, 607876
ADRB2	MENDELIOME	131.3	100	100	Beta-2-adrenoreceptor agonist, reduced response to, 0 {Asthma, nocturnal, susceptibility to}, 600807 {Obesity, susceptibility to}, 601665
ADSL	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	183.6	99.2	99.1	Adenylosuccinase deficiency, 103050

ADSSL1	MENDELIOME PRECONCEPTION SCREENING	113.6	89.8	85.9	Myopathy, distal, 5, 617030
AEBP1	MENDELIOME	134.7	99.8	97.9	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	INTELLECTUAL DISABILITY MENDELIOME	122.8	99.9	98.9	Mental retardation, X-linked, FRAXE type, 309548
AFF4	INTELLECTUAL DISABILITY MENDELIOME	110.7	99.4	97.3	CHOPS syndrome, 616368
AFG3L2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	121	91.9	84.9	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AFP	MENDELIOME	100.2	94.2	84	Alpha-fetoprotein deficiency, 615969 [Hereditary persistence of alpha-fetoprotein], 615970
AGA	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.2	100	100	Aspartylglucosaminuria, 208400
AGBL1	VISION DISORDERS MENDELIOME	132.3	98.5	98.4	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	111	100	99.8	Retinitis pigmentosa 75, 617023
AGK	VISION DISORDERS HEART PANEL METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	146.7	99.7	98	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGO2	INTELLECTUAL DISABILITY	128.5	99.1	99.1	No OMIM phenotype {Epithelial ovarian cancer,reduced risk,association with} (Permuth-Wey (2011) Cancer Res 71,3896)
AGPAT2	SKIN DISORDERS	109.5	99	95.1	Lipodystrophy, congenital generalized, type 1, 608594

	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				
AGPS	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	51.7	96.8	84.8	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	114.8	95.2	89.3	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	214.2	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	 RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	134.6	92	91.9	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.5	99.9	99.2	Hyperoxaluria, primary, type 1, 259900
AHCY	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.5	100	99.8	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	INTELLECTUAL DISABILITY MENDELIOME	120.9	98	96.3	Xia-Gibbs syndrome, 615829
AHI1	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.3	99.2	95.1	Joubert syndrome 3, 608629
AHSG	MENDELIOME	177	100	99.8	?Alopecia-mental retardation syndrome 1, 203650
AICDA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	139	89.8	82.6	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	HEARING IMPAIRMENT NEUROPATHIES INTELLECTUAL DISABILITY	106.2	100	99.7	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614

	MENDELIOME MITOCHONDRIAL DISORDERS				
AIMP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	84.8	97.3	89.7	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	INTELLECTUAL DISABILITY MENDELIOME	119.1	93.6	86.9	Leukodystrophy, hypomyelinating, 17, 618006
AIP	MENDELIOME HEREDITARY CANCER	154.2	99.9	99	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102000
AIPL1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	116	100	99.5	Cone-rod dystrophy, 604393 Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393
AIRE	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	68.2	98.9	92	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	119.8	100	99.2	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING SCID	111.8	99.8	96.6	Reticular dysgenesis, 267500
AK7	MENDELIOME	125.9	99.3	94.5	?Spermatogenic failure 27, 617965
AKAP9	HEART PANEL MENDELIOME RITME	98	98.3	94.2	?Long QT syndrome-11, 611820
AKR1C2	DSD MENDELIOME PRECONCEPTION SCREENING	179.5	96.3	89.6	46XY sex reversal 8, 614279
AKR1D1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	106.1	98.5	94.3	Bile acid synthesis defect, congenital, 2, 235555
AKT1	SKIN DISORDERS MENDELIOME	156.5	99.9	99.5	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500

AKT2	MENDELIOME	148.8	100	99.5	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	79.6	97.8	88.6	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.6	99.8	97.4	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME	89.7	99.6	97.1	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	MENDELIOME PRECONCEPTION SCREENING	156	99.9	98.7	Analbuminemia, 616000 [Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	MOVEMENT DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	131.1	100	99.9	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH1A2	CONGENITAL HEART DISEASE HEART PANEL	114.8	100	99.6	No OMIM phenotype Tetralogy of Fallot (Pavan (2009) BMC Med Genet 10, 113) Pentalogy of Cantrell (Steiner (2013) J Med Case Rep 7,287) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89,476)
ALDH1A3	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	104.7	93.4	89.6	Microphthalmia, isolated 8, 615113
ALDH1B1	MITOCHONDRIAL DISORDERS	206.6	100	100	No OMIM phenotype Bladder cancer (Nickerson (2014) Clin Cancer Res 20,4935)
ALDH2	METABOLIC DISORDERS MENDELIOME	124.7	100	99.7	Alcohol sensitivity, acute, 610251 {Esophageal cancer, alcohol-related, susceptibility to}, 0 {Hangover, susceptibility to}, 610251 {Sublingual nitroglycerin, susceptibility to poor response to}, 0
ALDH3A2	MOVEMENT DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	125.7	95.3	94.6	Sjogren-Larsson syndrome, 270200

	PRECONCEPTION SCREENING				
ALDH4A1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	116	100	98.6	Hyperprolinemia, type II, 239510
ALDH5A1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	87.6	86.4	80.1	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.3	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	77.1	93.7	85.4	Epilepsy, pyridoxine-dependent, 266100
ALDOA	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.1	76.3	74.7	Glycogen storage disease XII, 611881
ALDOB	SKIN DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	165.7	100	99.4	Fructose intolerance, hereditary, 229600
ALG1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	50.9	53.6	48.8	Congenital disorder of glycosylation, type Ik, 608540
ALG10	METABOLIC DISORDERS	304.9	100	100	{Long QT syndrome, acquired, reduced susceptibility to}, 613688
ALG11	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.6	96.7	96	Congenital disorder of glycosylation, type Ip, 613661
ALG12	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	156.2	100	100	Congenital disorder of glycosylation, type Ig, 607143

ALG13	EPILEPSY PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	86.7	98.7	94.1	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG14	METABOLIC DISORDERS MENDELIOME	233.8	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	115.9	100	100	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	96.4	96	93.3	Congenital disorder of glycosylation, type Ic, 603147
ALG8	METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	126	96.5	95.1	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.3	100	99.6	Congenital disorder of glycosylation, type Il, 608776 Gillessen-Kaesbach-Nishimura syndrome, 263210
ALK	HEREDITARY CANCER	128.8	99.4	98.1	{Neuroblastoma, susceptibility to, 3}, 613014
ALKBH1	MITOCHONDRIAL DISORDERS	113.8	100	99.4	No OMIM phenotype Arumugam et al ESHG 2018
ALMS1	VISION DISORDERS CILIO HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY	179.8	99.9	99.7	Alstrom syndrome, 203800

	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
ALOX12B	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.6	100	99.5	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	122.2	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	HEART PANEL MENDELIOME	98.7	94.6	92.5	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	156.4	100	100	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALS2	ALS MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	170.2	99.9	99.2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ALX1	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	153.2	99.9	98.4	?Frontonasal dysplasia 3, 613456
ALX3	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	102.7	73.3	70.9	Frontonasal dysplasia 1, 136760
ALX4	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	132.7	98.4	92.5	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	157.9	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	175.1	94.6	88.5	Amelogenesis imperfecta, type IF, 616270
AMELX	CRANIOFACIAL ANOMALIES SKIN DISORDERS	98.2	99	95	Amelogenesis imperfecta, type 1E, 301200

	MENDELIOME				
AMER1	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	96.9	99.8	98.9	Osteopathia striata with cranial sclerosis, 300373
AMH	DSD MENDELIOME	42.1	92.8	74.2	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	DSD MENDELIOME	158	100	99.5	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	72.2	99	94	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	66.8	83.5	71.6	Megaloblastic anemia-1, Norwegian type, 261100
AMPD1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	126.7	100	99.9	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135.5	99.9	99.2	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMPD3	METABOLIC DISORDERS	131.9	99.8	98.6	[AMP deaminase deficiency, erythrocytic], 612874
AMT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	173.1	100	100	Glycine encephalopathy, 605899
AMTN	CRANIOFACIAL ANOMALIES MENDELIOME	125	98.8	95.4	?Amelogenesis imperfecta, type IIIB, 617607
ANG	ALS MENDELIOME	178	100	99.9	Amyotrophic lateral sclerosis 9, 611895
ANGPTL3	MENDELIOME PRECONCEPTION SCREENING	89.9	97.5	92.2	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	MENDELIOME	111.9	99	94.2	Plasma triglyceride level QTL, low, 615881
ANK1	MENDELIOME	136.5	100	99.3	Spherocytosis, type 1, 182900
ANK2	HEART PANEL MENDELIOME RITME	160.3	100	99.9	Cardiac arrhythmia, ankyrin-B-related, 600919 Long QT syndrome 4, 600919

ANK3	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	155.1	99.1	98.8	?Mental retardation, autosomal recessive, 37, 615493
ANKH	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	118.6	100	99.7	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	162.5	98	94.7	?Microcephaly 16, primary, autosomal recessive, 616681
ANKRD1	CARDIO CONGENITAL HEART DISEASE HEART PANEL	101.7	99.5	96.8	No OMIM phenotype Cardiomyopathy,hypertrophic (Arimura (2009) J Am Coll Cardiol 54,334) Cardiomyopathy,dilated (Duboscq-Bidot (2009) Eur Heart J 30,2128) ?Total anomalous pulmonary venous return (Cinquetti (2008) Hum Mutat 29,468) ?Neurodevelo
ANKRD11	CRANIOFACIAL ANOMALIES SKIN DISORDERS EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	96.3	97.4	94.1	KBG syndrome, 148050
ANKRD26	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME HEREDITARY CANCER	81	88.7	76.8	Thrombocytopenia 2, 188000
ANKS6	CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	91.8	92.8	88.6	Nephronophthisis 16, 615382
ANLN	RENAL DISORDERS MENDELIOME	146.2	97.2	93.3	Focal segmental glomerulosclerosis 8, 616032
ANO10	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	116.7	98.8	96.5	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	MOVEMENT DISORDERS MENDELIOME	138.8	99.7	98.2	Dystonia 24, 615034
ANO5	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	142.2	99.5	95.9	Gnathodiaphyseal dysplasia, 166260 Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307

	MUSCLE DISORDERS				
ANO6	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.4	98	92.9	Scott syndrome, 262890
ANOS1	SKIN DISORDERS HH RENAL DISORDERS MENDELIOME	90.3	89.4	87.6	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123	98.3	95.7	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	100	98.9	94.9	Hyaline fibromatosis syndrome, 228600
ANXA11	ALS MENDELIOME	80.8	98.9	88.8	Amytrophic lateral sclerosis 23, 617839
AP1S1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	111.3	99.9	99.5	MEDNIK syndrome, 609313
AP1S2	INTELLECTUAL DISABILITY MENDELIOME	65.7	78.6	70.9	Mental retardation, X-linked syndromic 5, 304340
AP1S3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES	114.2	90.3	90.1	{Psoriasis 15, pustular, susceptibility to}, 616106
AP2S1	RENAL DISORDERS MENDELIOME	115.7	90.4	89.7	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	95	97.8	90.2	Hermansky-Pudlak syndrome 2, 608233
AP3B2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135.1	97.5	94.2	Epileptic encephalopathy, early infantile, 48, 617276
AP3D1	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES	121	98.1	97.8	?Hermansky-Pudlak syndrome 10, 617050

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
AP4B1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147.4	100	99.8	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	98.7	99.7	97.9	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	127.2	99.1	96.4	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	65.8	71.8	69.3	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	MENDELIOME PRECONCEPTION SCREENING	96.8	99.7	96.6	Spastic paraplegia 48, autosomal recessive, 613647
APC	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	159	99.9	98.9	Adenoma, periampullary, somatic, 0 Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500 Desmoid disease, hereditary, 135290 Gardner syndrome, 175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
APC2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	63.5	93.3	85.3	?Sotos syndrome 3, 617169
APCDD1	SKIN DISORDERS MENDELIOME	179.6	100	99.3	Hypotrichosis 1, 605389
APOA1	MENDELIOME	99.3	100	100	Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 0 Corneal clouding, autosomal recessive, 0 Hypoalphalipoproteinemia, 604091
APOA2	MENDELIOME	106	88.5	82.2	Apolipoprotein A-II deficiency, 0 {Hypercholesterolemia, familial, modifier of}, 143890
APOA5	MENDELIOME	152.5	100	100	Hyperchylomicronemia, late-onset, 144650

					{Hypertriglyceridemia, susceptibility to}, 145750
APOB	MENDELIOME PRECONCEPTION SCREENING	181.7	99.6	99.3	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558
APOC2	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	99.4	100	100	Hyperlipoproteinemia, type Ib, 207750
APOC3	MENDELIOME	88.8	100	100	Apolipoprotein C-III deficiency, 614028
APOE	MENDELIOME PRECONCEPTION SCREENING	56.2	93.9	83.1	Alzheimer disease-2, 104310 Hyperlipoproteinemia, type III, 617347 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 {Coronary artery disease, severe, susceptibility to}, 617347
APOL1	PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS	192.2	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
APOPT1	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	63.8	81.4	78.1	Mitochondrial complex IV deficiency, 220110
APP	MENDELIOME	138.9	100	100	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714
APRT	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	68.2	100	98.7	Adenine phosphoribosyltransferase deficiency, 614723
APTX	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	118.9	94.2	91.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.4	99.5	95.6	Diabetes insipidus, nephrogenic, 125800
AQP5	SKIN DISORDERS MENDELIOME	110.9	99.8	97.4	Palmoplantar keratoderma, Bothnian type, 600231
AR	DSD MENDELIOME	85.3	93.8	88.3	Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300

					Hypospadias 1, X-linked, 300633 Spinal and bulbar muscular atrophy of Kennedy, 313200 {Prostate cancer, susceptibility to}, 176807
ARCN1	INTELLECTUAL DISABILITY MENDELIOME	167.9	96.6	96.6	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	MENDELIOME	179.9	100	100	Periventricular nodular heterotopia 8, 618185
ARFGEF2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	154.9	100	99.3	Periventricular heterotopia with microcephaly, 608097
ARG1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	167.8	100	100	Argininemia, 207800
ARHGAP26	MENDELIOME	153.2	100	99.9	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP29	CRANIOFACIAL ANOMALIES	136.4	98.9	95	No OMIM phenotype Cleft lip with or without cleft palate (Leslie (2015) Am J Hum Genet 96,397)
ARHGAP31	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	133.4	99.8	98.3	Adams-Oliver syndrome 1, 100300
ARHGDIA	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	143.9	100	99.9	Nephrotic syndrome, type 8, 615244
ARHGEF10	NEUROPATHIES MENDELIOME	132.8	99.8	98	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	VISION DISORDERS MENDELIOME	114	97.7	93	Retinitis pigmentosa 78, 617433
ARHGEF2	MENDELIOME	107.6	100	99.7	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF6	INTELLECTUAL DISABILITY	133.7	99.1	95.4	Mental retardation, X-linked 46, 300436
ARHGEF9	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	60.2	76.4	74.4	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	150	92.2	89.7	Coffin-Siris syndrome 2, 614607
ARID1B	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	156.7	94.3	89.5	Coffin-Siris syndrome 1, 135900
ARID2	INTELLECTUAL DISABILITY MENDELIOME	216.9	99.2	95.7	Coffin-Siris syndrome 6, 617808

ARL13B	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	97.3	98.9	92.8	Joubert syndrome 8, 612291
ARL2BP	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	66.3	88.3	79.3	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	VISION DISORDERS MENDELIOME	85	98.9	93.3	?Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	85.2	99.8	95.3	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARL6IP1	MENDELIOME	66	98.8	85.8	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC4	CILIO MENDELIOME PRECONCEPTION SCREENING	129.1	94.4	93.4	Ciliary dyskinesia, primary, 23, 615451
ARMC5	MENDELIOME HEREDITARY CANCER	136.5	99.8	97.5	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	CILIO INTELLECTUAL DISABILITY MENDELIOME	127.3	99.8	98.5	Joubert syndrome 30, 617622
ARNT2	MENDELIOME	153	99.6	99	?Webb-Dattani syndrome, 615926
ARPC1B	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	126.4	100	99.9	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ARR3	VISION DISORDERS MENDELIOME	98.1	100	99.9	Myopia 26, X-linked, female-limited, 301010
ARSA	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	97.8	100	99.7	Metachromatic leukodystrophy, 250100
ARSB	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME	117.5	94.9	87.7	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200

	PRECONCEPTION SCREENING				
ARSE	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	102.1	99.2	93	Chondrodysplasia punctata, X-linked recessive, 302950
ARSG	VISION DISORDERS MENDELIOME	141.1	100	99.4	Usher syndrome, type IV, 618144
ARV1	MENDELIOME PRECONCEPTION SCREENING	133.5	100	99.3	Epileptic encephalopathy, early infantile, 38, 617020
ARX	MOVEMENT DISORDERS DSD EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	29.1	75.8	59.5	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105.9	97.6	92.1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASB10	MENDELIOME	101.6	99.6	96.8	Glaucoma 1, open angle, F, 603383
ASCC1	MENDELIOME	139.1	96.7	93.2	?Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	MENDELIOME	213.1	90.2	79.5	Central hypoventilation syndrome, congenital, 209880 Haddad syndrome, 209880
ASH1L	INTELLECTUAL DISABILITY MENDELIOME	160.1	98.7	98.5	Mental retardation, autosomal dominant 52, 617796
ASIP	SKIN DISORDERS	102.3	100	99.9	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 [Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.4	99.9	98.8	Argininosuccinic aciduria, 207900
ASNS	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105.8	97.8	90.5	Asparagine synthetase deficiency, 615574
ASPA	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	127.6	99.1	95.8	Canavan disease, 271900

	PRECONCEPTION SCREENING				
ASPH	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.3	98.8	93.9	Traboulsi syndrome, 601552
ASPM	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101.2	98	92.2	Microcephaly 5, primary, autosomal recessive, 608716
ASPSCR1	MENDELIOME	88.4	98.4	93.7	Alveolar soft-part sarcoma, 606243
ASRGL1	VISION DISORDERS	131	100	99.6	No OMIM phenotype Retinal degeneration (Biswas (2016) Hum Mol Genet 25,2483)
ASS1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	97.9	95.7	87.5	Citrullinemia, 215700
ASXL1	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	159.8	99.1	97.7	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL2	INTELLECTUAL DISABILITY MENDELIOME	152	98.1	98	Shashi-Pena syndrome, 617190
ASXL3	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	162.9	99.8	98.9	Bainbridge-Ropers syndrome, 615485
ATAD1	MENDELIOME MITOCHONDRIAL DISORDERS	59.1	94.9	86.4	Hyperekplexia 4, 618011
ATAD3A	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	87.9	89	86.2	Harel-Yoon syndrome, 617183
ATAD3B	MITOCHONDRIAL DISORDERS	83.8	88.6	82.8	No OMIM phenotype Late-onset encephalopathy with cerebellar atrophy, ataxia and dystonia (Desai (2017) Brain 140,1595)
ATCAY	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	146.1	100	99.7	Ataxia, cerebellar, Cayman type, 601238
ATF3	DSD	138.1	99.8	98.7	No OMIM phenotype Eur J Endocrinol. 2008 May 158(5):729-39. doi
ATF6	VISION DISORDERS MENDELIOME	134.1	100	99.6	Achromatopsia 7, 616517

	PRECONCEPTION SCREENING				
ATG4B		153	100	100	
ATG5	MENDELIOME	130.5	98.6	90.5	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	119.5	99.7	99	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	161	99.7	97.9	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	NEUROPATHIES MENDELIOME	125	98.1	93.8	Neuropathy, hereditary sensory, type IF, 615632
ATM	MOVEMENT DISORDERS BRSTKNK PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	109.7	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATN1	MENDELIOME	120.9	99.7	97.7	Dentatorubro-pallidoluysian atrophy, 125370
ATOH7	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	102.8	95.8	89.6	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	MENDELIOME	71.9	95.9	86.9	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PARK PRECONCEPTION SCREENING	117.4	100	98.8	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A1	INTELLECTUAL DISABILITY MENDELIOME	142.9	100	99.9	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	190.8	100	99.6	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY MENDELIOME PARK	177.3	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2A1	MENDELIOME	155.9	100	100	Brody myopathy, 601003

	PRECONCEPTION SCREENING MUSCLE DISORDERS				
ATP2A2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	175.2	100	99.9	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2B2	HEARING IMPAIRMENT	186.5	100	100	{Deafness, autosomal recessive 12, modifier of}, 601386
ATP2B3	MOVEMENT DISORDERS MENDELIOME	135.2	99.5	97.7	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	SKIN DISORDERS MENDELIOME	118.6	99.9	99	Hailey-Hailey disease, 169600
ATP4A	IRON DISORDERS	148.5	100	99	No OMIM-phenotype Gastric neuroendocrine tumor, type 1 (Calvete (2015) Hum Mol Genet 24,2914)
ATP5A1	MENDELIOME MITOCHONDRIAL DISORDERS	85.3	94.8	85.8	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228
ATP5B	MITOCHONDRIAL DISORDERS	129.2	100	99.8	No OMIM phenotype
ATP5C1	MITOCHONDRIAL DISORDERS	90.1	95.4	84.9	No OMIM phenotype
ATP5D	MENDELIOME MITOCHONDRIAL DISORDERS	66.1	98.8	90.3	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	MENDELIOME MITOCHONDRIAL DISORDERS	135.5	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	MITOCHONDRIAL DISORDERS	80.9	96.9	84.5	No OMIM phenotype
ATP5G1	MITOCHONDRIAL DISORDERS	110	100	98.2	No OMIM phenotype
ATP5G2	MITOCHONDRIAL DISORDERS	93.8	100	98.4	No OMIM phenotype
ATP5G3	MITOCHONDRIAL DISORDERS	118.5	100	100	No OMIM phenotype
ATP5H	MITOCHONDRIAL DISORDERS	109.7	93	71.8	No OMIM phenotype
ATP5I	MITOCHONDRIAL DISORDERS	69.6	99.9	97	No OMIM phenotype
ATP5J	MITOCHONDRIAL DISORDERS	66.4	99	90.6	No OMIM phenotype
ATP5J2	MITOCHONDRIAL DISORDERS	109.7	100	99.9	No OMIM phenotype
ATP5L	MITOCHONDRIAL DISORDERS	144.6	100	99.9	No OMIM phenotype
ATP5L2	MITOCHONDRIAL DISORDERS	217.8	100	100	No OMIM phenotype
ATP5O	MITOCHONDRIAL DISORDERS	101.3	99.1	90.9	No OMIM phenotype
ATP5S	MITOCHONDRIAL DISORDERS	133.6	100	100	No OMIM phenotype Complex V deficiency
ATP6AP1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	112.8	99.1	94.7	Immunodeficiency 47, 300972
ATP6AP2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	46.1	81.2	55.6	?Parkinsonism with spasticity, X-linked, 300911 Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP6V0A2	SKIN DISORDERS METABOLIC DISORDERS	130	100	99.3	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
ATP6V0A4	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	116.4	99.9	98.6	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	144.8	98.2	94.9	Cutis laxa, autosomal recessive, type IID, 617403 Epileptic encephalopathy, infantile or early childhood, 3, 618012
ATP6V1B1	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	176.6	100	100	Renal tubular acidosis with deafness, 267300
ATP6V1B2	INTELLECTUAL DISABILITY MENDELIOME	137	99.9	98.2	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 Zimmermann-Laband syndrome 2, 616455
ATP6V1E1	METABOLIC DISORDERS MENDELIOME	67.2	92.2	85.6	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	 SKIN DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	133.2	99.7	97.8	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP7B	MOVEMENT DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	168.9	100	99.8	Wilson disease, 277900
ATP8A2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	133.5	100	99.5	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	139	96.7	94.4	Cholestasis, benign recurrent intrahepatic, 243300 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600
ATPAF1	MITOCHONDRIAL DISORDERS	79.8	74	68.1	No OMIM phenotype
ATPAF2	HEART PANEL MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	101.4	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273

ATPIF1	MITOCHONDRIAL DISORDERS	174.3	100	100	No OMIM phenotype
ATR	BONE MARROW FAILURE SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	138.3	99.4	96.9	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
ATRX	DSD EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	82.6	98.2	92.2	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	MENDELIOME	159.7	100	100	Spinocerebellar ataxia 1, 164400
ATXN10	MENDELIOME	137.7	99.8	97.9	Spinocerebellar ataxia 10, 603516
ATXN2	MENDELIOME	89.3	85.8	77.8	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600
ATXN3	MENDELIOME	98.3	91.1	86.2	Machado-Joseph disease, 109150
ATXN7	MENDELIOME	131	96.6	94.1	Spinocerebellar ataxia 7, 164500
ATXN8OS	MENDELIOME	123456	123456	123456	Spinocerebellar ataxia 8, 608768
AUH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	90.9	99.9	97.6	3-methylglutaconic aciduria, type I, 250950
AURKC	MENDELIOME PRECONCEPTION SCREENING	79	99.8	97.5	Spermatogenic failure 5, 243060
AUTS2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	110.3	96.9	95.5	Mental retardation, autosomal dominant 26, 615834
AVP	RENAL DISORDERS MENDELIOME	53.3	76.3	57.3	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	127.5	99.3	97.1	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	MENDELIOME	141.2	98.6	96.8	?Caudal duplication anomaly, 607864 Hepatocellular carcinoma, somatic, 114550
AXIN2	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME HEREDITARY CANCER	114.5	99.7	98.9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B2M	PRIMARY IMMUNODEFICIENCIES MENDELIOME	252.1	100	99.9	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600

	PRECONCEPTION SCREENING SCID				
B3GALNT1	METABOLIC DISORDERS	130.7	100	99.9	[Blood group, globoside system], 615021 [Blood group, P1PK system, P(k) phenotype], 111400
B3GALNT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	115	92.4	89.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	47.5	76.4	71.7	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.6	99.4	95.9	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101.2	97.4	93.4	Peters-plus syndrome, 261540
B4GALNT1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	151	95.6	90.1	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	105.4	99.9	99	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	104.3	96.1	95	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	120.4	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287

	MUSCLE DISORDERS				
B9D1	CILIO DSD RENAL DISORDERS MENDELIOME	115.3	92.1	91.4	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.9	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.4	98.3	95.3	Hypercholanemia, familial, 607748
BACH2	PRIMARY IMMUNODEFICIENCIES	154.8	100	99.9	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
BAG3	CARDIO HEART PANEL MENDELIOME MUSCLE DISORDERS	136.5	100	100	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	58.3	98	88.1	Nestor-Guillermo progeria syndrome, 614008
BAP1	BRSTKNK SKIN DISORDERS MENDELIOME HEREDITARY CANCER	111	85.1	82.7	Tumor predisposition syndrome, 614327
BARD1	BRSTKNK HEREDITARY CANCER	150.3	100	99.7	{Breast cancer, susceptibility to}, 114480
BAX	MENDELIOME	92.9	96.9	95.3	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BBIP1	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME	132	99.4	94.8	?Bardet-Biedl syndrome 18, 615995
BBS1	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.9	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	VISION DISORDERS	172.6	100	100	Bardet-Biedl syndrome 10, 615987

	CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
BBS12	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	208.6	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	181.8	100	99.8	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.9	99.7	97.3	Bardet-Biedl syndrome 4, 615982
BBS5	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	81.1	95.8	84.1	Bardet-Biedl syndrome 5, 615983
BBS7	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	120.7	98.1	91.7	Bardet-Biedl syndrome 7, 615984
BBS9	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	112.9	96	93.8	Bardet-Biedl syndrome 9, 615986

BCAP31	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	70.6	93.1	82.5	Deafness, dystonia, and cerebral hypomyelination, 300475
BCHE	MENDELIOME	162.9	100	99.8	Butyrylcholinesterase deficiency, 617936 {Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936
BCKDHA	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	171.5	100	99.5	Maple syrup urine disease, type Ia, 248600
BCKDHB	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	112.6	88.9	81.3	Maple syrup urine disease, type Ib, 248600
BCKDK	MENDELIOME PRECONCEPTION SCREENING	178.8	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	PRIMARY IMMUNODEFICIENCIES MENDELIOME	97.9	100	99.8	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic},, 273300 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCL11A	INTELLECTUAL DISABILITY MENDELIOME	139.3	98.2	97	Dias-Logan syndrome, 617101
BCL11B	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	79.7	96.6	88.6	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	MENDELIOME	156	99.9	98.4	Leukemia/lymphoma, B-cell, 2, 0
BCL7A	MENDELIOME	136	100	100	B-cell non-Hodgkin lymphoma, high-grade, 0
BCO1	METABOLIC DISORDERS MENDELIOME	164.9	100	100	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	109.7	99.3	96.8	Microphthalmia, syndromic 2, 300166
BCR	MENDELIOME	110.6	87.7	84.3	Leukemia, acute lymphocytic, somatic, 613065 Leukemia, chronic myeloid, somatic, 608232
BCS1L	SKIN DISORDERS HEARING IMPAIRMENT	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358

	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BDP1	HEARING IMPAIRMENT MENDELIOME	125.5	95.6	90	?Deafness, autosomal recessive 112, 618257
BEAN1	MENDELIOME	146.2	99.1	95.3	Spinocerebellar ataxia 31, 117210
BECN1		135.2	100	100	
BEST1	VISION DISORDERS MENDELIOME	144.6	99.6	97.5	Bestrophinopathy, autosomal recessive, 611809 Macular dystrophy, vitelliform, 2, 153700 Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 Retinitis pigmentosa, concentric, 613194 Retinitis pigmentosa-50, 613194 Vitreoretinochoroidopathy, 193220
BFSP1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	98	98.2	88.9	Cataract 33, multiple types, 611391
BFSP2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	89.5	99.8	97.6	Cataract 12, multiple types, 611597
BGN	HEART PANEL MENDELIOME	128.9	100	99.5	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	MENDELIOME PRECONCEPTION SCREENING	13.8	57.8	41.3	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BICC1	RENAL DISORDERS	152.3	100	99.6	{Renal dysplasia, cystic, susceptibility to}, 601331
BICD2	NEUROPATHIES MENDELIOME MUSCLE DISORDERS	158.6	100	99.9	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290
BIN1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	100.2	99.1	95.3	Centronuclear myopathy 2, 255200
BLK	PRIMARY IMMUNODEFICIENCIES MENDELIOME	115.7	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	116.3	99.4	96.5	Bloom syndrome, 210900

	HEREDITARY CANCER				
BLNK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	95.7	93.7	91.3	?Agammaglobulinemia 4, 613502
BLOC1S3	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	28.7	88.7	65.3	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	97.2	98.7	91.3	?Hermansky-pudlak syndrome 9, 614171
BLVRA	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.1	100	99.7	Hyperbiliverdinemia, 614156
BMP1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	143.9	99.9	99.1	Osteogenesis imperfecta, type XIII, 614856
BMP15	MENDELIOME	120.5	100	99.4	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP2	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME	173.4	100	99.9	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT MENDELIOME	151.7	100	99.9	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMP6	IRON DISORDERS	107.9	92.9	90	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008)).
BMPER	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	159.9	99.9	99	Diaphanospondylodysostosis, 608022
BMPR1A	 MENDELIOME HEREDITARY CANCER	98	99.7	94.2	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	172.4	100	98.9	Acromesomelic dysplasia, Demirhan type, 609441 Brachydactyly, type A1, D, 616849

	PRECONCEPTION SCREENING				Brachydactyly, type A2, 112600
BMPR2	MENDELIOME	194.7	99.9	99.7	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BMS1	SKIN DISORDERS MENDELIOME	96.6	66.7	65.6	?Aplasia cutis congenita, nonsyndromic, 107600
BNIP3		76.6	77.2	64.2	
BOLA1	MITOCHONDRIAL DISORDERS	110.6	100	99.9	No OMIM phenotype
BOLA2	MITOCHONDRIAL DISORDERS	120.5	100	100	No OMIM phenotype ?Autism and developmental delay (Nuttle (2016) Nature 536, 205)
BOLA3	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	50.1	92.3	81.7	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.8	100	100	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800
BPTF	INTELLECTUAL DISABILITY MENDELIOME	177.7	96.1	95.1	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	74.4	87.6	77.2	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRAT1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	108.5	99.8	97.4	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	BONE MARROW FAILURE BRSTKNK MENDELIOME HEREDITARY CANCER	177.5	98.9	96.9	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	BONE MARROW FAILURE BRSTKNK MENDELIOME HEREDITARY CANCER	102.7	99	97.4	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070 {Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255

					{Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRDT	MENDELIOME	100.3	92.6	86.5	?Spermatogenic failure 21, 617644
BRF1	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	100	96.6	92.9	Cerebellofaciodental syndrome, 616202
BRIP1	BONE MARROW FAILURE BRSTKNK SKIN DISORDERS MENDELIOME HEREDITARY CANCER	117.8	99.8	97.7	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BRPF1	INTELLECTUAL DISABILITY MENDELIOME	179.1	100	99.5	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRWD3	INTELLECTUAL DISABILITY MENDELIOME	97.1	97	92.3	Mental retardation, X-linked 93, 300659
BSCL2	MOVEMENT DISORDERS SKIN DISORDERS HEART PANEL NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	113.5	100	100	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BSND	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.1	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
BTD	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	166.6	100	99.9	Biotinidase deficiency, 253260
BTK	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	116.2	100	99.6	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BUB1	MENDELIOME HEREDITARY CANCER	136.4	99.8	97.9	Colorectal cancer with chromosomal instability, somatic, 0
BUB1B	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	136.5	98.6	97.9	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430

	HEREDITARY CANCER				
BUB3	HEREDITARY CANCER	146.1	98.6	97.8	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
BVES	HEART PANEL MENDELIOME	115	99.8	98.2	?Cardiac arrhythmia with increased serum creatine kinase, 616812
C11orf70	CILIO MENDELIOME	60.7	92.3	82.2	Ciliary dyskinesia, primary, 38, 618063
C12orf4	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	131.3	99.1	94.7	Mental retardation, autosomal recessive 66, 618221
C12orf57	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	152	100	100	Temtamy syndrome, 218340
C12orf65	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	88.2	97.3	91.9	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C15orf41	IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	124.9	99.9	97.9	Dyserythropoietic anemia, congenital, type Ib, 615631
C19orf12	MOVEMENT DISORDERS VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PARK PRECONCEPTION SCREENING	93.9	100	99.7	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C19orf70	MITOCHONDRIAL DISORDERS	63	100	98.8	No OMIM phenotype Mitochondrial encephalopathy with liver disease, early-onset fatal (Guarani (2016) Elife 5, e17163) Mitochondrial hepato-encephalopathy (Zeharia (2016) Eur J Hum Genet 24,1778)
C1GALT1C1	METABOLIC DISORDERS MENDELIOME	147.2	99.8	98.7	Tn polyagglutination syndrome, somatic, 300622
C1QA	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	120.4	100	99	C1q deficiency, 613652
C1QB	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	183.4	100	99.9	C1q deficiency, 613652

	PRECONCEPTION SCREENING				
C1QBP	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	80.7	81.7	71	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	198.1	100	98.9	C1q deficiency, 613652
C1QTNF5	VISION DISORDERS MENDELIOME	151.2	79.7	65.5	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	PRIMARY IMMUNODEFICIENCIES MENDELIOME	156.9	100	100	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	117.6	100	99.7	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	129.9	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C21orf2	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	104.4	99.9	98.7	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C21orf59	CILIO MENDELIOME PRECONCEPTION SCREENING	145.8	98.7	94.6	Ciliary dyskinesia, primary, 26, 615500
C2CD3	CILIO SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.1	95.8	95.6	?Orofaciodigital syndrome XIV, 615948
C2orf71	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	124.6	99.7	98.8	Retinitis pigmentosa 54, 613428
C3	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	145.5	100	99.7	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	91.5	98.1	95.9	C4a deficiency, 614380 [Blood group, Rodgers], 614374

C4B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	90.5	98.5	96.5	C4B deficiency, 614379
C4orf26	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	197.6	100	100	Amelogenesis imperfecta, type IIA4, 614832
C5	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	134.4	98.4	95.3	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C5orf42	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	122.8	98.6	95.5	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C6	PRIMARY IMMUNODEFICIENCIES MENDELIOME	157.5	100	99.9	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	PRIMARY IMMUNODEFICIENCIES MENDELIOME	132.3	99.1	94.8	C7 deficiency, 610102
C8A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	120.2	100	99.8	C8 deficiency, type I, 613790
C8B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	135.8	99.9	99.5	C8 deficiency, type II, 613789
C8G	PRIMARY IMMUNODEFICIENCIES	130.1	100	100	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
C8orf37	VISION DISORDERS CILIO MENDELIOME PRECONCEPTION SCREENING	126.4	100	99	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
C9	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	133.7	100	98.5	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
C9orf72	MENDELIOME	108.5	99	94.2	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	MENDELIOME PRECONCEPTION SCREENING	109.6	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
CA4	VISION DISORDERS MENDELIOME	142.4	100	99.9	Retinitis pigmentosa 17, 600852
CA5A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	124.1	99.5	94.9	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.6	96.8	93	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	67.1	74.6	65.5	Deafness, autosomal recessive 93, 614899
CABP4	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	98.4	99.7	97.7	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	87.8	92.7	89.1	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1B	MENDELIOME	137.5	94.1	91.5	?Dystonia 23, 614860
CACNA1C	HEART PANEL INTELLECTUAL DISABILITY MENDELIOME RITME	154.6	99.9	99.2	Brugada syndrome 3, 611875 Timothy syndrome, 601005
CACNA1D	HEARING IMPAIRMENT HEART PANEL MENDELIOME RITME	149.9	98	97.8	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	MOVEMENT DISORDERS EPILEPSY	139.3	99.8	99.3	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1F	VISION DISORDERS MENDELIOME	99.4	99.8	97.9	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071
CACNA1G	MOVEMENT DISORDERS	132.8	99	97.5	Spinocerebellar ataxia 42, 616795

	INTELLECTUAL DISABILITY MENDELIOME				Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	RENAL DISORDERS MENDELIOME	103.4	97.6	94.3	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1S	MENDELIOME MUSCLE DISORDERS	135.5	100	99.7	Hypokalemic periodic paralysis, type 1, 170400 {Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580
CACNA2D1	HEART PANEL INTELLECTUAL DISABILITY RITME	82.7	93.1	84.4	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077) Histiocytoid cardiomyopathy (Cataldo (2014) Cardiol Young epub) West syndrome (Hino-Fukuyo (2015) Hum Genet 134,
CACNA2D2	EPILEPSY	135.2	93.7	92.6	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)
CACNA2D4	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	112	99.2	97.7	Retinal cone dystrophy 4, 610478
CACNB2	HEART PANEL MENDELIOME RITME	150.9	99.5	96.9	Brugada syndrome 4, 611876
CACNB4	MOVEMENT DISORDERS EPILEPSY MENDELIOME	106.1	96.3	94.6	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CACNG2	MENDELIOME	114.9	100	99.9	?Mental retardation, autosomal dominant 10, 614256
CAD	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	158.9	100	99.7	Epileptic encephalopathy, early infantile, 50, 616457
CALM1	HEART PANEL MENDELIOME RITME	114.3	100	99.7	Long QT syndrome 14, 616247 Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916
CALM2	HEART PANEL MENDELIOME RITME	54	67.8	65.8	Long QT syndrome 15, 616249
CALM3	HEART PANEL RITME	117.4	99.9	99.5	No OMIM phenotype Catecholaminergic polymorphic ventricular tachycardia (Boczek (2013) Circulation 128,A14699) Long QT syndrome (Reed (2015) Heart Rhythm 12,419)

					{Cardiomyopathy,hypertrophic,modifier of} (Friedrich (2009) Eur Heart J 30,1648)
CALR	HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME	113.5	99.9	97.4	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	INTELLECTUAL DISABILITY MENDELIOME	123.2	100	99.9	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CAMK2B	INTELLECTUAL DISABILITY MENDELIOME	103.4	98.7	93.7	Mental retardation, autosomal dominant 54, 617799
CAMTA1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	185.9	99.6	98.8	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	142.1	100	99.8	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	146	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	PRECONCEPTION SCREENING	95.9	99.9	98	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN12	SKIN DISORDERS	86.9	95.3	86.8	No OMIM phenotype Modifying factor in ichthyosis
CAPN3	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	111.4	99	96.7	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600
CAPN5	VISION DISORDERS MENDELIOME	166.1	100	99.9	Vitreoretinopathy, neovascular inflammatory, 193235
CARD11	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	154.6	99.9	98.6	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD14	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	116.2	99.7	97.7	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	119.7	98.3	96.4	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	121.1	94.9	92.7	Immunodeficiency 58, 618131
CARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	121.1	100	99.8	Combined oxidative phosphorylation deficiency 27, 616672

	PRECONCEPTION SCREENING				
CASK	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	92.3	98.7	93.7	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CASP10	PRIMARY IMMUNODEFICIENCIES MENDELIOME	117.4	99.5	98	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP14	SKIN DISORDERS MENDELIOME	85.5	100	100	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	144.8	95.6	95.5	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CASQ1	MENDELIOME MUSCLE DISORDERS	122	100	99.6	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	HEART PANEL MENDELIOME PRECONCEPTION SCREENING RITME	143.3	99.9	99.2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	178	100	99.7	Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.2	96.8	92.8	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.5	100	100	Acatalasemia, 614097
CATSPER1	MENDELIOME PRECONCEPTION SCREENING	147.2	100	99.4	Spermatogenic failure 7, 612997
CAV1	SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	265.4	100	100	?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 Pulmonary hypertension, primary, 3, 615343
CAV3	CARDIO HEART PANEL MENDELIOME RITME MUSCLE DISORDERS	304.7	100	100	Cardiomyopathy, familial hypertrophic, 192600 Creatine phosphokinase, elevated serum, 123320 Long QT syndrome 9, 611818 Myopathy, distal, Tateyama type, 614321 Rippling muscle disease, 606072

CAVIN1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	137	99.9	99.3	Lipodystrophy, congenital generalized, type 4, 613327
CAVIN4	HEART PANEL	159.5	100	100	No OMIM phenotype
CBL	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	129.8	96.9	95.7	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	116.2	97.1	91.1	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	DSD MENDELIOME PRECONCEPTION SCREENING	96.6	99.9	98.4	?46XY sex reversal 5, 613080
CC2D1A	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	119.4	99.8	98.3	Mental retardation, autosomal recessive 3, 608443
CC2D2A	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.4	99.5	97.1	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCBE1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	75.9	98.9	95.5	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	CILIO MENDELIOME PRECONCEPTION SCREENING	116.8	100	99	Ciliary dyskinesia, primary, 17, 614679
CCDC114	CILIO MENDELIOME PRECONCEPTION SCREENING	120.7	100	99.6	Ciliary dyskinesia, primary, 20, 615067

CCDC115	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	59.3	88	85.3	Congenital disorder of glycosylation, type IIo, 616828
CCDC141	HH	111.9	99.9	98	No OMIM phenotype PMID:27014940,PMID: 28324054
CCDC151	CILIO MENDELIOME PRECONCEPTION SCREENING	116.2	100	99.7	Ciliary dyskinesia, primary, 30, 616037
CCDC174	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	133.1	98.1	93.3	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC22	INTELLECTUAL DISABILITY MENDELIOME	93.3	97.3	89.4	Ritscher-Schinzel syndrome 2, 300963
CCDC28B	CILIO	83.7	100	98.6	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	CILIO MENDELIOME PRECONCEPTION SCREENING	74.3	96.6	88.9	Ciliary dyskinesia, primary, 14, 613807
CCDC40	CILIO MENDELIOME PRECONCEPTION SCREENING	126.5	98.9	97.8	Ciliary dyskinesia, primary, 15, 613808
CCDC50	HEARING IMPAIRMENT MENDELIOME	135.1	99.9	98.9	?Deafness, autosomal dominant 44, 607453
CCDC65	CILIO MENDELIOME PRECONCEPTION SCREENING	105.9	99.7	97.6	Ciliary dyskinesia, primary, 27, 615504
CCDC78	MENDELIOME MUSCLE DISORDERS	114.9	100	100	?Centronuclear myopathy 4, 614807
CCDC8	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	111.9	100	100	3-M syndrome 3, 614205
CCDC88A	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	78.9	94.7	84.9	?PEHO syndrome-like, 617507
CCDC88C	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101.4	99.8	97.4	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCL2	IRON DISORDERS	138.4	100	100	{Coronary artery disease, modifier of}, 0 {HIV-1, resistance to}, 609423 {Mycobacterium tuberculosis, susceptibility to}, 607948 {Spina bifida, susceptibility to}, 182940

CCM2	MENDELIOME	160	97.9	97.9	Cerebral cavernous malformations-2, 603284
CCND2	INTELLECTUAL DISABILITY MENDELIOME	152.3	100	100	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	INTELLECTUAL DISABILITY MENDELIOME	82.4	88	76.6	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	CILIO MENDELIOME PRECONCEPTION SCREENING	103.1	99	95.6	Ciliary dyskinesia, primary, 29, 615872
CCR5		196.5	100	100	{Diabetes mellitus, insulin-dependent, 22}, 612522 {Hepatitis C virus, resistance to}, 609532 {HIV infection, susceptibility/resistance to}, 0 {West nile virus, susceptibility to}, 610379
CCT2	VISION DISORDERS	150.4	99.9	98.7	No OMIM phenotype Leber congenital amaurosis
CCT5	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	164.5	99.9	99.1	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	132.3	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD164	HEARING IMPAIRMENT MENDELIOME	130.4	98	93.9	?Deafness, autosomal dominant 66, 616969
CD19	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	88.8	99.9	98.4	Immunodeficiency, common variable, 3, 613493
CD247	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	101.6	100	98.9	?Immunodeficiency 25, 610163
CD27	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	118.1	100	99.6	Lymphoproliferative syndrome 2, 615122
CD2AP	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	98.2	99.6	96	Glomerulosclerosis, focal segmental, 3, 607832
CD320	MENDELIOME PRECONCEPTION SCREENING	92	100	99.5	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.3	99.2	95.7	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia], 0 {Coronary heart disease, susceptibility to, 7}, 610938

					{Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD3D	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	193.8	100	100	Immunodeficiency 19, 615617
CD3E	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	152.1	100	99.9	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	156.8	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	MENDELIOME	119.9	100	99.7	OKT4 epitope deficiency, 613949
CD40	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	165.4	100	99.9	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	PRIMARY IMMUNODEFICIENCIES MENDELIOME	126.6	95.9	86.8	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS	115.1	97.8	93.2	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	PRIMARY IMMUNODEFICIENCIES MENDELIOME	138.9	94.4	86.2	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 [Blood group Cromer], 613793
CD59	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	200.9	93.6	86.5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	PRIMARY IMMUNODEFICIENCIES	116.4	100	99.3	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
CD79A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	128.3	99.8	97	Agammaglobulinemia 3, 613501
CD79B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	210.7	100	100	Agammaglobulinemia 6, 612692
CD81	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	142.6	99.9	98.1	Immunodeficiency, common variable, 6, 613496
CD8A	PRIMARY IMMUNODEFICIENCIES	110	99.9	99	CD8 deficiency, familial, 608957

	MENDELIOME PRECONCEPTION SCREENING SCID				
CD96	MENDELIOME	156.8	100	99.9	C syndrome, 211750
CDAN1	SKIN DISORDERS IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	97.6	97.6	95.2	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	161.4	98.3	93.5	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC42	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	97	96.7	89.5	Takenouchi-Kos syndrome, 616737
CDC45	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	160.7	99.4	97.5	Meier-Gorlin syndrome 7, 617063
CDC6	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	165.4	99.8	98.3	?Meier-Gorlin syndrome 5, 613805
CDC73	MENDELIOME HEREDITARY CANCER	102.7	99.8	97.7	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDCA7	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	109.2	100	99.3	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	BRSTKNK MENDELIOME HEREDITARY CANCER	125.3	99.1	98.4	Blepharocheilodontic syndrome 1, 119580 Endometrial carcinoma, somatic, 608089 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDH11	INTELLECTUAL DISABILITY MENDELIOME	154.2	100	100	Elsahy-Waters syndrome, 211380
CDH15	INTELLECTUAL DISABILITY MENDELIOME	120.6	99.8	97.2	Mental retardation, autosomal dominant 3, 612580
CDH2	HEART PANEL	137.5	98.4	97.5	No OMIM phenotype
CDH23	VISION DISORDERS HEARING IMPAIRMENT	197.2	100	100	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D, 601067

	MENDELIOME PRECONCEPTION SCREENING				Usher syndrome, type 1D/F digenic, 601067 {Pituitary adenoma 5, multiple types}, 617540
CDH3	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	159.3	99.5	97.3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	154.2	99.2	98	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.6	100	99.9	Al Kaissi syndrome, 617694
CDK13	INTELLECTUAL DISABILITY MENDELIOME	136.6	95.4	88.1	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK4	SKIN DISORDERS HEREDITARY CANCER	128	100	99.6	{Melanoma, cutaneous malignant, 3}, 609048
CDK5	MENDELIOME	110.4	99.9	99.1	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123.9	99.9	98.7	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	MENDELIOME	105.9	99.4	95.7	?Microcephaly 12, primary, autosomal recessive, 616080
CDKL5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	114.4	94.9	91.8	Epileptic encephalopathy, early infantile, 2, 300672
CDKN1A	HEREDITARY CANCER	145.9	100	100	No OMIM phenotype Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) {Cancer, association with} (Mousses (1995) Hum Mol Genet 4, 1089) {Breast cancer, association with} (Staalesen (2006) Clin Cancer Res 12, 6000)
CDKN1B	MENDELIOME HEREDITARY CANCER	93.2	100	99.5	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	21.1	68.1	51.8	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	79.6	92.2	91.4	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, 0 Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDKN2B	PRIMARY IMMUNODEFICIENCIES	85.9	100	99.8	No OMIM phenotype Renal cell carcinoma (Jafri (2015) Cancer Discov 5, 723)

	HEREDITARY CANCER				Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) ?Melanoma (Foley (2015) EBioMedicine 2,74) ?Parathyroid adenoma (Costa-Guda (2013) Horm
CDKN2B-AS1					
CDKN2C	HEREDITARY CANCER	153.6	100	100	No OMIM phenotype MEN-1-like
CDON	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	143.8	100	99.6	Holoprosencephaly 11, 614226
CDSN	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	119.3	100	99.5	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	99.3	96.8	93.8	Meier-Gorlin syndrome 4, 613804
CEACAM16	HEARING IMPAIRMENT MENDELIOME	144.9	100	99.7	Deafness, autosomal dominant 4B, 614614
CEBPA	MENDELIOME HEREDITARY CANCER	46.7	75.5	65.1	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626
CEBPE	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	71.1	99.3	95.8	Specific granule deficiency, 245480
CEL	METABOLIC DISORDERS MENDELIOME	130	85	82.7	Maturity-onset diabetes of the young, type VIII, 609812
CELSR1	SKIN DISORDERS	183.8	93.8	92.9	No OMIM phenotype Congenital heart defects (Qiao (2016) Clin Sci (Lond)) Craniorachischisis (Robinson (2012) Hum Mutat 33,440) Neural tube defects (Qiao (2016) Clin Sci (Lond)) Spina bifida (Lei (2014) PLoS One 9,e92207) Lymphoedema (Gonzal
CENPE	MENDELIOME	61.5	95	83.3	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	CILIO INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.5	99.5	97.6	Stromme syndrome, 243605
CENPJ	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	141.7	99.7	97.8	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CEP104	CILIO	119.9	99	97.9	Joubert syndrome 25, 616781

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
CEP120	CILIO SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	79.2	98.1	89.1	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	162.5	97.2	94.5	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	94.2	99.9	98	Nephronophthisis 15, 614845
CEP19	MENDELIOME PRECONCEPTION SCREENING	202.7	100	100	Morbid obesity and spermatogenic failure, 615703
CEP250	VISION DISORDERS	105	99.9	98.8	No OMIM phenotype Usher syndrome,atypical (Khateb (2014) J Med Genet 51,460) ?Miscarriage, recurrent (Filges (2014) Mol Hum Reprod epub,epub)
CEP290	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	66.1	88.4	76.7	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	VISION DISORDERS CILIO DSD INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	83.5	97.7	89.6	Joubert syndrome 15, 614464
CEP55	CILIO RENAL DISORDERS MENDELIOME	129.5	100	99.9	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500

CEP57	MENDELIOME PRECONCEPTION SCREENING	92.6	99.4	93.3	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	MENDELIOME	119.4	96.7	89.9	?Seckel syndrome 6, 614728
CEP78	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME	112.1	97	93.9	Cone-rod dystrophy and hearing loss, 617236
CEP83	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	96.7	98.3	89.1	Nephronophthisis 18, 615862
CEP89	INTELLECTUAL DISABILITY MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	125.5	94.7	91.4	No OMIM phenotype Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138) ?Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679)
CERKL	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.4	98.6	92.8	Retinitis pigmentosa 26, 608380
CERS1	MENDELIOME	55.7	70.5	60.5	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	106.8	100	98.8	Ichthyosis, congenital, autosomal recessive 9, 615023
CES1	MENDELIOME	155.1	99.8	98.7	Drug metabolism, altered, CES1-related, 618057
CETP	MENDELIOME	131.9	100	100	Hyperalphalipoproteinemia, 143470 [High density lipoprotein cholesterol level QTL 10], 143470
CRANIOFACIAL ANOMALIESP43	MENDELIOME	123.7	99.4	96.1	Spermatogenic failure 19, 617592
CRANIOFACIAL ANOMALIESP44	MENDELIOME	104	99.4	97.1	?Spermatogenic failure 20, 617593
CRANIOFACIAL ANOMALIESP53	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME PRECONCEPTION SCREENING	146.6	97.6	94.2	Heterotaxy, visceral, 6, autosomal recessive, 614779
CRANIOFACIAL	MENDELIOME	58.4	96.2	86.6	Spermatogenic failure 24, 617959

ANOMALIE SP69					
CFB	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	147.1	100	100	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFC1	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	74.5	82.7	71.3	Heterotaxy, visceral, 2, autosomal, 605376
CFD	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	80.6	89.7	81.6	Complement factor D deficiency, 613912
CFH	VISION DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	183.2	98.7	95.3	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS	236.5	95.8	94.2	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR2	PRIMARY IMMUNODEFICIENCIES	171.7	96.8	89	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
CFHR3	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS	101	90.6	85.6	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR4	PRIMARY IMMUNODEFICIENCIES	139.7	99.6	97.3	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
CFHR5	PRIMARY IMMUNODEFICIENCIES MENDELIOME	97.3	98.7	93.4	Nephropathy due to CFHR5 deficiency, 614809
CFI	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	145.5	96.6	92.8	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	117.3	94.3	86.7	Nemaline myopathy 7, autosomal recessive, 610687
CFP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	95.9	98.4	93.4	Properdin deficiency, X-linked, 312060
CFTR	PRIMARY IMMUNODEFICIENCIES	124	99.1	96.3	Congenital bilateral absence of vas deferens, 277180

	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800
CHAMP1	INTELLECTUAL DISABILITY MENDELIOME	160.6	100	100	Mental retardation, autosomal dominant 40, 616579
CHAT	 MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	130.3	89.3	86.8	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCONGENITAL HEART DISEASE10	ALS MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	20	43	35.2	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHCONGENITAL HEART DISEASE2	MENDELIOME MITOCHONDRIAL DISORDERS PARK	95.4	99.5	91.9	Parkinson disease 22, autosomal dominant, 616710
CONGENITAL HEART DISEASE1	INTELLECTUAL DISABILITY MENDELIOME	95.4	93.7	83.5	Pilarowski-Bjornsson syndrome, 617682
CONGENITAL HEART DISEASE2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	137.7	99.3	98.5	Epileptic encephalopathy, childhood-onset, 615369
CONGENITAL HEART DISEASE3	INTELLECTUAL DISABILITY MENDELIOME	106.1	94.7	92.1	Snijders Blok-Campeau syndrome, 618205
CONGENITAL HEART DISEASE4	INTELLECTUAL DISABILITY MENDELIOME	131.3	100	99.8	Sifrim-Hitz-Weiss syndrome, 617159
CONGENITAL HEART DISEASE7	VISION DISORDERS CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEART PANEL HH PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CONGENITAL HEART	INTELLECTUAL DISABILITY	148.1	100	99.9	{Autism, susceptibility to, 18}, 615032

DISEASE8					
CHEK2	BRSTKNK MENDELIOME HEREDITARY CANCER	100.8	82.4	78.7	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to}, 0 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CHIT1	METABOLIC DISORDERS	117.5	99.6	97.3	[Chitotriosidase deficiency], 614122
CHKB	SKIN DISORDERS HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CHM	VISION DISORDERS MENDELIOME	102.6	96.5	87.3	Choroideremia, 303100
CHMP1A	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	133.7	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	ALS MENDELIOME PARK	92	98.6	91.7	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CHMP4B	VISION DISORDERS MENDELIOME	139.3	99.9	99	Cataract 31, multiple types, 605387
CHN1	MENDELIOME	141.8	99.2	97.7	Duane retraction syndrome 2, 604356
CHRDL1	VISION DISORDERS MENDELIOME	109.7	100	99.4	Megalocornea 1, X-linked, 309300
CHRM2	HEART PANEL	142.7	100	99.8	No OMIM phenotype
CHRM3	MENDELIOME	160.2	100	100	?Prune belly syndrome, 100100
CHRNA1	 MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	121.8	94.7	94.6	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNA2	EPILEPSY MENDELIOME	229.3	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	142.1	96.7	95.8	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB1	MENDELIOME	131.8	98.8	96.7	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor

	PRECONCEPTION SCREENING MUSCLE DISORDERS				deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	EPILEPSY MENDELIOME	247.5	98.4	94.6	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	150.5	100	99	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	127.7	99.3	95.8	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNG	MENDELIOME PRECONCEPTION SCREENING	155.2	100	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	MENDELIOME	240.7	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	165.6	95.7	93.3	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	91.6	100	97.5	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	334	100	100	Macular corneal dystrophy, 217800
CHST8	MENDELIOME	263.6	100	100	?Peeling skin syndrome 3, 616265
CHSY1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.4	95.9	93.9	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.6	100	98.4	Cocoon syndrome, 613630
CIB1	SKIN DISORDERS	130.9	95.2	92.5	No OMIM phenotype Epidermodysplasia verruciformis

CIB2	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	229.9	99.9	99.6	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIC	INTELLECTUAL DISABILITY MENDELIOME	54.2	63.7	60.9	Mental retardation, autosomal dominant 45, 617600
CIDEC	MENDELIOME	96.4	99.9	96.4	?Lipodystrophy, familial partial, type 5, 615238
CIITA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	125	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CISD2	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	127.9	83.4	83.4	Wolfram syndrome 2, 604928
CIT	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	108.6	99.9	98.2	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	111.6	99.2	99	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	161.3	98.9	96.6	Filippi syndrome, 272440
CLCF1	MENDELIOME PRECONCEPTION SCREENING	76.2	98.7	97.7	Cold-induced sweating syndrome 2, 610313
CLCN1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	137.4	100	99.5	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	108.6	100	99.4	Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN4	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	123	100	99.8	Raynaud-Claes syndrome, 300114
CLCN5	SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS	134.6	99.6	98	Dent disease, 300009 Hypophosphatemic rickets, 300554

	MENDELIOME				Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCN7	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	129.7	99.5	98.2	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLCNKA	MENDELIOME	108.5	99.4	94.7	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.7	98.5	90.5	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.6	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	SKIN DISORDERS RENAL DISORDERS MENDELIOME	148.9	100	100	HELIX syndrome, 617671
CLDN14	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	130.6	100	99.9	Deafness, autosomal recessive 29, 614035
CLDN16	EPILEPSY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	136.3	100	99.9	Hypomagnesemia 3, renal, 248250
CLDN19	EPILEPSY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.7	98.2	93.7	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLEC4D	PRIMARY IMMUNODEFICIENCIES	140.4	100	100	No OMIM phenotype plays a role in immunity (Zhu et al.,Immunity 2013).
CLEC7A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	150.7	100	99.9	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLIC2	INTELLECTUAL DISABILITY MENDELIOME	72.5	99.3	95.2	?Mental retardation, X-linked, syndromic 32, 300886
CLIC5	HEARING IMPAIRMENT MENDELIOME	118.3	100	99.9	?Deafness, autosomal recessive 103, 616042
CLIP1	INTELLECTUAL DISABILITY PRECONCEPTION SCREENING	131.7	99.7	97.8	No OMIM phenotype Intellectual disability, autosomal recessive (Larti (2015) Eur J Hum Genet 23,331)
CLMP	MENDELIOME PRECONCEPTION SCREENING	111	100	99.9	Congenital short bowel syndrome, 615237
CLN3	VISION DISORDERS EPILEPSY	114.9	92.5	90.7	Ceroid lipofuscinosis, neuronal, 3, 204200

	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
CLN5	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	146.1	98.2	92.2	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	131.6	98.9	95.3	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	163.9	83.5	83.5	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	182.4	100	99.8	Pontocerebellar hypoplasia, type 10, 615803
CLPB	MOVEMENT DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	115.4	99.8	96.9	Perrault syndrome 3, 614129
CLPX	MENDELIOME	151	99.7	97	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	157.2	100	99.8	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902

CLTC	INTELLECTUAL DISABILITY MENDELIOME	171.5	99.8	99.2	Mental retardation, autosomal dominant 56, 617854
CLUAP1	VISION DISORDERS	152.6	99.9	99.5	No OMIM phenotype Leber congenital amaurosis (Soens (2016) Genet Med 18,1044)
CNBP	MENDELIOME	136.7	100	100	Myotonic dystrophy 2, 602668
CNGA1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.2	89.4	84.6	Retinitis pigmentosa 49, 613756
CNGA3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	167.7	100	99.9	Achromatopsia 2, 216900
CNGB1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	102.5	98.4	94.8	Retinitis pigmentosa 45, 613767
CNGB3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.4	97.7	93	Achromatopsia 3, 262300 Macular degeneration, juvenile, 248200
CNKSR2	INTELLECTUAL DISABILITY MENDELIOME	98.1	96.5	89.7	Mental retardation, X-linked, syndromic, Houge type, 301008
CNNM2	EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	188.4	100	99.2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	190.5	98.7	97.8	Jalili syndrome, 217080
CNPY3	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	87.1	99.8	96.7	Epileptic encephalopathy, early infantile, 60, 617929
CNTN1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	151.7	99.8	98.3	?Myopathy, congenital, Compton-North, 612540
CNTN2	EPILEPSY MENDELIOME	125.4	92.7	92.6	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTN3		151.3	100	99.7	
CNTNAP1	NEUROPATHIES MENDELIOME	161.2	99.2	97.5	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286

	PRECONCEPTION SCREENING				
CNTNAP2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	148	100	99.9	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COA1	MITOCHONDRIAL DISORDERS	89.4	100	100	No OMIM phenotype
COA3	MITOCHONDRIAL DISORDERS	143.3	100	100	No OMIM phenotype Neuropathy,exercise intolerance,obesity and short stature (Ostergaard (2015) J Med Genet 52,203
COA5	MENDELIOME MITOCHONDRIAL DISORDERS	59.2	85.6	84	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	78.7	98.8	91.9	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	MENDELIOME MITOCHONDRIAL DISORDERS	146	100	100	?Mitochondrial complex IV deficiency, 220110
COASY	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	168.5	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COCH	HEARING IMPAIRMENT MENDELIOME	194.4	99.9	99.6	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.2	100	99.9	Congenital disorder of glycosylation, type IIg, 611209
COG2	METABOLIC DISORDERS MENDELIOME	122.6	97.2	94.7	?Congenital disorder of glycosylation, type IIq, 617395
COG4	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123.8	100	99.9	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	107	97.4	93.8	Congenital disorder of glycosylation, type IIi, 613612
COG6	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	78.4	95	85.9	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328

	PRECONCEPTION SCREENING				
COG7	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.1	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	122.4	99.9	98.4	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	88.4	98.6	93.9	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	90.8	94.9	89.6	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	92.2	99.9	98.3	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL12A1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	137.5	99.5	97.5	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL13A1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	85.4	99.8	97.1	Myasthenic syndrome, congenital, 19, 616720
COL14A1	SKIN DISORDERS	131	98	96.6	No OMIM phenotype Keratoderma, palmoplantar, punctate (Guo (2012) J Med Genet 49,563)
COL17A1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	107.9	99.2	96.6	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL18A1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	88.7	93.9	87.7	Knobloch syndrome, type 1, 267750
COL1A1	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	134.9	98.1	96.3	Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210

					Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL1A2	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	101.8	96.7	93.6	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL25A1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	126.8	98.4	95.8	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	MENDELIOME	116.1	99.5	97.4	Steel syndrome, 615155
COL2A1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	103.4	99.9	99	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL3A1	SKIN DISORDERS HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	104.3	97.8	92.3	Ehlers-Danlos syndrome, vascular type, 130050
COL4A1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	92.8	97.9	94	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, s, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	INTELLECTUAL DISABILITY	96.8	98.5	93.9	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519

	MENDELIOME				
COL4A3	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	89.6	97.8	95.5	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A3BP	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	121.6	98.3	92.7	Mental retardation, autosomal dominant 34, 616351
COL4A4	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	85	97.6	93.5	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign, 0
COL4A5	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	52.3	92.1	77.5	Alport syndrome, 301050
COL4A6	HEARING IMPAIRMENT MENDELIOME	81.2	96	89.4	?Deafness, X-linked 6, 300914
COL5A1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	114.3	97.7	95	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	89.1	99.5	97.4	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	137.2	99.5	97.8	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	165.3	99.3	98.4	?Myosclerosis, congenital, 255600 Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	174.7	100	99.9	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	SKIN DISORDERS	129.5	99.6	97.5	EBD inversa, 226600

	MENDELIOME PRECONCEPTION SCREENING				EBD, Bart type, 132000 EBD, localisata variant, 0 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COL8A2	VISION DISORDERS MENDELIOME	37.5	84.6	69.4	Corneal dystrophy, Fuchs endothelial, 1, 136800 Corneal dystrophy, posterior polymorphous 2, 609140
COL9A1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	121.2	99.5	96.9	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	65.1	98.3	88.8	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	65.5	95.6	86.7	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
COLEC10	MENDELIOME	162.6	100	99.1	3MC syndrome 3, 248340
COLEC11	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	203	100	100	3MC syndrome 2, 265050
COLQ	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	113.2	99.8	98.1	Myasthenic syndrome, congenital, 5, 603034
COMP	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	121.4	93.6	92.4	Epiphyseal dysplasia, multiple, 1, 132400 Pseudoachondroplasia, 177170
COMT	METABOLIC DISORDERS	206.6	100	99.9	{Panic disorder, susceptibility to}, 167870 {Schizophrenia, susceptibility to}, 181500
COPA	PRIMARY IMMUNODEFICIENCIES	133.2	100	100	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
COPB2	MENDELIOME	149.7	99.9	99.1	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	MOVEMENT DISORDERS	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426

	EPILEPSY HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				{Multiple system atrophy, susceptibility to}, 146500
COQ4	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	89.8	88.4	84.9	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	METABOLIC DISORDERS MITOCHONDRIAL DISORDERS	184.4	100	100	No OMIM phenotype Cerebellar ataxia and static encephalopmyopathy (Malicdan (2018) Hum Mutat 39,69) Intellectual disability (Najmabadi (2011) Nature 478,57)
COQ6	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	143.9	99.3	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	158.5	99.7	98.9	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	134.3	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	90.5	100	99.1	Nephrotic syndrome, type 9, 615573
COQ9	MOVEMENT DISORDERS METABOLIC DISORDERS	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654

	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
CORIN	MENDELIOME	179.1	99.9	99.4	Preeclampsia/eclampsia 5, 614595
CORO1A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	154.4	99.8	96.9	Immunodeficiency 8, 615401
COX10	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	241.9	100	99.6	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	MENDELIOME MITOCHONDRIAL DISORDERS	108.1	100	99.9	?Mitochondrial complex IV deficiency, 220110
COX15	HEART PANEL INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	58.1	83	65.4	Mitochondrial complex IV deficiency, 220110
COX4I1	MITOCHONDRIAL DISORDERS	133.9	100	100	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
COX4I2	SKIN DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	120.1	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	MITOCHONDRIAL DISORDERS	37.6	80.2	57.7	No OMIM phenotype
COX5B	MITOCHONDRIAL DISORDERS	126.9	100	100	No OMIM phenotype
COX6A1	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	180.6	100	99.4	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	MITOCHONDRIAL DISORDERS	32.4	97.5	80.8	No OMIM phenotype
COX6B1	INTELLECTUAL DISABILITY MENDELIOME	159.6	100	100	Mitochondrial complex IV deficiency, 220110

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
COX6B2	MITOCHONDRIAL DISORDERS	62.3	100	99.3	No OMIM phenotype
COX6C	MITOCHONDRIAL DISORDERS	131.4	99.2	90.9	No OMIM phenotype
COX7A1	MITOCHONDRIAL DISORDERS	81.7	99.9	98	No OMIM phenotype
COX7A2	MITOCHONDRIAL DISORDERS	82.9	99.3	92.7	No OMIM phenotype {insulin secretion,association with} (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	SKIN DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	47.9	73.3	42	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	MITOCHONDRIAL DISORDERS	247.3	100	100	No OMIM phenotype
COX7C	MITOCHONDRIAL DISORDERS	48.9	99	94.4	No OMIM phenotype
COX8A	MENDELIOME MITOCHONDRIAL DISORDERS	98.1	100	100	?Mitochondrial complex IV deficiency, 220110
COX8C	MITOCHONDRIAL DISORDERS	159.2	99.9	97.9	No OMIM phenotype ?Tethered spinal cord syndrome (Zhao (2016) Neural Regen Res 11, 1333)
CP	MOVEMENT DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	120	93.9	89.6	Cerebellar ataxia, 604290 HEMOSTATIC/THROMBOTIC DISORDERSiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CPA6	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	118.3	99.8	98.5	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPAMD8	MENDELIOME	102	94.7	91.4	Anterior segment dysgenesis 8, 617319
CPLX1	INTELLECTUAL DISABILITY MENDELIOME	79.8	99.9	97.9	Epileptic encephalopathy, early infantile, 63, 617976
CPN1	MENDELIOME PRECONCEPTION SCREENING	118.3	100	98.8	Carboxypeptidase N deficiency, 212070
CPOX	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	116.8	95.2	88.1	Coproporphyria, 121300 Harderoporphyria, 121300
CPS1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.8	100	99.8	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT1A	HEART PANEL METABOLIC DISORDERS	169.3	100	98.7	CPT deficiency, hepatic, type IA, 255120

	MENDELIOME PRECONCEPTION SCREENING				
CPT1C	MENDELIOME	111.8	100	99.9	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	EPILEPSY HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	162.8	97.2	95.4	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	160.7	100	99.8	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CRADD	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	115.2	99.9	98.5	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	MENDELIOME	126.6	100	99.9	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	191.8	100	100	Leber congenital amaurosis 8, 613835 Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105
CRB2	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	112.4	99.4	94.7	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.7	87.8	83.8	Mental retardation, autosomal recessive 2, 607417
CREB1	MENDELIOME	147.3	98.6	93.7	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	113.8	99.4	97	Osteogenesis imperfecta, type XVI, 616229
CREBBP	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	123.5	99.4	96.7	Rubinstein-Taybi syndrome 1, 180849
CRELD1	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	114.4	99.9	97.8	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 {Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	MENDELIOME PRECONCEPTION SCREENING	34.5	96.5	74.8	Short stature with microcephaly and distinctive facies, 615789
CRKL	CONGENITAL HEART DISEASE HEART PANEL	166.4	100	99.8	No OMIM phenotype ?Congenital heart defect (Breckpot (2012) Am J Med Genet A 158A,574) ?Tetralogy of Fallot (Tomita-Mitchell (2012) Physiol Genomics 44,518)

					?Ventricular septal defect (Zhao (2013) Am J Med Genet A 161,3087
CRLF1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105.6	90.9	89.2	Cold-induced sweating syndrome 1, 272430
CRP		242.2	100	98.4	
CRTAP	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	110.4	99.8	97.3	Osteogenesis imperfecta, type VII, 610682
CRTC1	MENDELIOME	135.2	99	95.7	Mucoepidermoid salivary gland carcinoma, 0
CRX	VISION DISORDERS MENDELIOME	114.2	99.9	98.7	Cone-rod retinal dystrophy-2, 120970 Leber congenital amaurosis 7, 613829
CRYAA	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.3	92.7	86.2	Cataract 9, multiple types, 604219
CRYAB	VISION DISORDERS CARDIO HEART PANEL MENDELIOME MUSCLE DISORDERS	125.7	99.9	98.7	Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, 2, 608810 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBA1	VISION DISORDERS MENDELIOME	135.7	100	99.2	Cataract 10, multiple types, 600881
CRYBA2	VISION DISORDERS MENDELIOME	150.6	100	100	?Cataract 42, 115900
CRYBA4	VISION DISORDERS MENDELIOME	117.5	100	100	Cataract 23, 610425
CRYBB1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	129.1	100	99.4	Cataract 17, multiple types, 611544
CRYBB2	VISION DISORDERS MENDELIOME	150.3	100	100	Cataract 3, multiple types, 601547
CRYBB3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	144.3	100	100	Cataract 22, 609741
CRYGB	VISION DISORDERS MENDELIOME	97.4	99.8	97.4	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	VISION DISORDERS MENDELIOME	128.5	100	99.3	Cataract 2, multiple types, 604307
CRYGD	VISION DISORDERS MENDELIOME	100.4	100	99.8	Cataract 4, multiple types, 115700
CRYGS	VISION DISORDERS MENDELIOME	105.4	96.4	88	Cataract 20, multiple types, 116100

CRYM	HEARING IMPAIRMENT MENDELIOME	97.4	99.9	98.3	Deafness, autosomal dominant 40, 616357
CSF1R	MOVEMENT DISORDERS MENDELIOME PARK	139.5	99.5	98.4	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	66	89.9	88.2	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	94.8	99.6	97.8	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	94.4	99.3	96.5	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSGALNACT1	SHORT STATURE/SKELETAL DYSPLASIA	193.1	100	100	No OMIM phenotype Skeletal dysplasia and joint laxity (Vodopiutz (2017) Hum Mutat 38,34) ?Hemi-facial palsy (Saigoh (2011) J Hum Genet 56,143) ?Neuropathy, hereditary motor and sensory (Saigoh (2011) J Hum Genet 56,143)
CSNK1D	MENDELIOME	148.4	94.8	90.4	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK2A1	INTELLECTUAL DISABILITY MENDELIOME	126.4	94.1	86.2	Okur-Chung neurodevelopmental syndrome, 617062
CSPP1	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	112	99.8	97.8	Joubert syndrome 21, 615636
CSRP3	CARDIO HEART PANEL MENDELIOME	103	100	99.9	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CST3	MENDELIOME	106.4	92	74.3	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CST6	SKIN DISORDERS	112.7	99.2	93.3	No OMIM phenotype Hypotrichosis
CSTA	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	119	99.9	99	Peeling skin syndrome 4, 607936
CSTB	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	82.5	97.1	82.7	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800

CTBP1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	93.4	96.1	85	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	INTELLECTUAL DISABILITY MENDELIOME	158.4	98.6	96.7	Mental retardation, autosomal dominant 21, 615502
CTDP1	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105	86.6	83.6	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTF1	HEART PANEL	24.5	28	20	No OMIM phenotype Cardiomyopathy,dilated (Erdmann (2000) Hum Mutat 16,448)
CTH	METABOLIC DISORDERS MENDELIOME	152.6	99.9	98.1	Cystathioninuria, 219500 Homocysteine, total plasma, elevated, 0
CTHRC1	MENDELIOME	90.2	91.9	84.5	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	193.9	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700
CTNNA1	VISION DISORDERS MENDELIOME HEREDITARY CANCER	125.6	99.9	99	Macular dystrophy, patterned, 2, 608970 Gastric cancer
CTNNA2	MENDELIOME	124.6	99.9	99.3	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	CARDIO HEART PANEL MENDELIOME	138.3	100	99.9	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	163.8	100	99.9	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600

CTNND1	MENDELIOME	153.8	100	99.9	Blepharocheilodontic syndrome 2, 617681
CTNND2	INTELLECTUAL DISABILITY	113.7	93.2	91	No OMIM phenotype Autism (Turner (2015) Nature 520,51) Intellectual disability (Hofmeister (2015) J Med Genet 52,111)
CTNS	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	120.1	100	99.9	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CTPS1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	143.1	100	99.6	Immunodeficiency 24, 615897
CTR9	HEREDITARY CANCER	154.2	100	99.8	No OMIM phenotype Wilms tumor (Hanks (2014) Nat Commun 5, 4398)
CTSA	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	134.1	100	99.4	Galactosialidosis, 256540
CTSB	SKIN DISORDERS	137.3	100	100	Keratolytic winter erythema, 148370
CTSC	 SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.5	100	100	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CTSD	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	163.7	98	95.3	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	112.8	84.2	80.2	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	105	100	99.9	Pycnodysostosis, 265800
CTTNBP2	INTELLECTUAL DISABILITY	140.7	99.6	97.3	No OMIM phenotype

					?Autism (Iossifov (2012) Neuron 74,285)
CTU2	MENDELIOME	106.7	98.6	91.7	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.8	99.8	98.4	Megaloblastic anemia-1, Finnish type, 261100
CUL3	RENAL DISORDERS MENDELIOME	110.3	98.6	94.7	Pseudohypoaldosteronism, type IIE, 614496
CUL4B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	72.8	98	88.5	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	149.6	99.8	97.9	3-M syndrome 1, 273750
CUX1	INTELLECTUAL DISABILITY	109.2	94.6	91.9	No OMIM phenotype
CUX2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	85.6	99.8	97.9	Epileptic encephalopathy, early infantile, 67, 618141
CWC27	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	74.7	97.2	89.7	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	119.1	99.5	96.7	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCL10		174.4	100	99.9	
CXCL13		193.2	100	100	
CXCL2		64	99.9	93.8	
CXCR4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	202.7	100	99.9	Myelokathexis, isolated, 0 WHIM syndrome, 193670
CXorf56	INTELLECTUAL DISABILITY MENDELIOME	91.8	99.7	95.5	?Mental retardation, X-linked 107, 301013
CYB561	METABOLIC DISORDERS MENDELIOME	131	92.8	92.6	Orthostatic hypotension 2, 618182
CYB5A	DSD MENDELIOME PRECONCEPTION SCREENING	133.5	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	147.3	98	98	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800

	PRECONCEPTION SCREENING				
CYBA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	97.3	77.9	71	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	PRIMARY IMMUNODEFICIENCIES MENDELIOME	110.8	99.9	99.2	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
CYBRD1	IRON DISORDERS	131.6	100	100	No OMIM phenotype Iron overload (Zaahl (2004) Hum Genet 115,409 {Haemochromatosis,phenotype modifier,association with} (Constantine (2009) Br J Haematol 147,140)
CYC1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	184.5	88.1	86.8	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	72.1	99.6	95	Thrombocytopenia 4, 612004
CYFIP2	MENDELIOME	139.9	100	99.6	Epileptic encephalopathy, early infantile, 65, 618008
CYLD	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	119.9	98.1	93	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP11A1	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.9	99.6	97.7	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	175.9	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	173.4	100	100	Aldosterone to renin ratio raised, 0 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 {Low renin hypertension, susceptibility to}, 0
CYP17A1	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	135	100	99.7	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	160.6	99.1	97.3	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	VISION DISORDERS	134.8	100	100	Anterior segment dysgenesis 6, multiple subtypes, 617315

	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300
CYP21A2	DSD METABOLIC DISORDERS MENDELIOME	93.8	95.8	86.6	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	165.3	100	100	Hypercalcemia, infantile, 1, 143880
CYP26B1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	178.1	100	99.9	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	87.2	99.5	95.6	Focal facial dermal dysplasia 4, 614974
CYP27A1	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	175.1	98.3	96.1	Cerebrotendinous xanthomatosis, 213700
CYP27B1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.1	100	99.1	Vitamin D-dependent rickets, type I, 264700
CYP2A6	MENDELIOME	167.9	100	99.8	Coumarin resistance, 122700 {Lung cancer, resistance to}, 211980 {Nicotine addiction, protection from}, 188890
CYP2B6	MENDELIOME	120.3	100	98.9	Efavirenz, poor metabolism of, 614546 {Efavirenz central nervous system toxicity, susceptibility to}, 614546
CYP2C19	MENDELIOME	186.3	99.5	96.3	Clopidogrel, impaired responsiveness to, 609535 Mephenytoin poor metabolizer, 609535 Omeprazole poor metabolizer, 609535 Proguanil poor metabolizer, 609535
CYP2C8	PRECONCEPTION SCREENING	109.1	98	93.7	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2C9	MENDELIOME	186.9	99.3	94.4	Tolbutamide poor metabolizer, 0 Warfarin sensitivity, 122700
CYP2R1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.5	96.4	86.6	Rickets due to defect in vitamin D 25-hydroxylation, 600081
CYP2U1	MOVEMENT DISORDERS	119.2	93.7	91.2	Spastic paraplegia 56, autosomal recessive, 615030

	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
CYP4F22	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.7	100	99.7	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.5	99.8	98.5	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.2	94.7	87.7	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
D2HGDH	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	134.5	97.5	95.2	D-2-hydroxyglutaric aciduria, 600721
DAB1	MENDELIOME	123.9	100	100	Spinocerebellar ataxia 37, 615945
DAB2IP		168.7	98.6	96.9	
DACT1	MENDELIOME	127.1	92.5	89.4	?Townes-Brocks syndrome 2, 617466
DAG1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	220.8	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DAO	METABOLIC DISORDERS	134.5	100	99.9	{Schizophrenia}, 181500
DARS	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	98.6	98.7	93.8	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	122.3	100	99.6	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	140.7	100	99.8	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	102.1	97.3	93.8	Maple syrup urine disease, type II, 248600

	PRECONCEPTION SCREENING				
DCAF17	MOVEMENT DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	91.9	95.6	89.3	Woodhouse-Sakati syndrome, 241080
DCAF8	NEUROPATHIES MENDELIOME	124.3	100	99.9	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	MOVEMENT DISORDERS HH INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	138.5	100	99.9	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCDC2	CILIO HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	150.5	99.9	99.6	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DCHS1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	160.1	99.8	99.2	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	128.8	98.2	94.5	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCN	VISION DISORDERS MENDELIOME	140	95.7	94.9	Corneal dystrophy, congenital stromal, 610048
DCPS	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.8	100	99.8	Al-Raqad syndrome, 616459
DCTN1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PARK	131.6	99.7	98.3	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCX	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	113.2	100	99.7	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067
DCXR	METABOLIC DISORDERS	163	99.9	98.4	[Pentosuria], 260800
DDB2	SKIN DISORDERS	162.4	100	99.7	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740

	MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER				
DDC	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101	99.1	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	MOVEMENT DISORDERS VISION DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	141.8	97.1	94.8	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	149.7	99.9	98	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	METABOLIC DISORDERS MENDELIOME	120.5	99.9	99.1	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	155	100	99.9	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	MENDELIOME	87.2	100	99.9	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	113.9	86	81	Warsaw breakage syndrome, 613398
DDX3X	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	80.5	85.9	82.1	Mental retardation, X-linked 102, 300958
DDX58	PRIMARY IMMUNODEFICIENCIES MENDELIOME	123.3	98.6	95	Singleton-Merten syndrome 2, 616298
DDX59	CILIO INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	151.7	99.7	97.6	Orofaciodigital syndrome V, 174300
DEAF1	INTELLECTUAL DISABILITY MENDELIOME	125.9	88.3	83.7	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828
DENND5A	EPILEPSY INTELLECTUAL DISABILITY	123	99.8	97.9	Epileptic encephalopathy, early infantile, 49, 617281

	MENDELIOME PRECONCEPTION SCREENING				
DEPDC5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	148.3	99.8	99.3	Epilepsy, familial focal, with variable foci 1, 604364
DES	CARDIO HEART PANEL MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	120.8	99.9	98.1	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DFNA5	HEARING IMPAIRMENT MENDELIOME	112.9	99.9	99.4	Deafness, autosomal dominant 5, 600994
DFNB59	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	123.6	100	99.2	Deafness, autosomal recessive 59, 610220
DGAT1	METABOLIC DISORDERS MENDELIOME	156.2	88.5	86.3	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	142.3	99.5	95.2	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DGUOK	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHCR24	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	183	100	100	Desmosterolosis, 602398
DHCR7	SKIN DISORDERS DSD METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	158.3	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	MOVEMENT DISORDERS	93.5	97.8	94.8	?Congenital disorder of glycosylation, type 1bb, 613861

	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DHFR	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	48.4	91.1	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	DSD MENDELIOME PRECONCEPTION SCREENING	117.7	100	100	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	CRANIOFACIAL ANOMALIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	92.2	100	99.9	Miller syndrome, 263750
DHTKD1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	141	99.6	98.2	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX30	INTELLECTUAL DISABILITY MENDELIOME	160.6	99.9	99.2	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX38	VISION DISORDERS MENDELIOME	130.3	99.9	99	Retinitis pigmentosa 84, 618220
DIABLO	HEARING IMPAIRMENT MENDELIOME	206	100	99.5	Deafness, autosomal dominant 64, 614152
DIAPH1	HEARING IMPAIRMENT HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	120.7	99.3	97.8	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	MENDELIOME	61.7	93.1	79.7	?Premature ovarian failure 2A, 300511
DIAPH3	HEARING IMPAIRMENT MENDELIOME	73.9	97.9	90.1	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	MENDELIOME HEREDITARY CANCER	145.4	99.5	98.1	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295

DIP2B	INTELLECTUAL DISABILITY MENDELIOME	160.3	99.2	98.1	Mental retardation, FRA12A type, 136630
DIS3L2	MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	158.5	99.8	99	Perlman syndrome, 267000
DISP1	CRANIOFACIAL ANOMALIES	211.4	99.9	99.1	No OMIM phenotype Craniofacial and neuro-developmental abnormalities (Roessler (2009) Hum Genet 125,393) Diaphragmatic hernia, congenital (Kantarci (2010) Am J Med Genet A 152A,2493) Tetralogy of Fallot (Silversides (2012) PLoS Genet 8, e1002843)
DYSKERATOSIS CONGENITA1	VISION DISORDERS BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
DLAT	MOVEMENT DISORDERS EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	91.6	99.1	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	MENDELIOME	179.2	99.9	99.4	Colorectal cancer, somatic, 114500
DLD	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	123.5	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	INTELLECTUAL DISABILITY MENDELIOME	90	99.3	94.3	Mental retardation, X-linked 90, 300850
DLG4	INTELLECTUAL DISABILITY	170.4	100	99.4	no OMIM phenotype Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394)
DLL3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	64.1	88.8	79.9	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	MENDELIOME	181.7	99.8	98.8	Adams-Oliver syndrome 6, 616589
DLST	MITOCHONDRIAL DISORDERS	95.8	94.2	89.7	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
DLX3	CRANIOFACIAL ANOMALIES	109.8	100	99.1	Amelogenesis imperfecta, type IV, 104510

	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME				Trichodontoosseous syndrome, 190320
DLX4	CRANIOFACIAL ANOMALIES MENDELIOME	162.4	100	100	?Orofacial cleft 15, 616788
DLX5	SKIN DISORDERS MENDELIOME	123.6	99.9	97	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMAC1	MITOCHONDRIAL DISORDERS	50.4	99.8	96.3	No OMIM phenotype Complex I assembly factor
DMAC2	MITOCHONDRIAL DISORDERS	135.1	98.3	98.3	No OMIM phenotype Complex I assembly factor
DMD	HEART PANEL INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	112.4	99.4	97.4	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	157	98.8	97.2	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	159.5	99.9	99.1	Hypophosphatemic rickets, AR, 241520
DMPK	INTELLECTUAL DISABILITY MENDELIOME	117.7	99.9	97.9	Myotonic dystrophy 1, 160900
DMRT1	DSD	99.4	99.3	92.4	No OMIM phenotype XY gonadal dysgenesis (Ledig (2010) Hum Reprod 25,2637) Azoospermia (Lopes (2013) PLoS Genet 9,e1003349) ?Male infertility (Tewes (2014) Fertil Steril 102, 816) ?XY sex reversal (Raymond (1999) Hum Mol Genet 8, 989)
DMRT2	DSD	154.3	98.4	90.6	No OMIM phenotype 46,XY DSD (deletion)
DMXL2	HEARING IMPAIRMENT MENDELIOME	181.4	98.6	96	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	123.6	99.8	96.9	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAAF1	CILIO MENDELIOME PRECONCEPTION SCREENING	115.8	100	99.7	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	CILIO MENDELIOME	105.1	99.7	96.9	Ciliary dyskinesia, primary, 10, 612518

	PRECONCEPTION SCREENING				
DNAAF3	CILIO MENDELIOME PRECONCEPTION SCREENING	91.8	97.7	90.6	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	CILIO MENDELIOME PRECONCEPTION SCREENING	79.6	96.3	84.1	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	CILIO MENDELIOME PRECONCEPTION SCREENING	107.9	84.5	78.2	Ciliary dyskinesia, primary, 18, 614874
DNAH1	CILIO MENDELIOME	183.3	100	99.7	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	CILIO MENDELIOME PRECONCEPTION SCREENING	134	99.8	98.4	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	CILIO	136.9	100	99.7	Ciliary dyskinesia,primary
DNAH5	CILIO MENDELIOME PRECONCEPTION SCREENING	123.8	99.7	98.5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	CILIO MENDELIOME PRECONCEPTION SCREENING	135.3	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	CILIO MENDELIOME PRECONCEPTION SCREENING	156.6	98.4	95.5	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB11	RENAL DISORDERS MENDELIOME	111.3	100	99.6	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	CILIO MENDELIOME	138.3	100	97.3	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	102	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB6	MENDELIOME MUSCLE DISORDERS	59.8	91.7	79.3	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.3	87.4	87.3	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	97.9	98.5	90	3-methylglutaconic aciduria, type V, 610198

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
DNAJC21	BONE MARROW FAILURE MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	125.7	99.8	98.5	Bone marrow failure syndrome 3, 617052
DNAJC3	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	116.3	99.9	98.1	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC5	EPILEPSY MENDELIOME	200.2	100	99.9	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNAJC6	MENDELIOME PARK PRECONCEPTION SCREENING	160.8	99.9	98.9	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	CILIO MENDELIOME PRECONCEPTION SCREENING	99	95.7	84.5	Ciliary dyskinesia, primary, 16, 614017
DNAL4	MOVEMENT DISORDERS MENDELIOME	72.6	99.3	93.5	?Mirror movements 3, 616059
DNASE1	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES	198.8	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE1L3	MENDELIOME PRECONCEPTION SCREENING	141.3	100	100	Systemic lupus erythematosus 16, 614420
DNHD1	CILIO	173.1	100	99.9	Joubert syndrome
DNM1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	156.7	89.3	87.5	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	VISION DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	123.5	99.7	96.6	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	NEUROPATHIES METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS	127.4	97.5	94.4	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME	113.4	99.2	98.3	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116

DNMT3A	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	115.5	98.8	95.8	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879
DNMT3B	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	143.8	100	99.9	Immunodeficiency 40, 616433
DOCK6	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	119.9	98.9	96.5	Adams-Oliver syndrome 2, 614219
DOCK7	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.4	97.9	95.6	Epileptic encephalopathy, early infantile, 23, 615859
DOCK8	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	129.1	100	99.8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	 MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	105.7	93.3	92.5	?Fetal nesia deformation sequence, 208150 Myasthenic syndrome, congenital, 10, 254300
DOLK	SKIN DISORDERS HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	202.9	100	99.9	Congenital disorder of glycosylation, type Im, 610768
DONSON	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	104.9	83.9	78.3	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DPAGT1	EPILEPSY METABOLIC DISORDERS	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS				
DPF2	INTELLECTUAL DISABILITY MENDELIOME	102.4	100	97.9	Coffin-Siris syndrome 7, 618027
DPH1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	157.1	100	99.7	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901
DPM1	EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	131.1	91.7	86.7	Congenital disorder of glycosylation, type Ie, 608799
DPM2	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	102.1	100	99.4	Congenital disorder of glycosylation, type Iu, 615042
DPM3	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	183.9	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	145.5	96.5	94.5	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPY19L2	MENDELIOME PRECONCEPTION SCREENING	94.8	72.9	66.6	Spermatogenic failure 9, 613958
DPYD	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	158.3	95.6	93.7	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	133.5	100	99.5	Dihydropyrimidinuria, 222748

	PRECONCEPTION SCREENING				
DRAM2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.8	100	99.7	Cone-rod dystrophy 21, 616502
DRC1	CILIO MENDELIOME PRECONCEPTION SCREENING	97	99.9	98.6	Ciliary dyskinesia, primary, 21, 615294
DRD4	MENDELIOME	79	80.9	70.7	Autonomic nervous system dysfunction, 0 [Novelty seeking personality], 601696 {Attention deficit-hyperactivity disorder}, 143465
DSC2	CARDIO SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	128.5	99.4	96.2	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	87.1	97.2	89.6	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	SKIN DISORDERS MENDELIOME	124.3	99.8	98.5	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	SKIN DISORDERS MENDELIOME	175	98.4	96.1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG2	CARDIO HEART PANEL MENDELIOME	140.6	99.9	98.7	Arrhythmogenic right ventricular dysplasia 10, 610193 Cardiomyopathy, dilated, 1BB, 612877
DSG3	SKIN DISORDERS	148.9	99.8	98.5	No OMIM phenotype
DSG4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	198.4	98.5	95.7	Hypotrichosis 6, 607903
DSP	CARDIO SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	154	100	99.8	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DSPP	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME	155.7	99.9	99.3	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	SKIN DISORDERS	154.1	99.7	98.2	?Neuropathy, hereditary sensory and autonomic, type VI, 614653

	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING				Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	RENAL DISORDERS MENDELIOME	144.7	100	99.4	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNA	HEART PANEL MENDELIOME	156.5	100	100	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.2	99.3	95.1	Hermansky-Pudlak syndrome 7, 614076
DUOX2	MENDELIOME PRECONCEPTION SCREENING	136.4	96.7	94.8	Thyroid dyshormonogenesis 6, 607200
DUOXA2	MENDELIOME PRECONCEPTION SCREENING	122.2	100	99.9	Thyroid dyshormonogenesis 5, 274900
DUSP6	SKIN DISORDERS HH MENDELIOME	175.9	100	99.9	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	113.9	97.8	94.1	Robinow syndrome, autosomal dominant 2, 616331
DVL3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	149.4	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYM	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101.3	97.2	94.8	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326
DYNC1H1	EPILEPSY NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	179.8	100	99.6	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYNC2H1	CILIO DSD SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	CILIO SHORT STATURE/SKELETAL DYSPLASIA	95.1	99.3	96	Short-rib thoracic dysplasia 15 with polydactyly, 617088

	MENDELIOME PRECONCEPTION SCREENING				
DYRK1A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	159.6	100	100	Mental retardation, autosomal dominant 7, 614104
DYRK1B	MENDELIOME	105	93	85	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	133.1	100	99.8	Miyoshi muscular dystrophy 1, 254130 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	RENAL DISORDERS MENDELIOME	98.3	99.4	95.5	Polycystic kidney disease 5, 617610
EARS2	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	103.4	99.7	98.3	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	INTELLECTUAL DISABILITY MENDELIOME	130.4	100	99.1	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	83.3	100	98	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECE1	MENDELIOME	144.6	97.9	97.8	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 {Hypertension, essential, susceptibility to}, 145500
ECEL1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	100.7	88.8	83.1	Arthrogryposis, distal, type 5D, 615065
ECHS1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	112.8	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	170.8	100	99.7	Urbach-Wiethe disease, 247100
ECSIT	MITOCHONDRIAL DISORDERS	141.4	99.7	98	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
EDA	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	88.5	85.7	77.3	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500

EDAR	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.6	100	99.6	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	99.1	99.3	93.3	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDC3	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	144.7	100	99.6	?Mental retardation, autosomal recessive 50, 616460
EDN1	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	145.5	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDN3	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	134.4	100	99.5	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	218.9	100	99.7	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	131	95.6	90.9	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EED	INTELLECTUAL DISABILITY MENDELIOME	94.2	98.4	91.6	Cohen-Gibson syndrome, 617561
EEF1A2	EPILEPSY HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	177.7	98.8	93.8	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EEF2	MENDELIOME	167.6	100	99.9	?Spinocerebellar ataxia 26, 609306
EFEMP1	VISION DISORDERS MENDELIOME	167.9	100	99.6	Doyne honeycomb degeneration of retina, 126600
EFEMP2	SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	120.9	100	99.9	Cutis laxa, autosomal recessive, type IB, 614437
EFHC1	EPILEPSY	136.3	92.2	91.5	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631

					{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770
EFL1	BONE MARROW FAILURE SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	174.8	99.4	97.7	Shwachman-Diamond syndrome 2, 617941
EFNA4	CRANIOFACIAL ANOMALIES	106.6	100	100	No OMIM phenotype Craniosynostosis 1 (Merrill et al. (2006) Hum Molec Genet 15)
EFNB1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	118.5	100	99.9	Craniofrontonasal dysplasia, 304110
EFNB2	INTELLECTUAL DISABILITY	179.7	99.8	97.7	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/29508392
EFTUD2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	124.2	100	99.4	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	EPILEPSY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.2	100	99.8	Hypomagnesemia 4, renal, 611718
EGFR	MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	160.8	100	99.1	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	MENDELIOME	39	79	66.3	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070
EGR2	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	124.4	100	100	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
EHHADH	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	163.2	100	99.7	?Fanconi renotubular syndrome 3, 615605
EHMT1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	141.4	94	92.9	Kleefstra syndrome 1, 610253
EIF2AK3	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147.1	95.1	91.3	Wolcott-Rallison syndrome, 226980
EIF2AK4	MENDELIOME PRECONCEPTION SCREENING	146.3	99.8	98	Pulmonary venoocclusive disease 2, 234810

EIF2B1	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	149.9	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.9	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	163.8	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	146	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	128	99.6	97.9	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2S3	INTELLECTUAL DISABILITY MENDELIOME	84.9	97	89.8	MEHMO syndrome, 300148
EIF4A3	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	106.8	100	99.9	Robin sequence with cleft mandible and limb anomalies, 268305
ELAC2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	123.8	100	99.3	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELANE	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	80.9	99.7	95.9	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	PRIMARY IMMUNODEFICIENCIES	76.8	99.8	97.5	No OMIM phenotype ?Immunodeficiency, primary, modifier of (Stray-Pedersen (2017) J Allergy Clin Immunol 139,232) ?Hypogammaglobulinaemia (Stewart (2005) Curr Opin Allergy Clin Immunol 5,510)
ELMO2	MENDELIOME	134.7	100	99.1	Vascular malformation, primary intraosseous, 606893
ELMOD3	HEARING IMPAIRMENT MENDELIOME	156.2	100	99.7	?Deafness, autosomal recessive 88, 615429
ELN	CONGENITAL HEART DISEASE SKIN DISORDERS HEART PANEL MENDELIOME	91.1	99.4	97.4	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500

ELOVL1	SKIN DISORDERS METABOLIC DISORDERS	117	100	99.5	No OMIM phenotype Ichthyotic keratoderma,spasticity,hypomyelination and dysmorphic features
ELOVL4	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	91.9	99.9	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELOVL5	MOVEMENT DISORDERS MENDELIOME	120.9	100	99.8	Spinocerebellar ataxia 38, 615957
ELP1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	142.6	99.7	98.1	Dysautonomia, familial, 223900
ELP2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.5	99.2	96.9	Mental retardation, autosomal recessive 58, 617270
ELP4	MENDELIOME	54.9	72.2	66.7	?Aniridia 2, 617141
EMC1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.3	100	99.8	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMD	CARDIO HEART PANEL MENDELIOME MUSCLE DISORDERS	100.3	99.8	97.2	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	MENDELIOME PRECONCEPTION SCREENING	137.7	100	100	Bowen-Conradi syndrome, 211180
EMILIN1	 HEART PANEL	75.6	96.8	87.5	No OMIM phenotype Connective tissue disease, autosomal dominant (Capuano (2016) Hum Mutat 37, 84)
EML1	INTELLECTUAL DISABILITY MENDELIOME	163.6	99	97.1	Band heterotopia, 600348
EMP2	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.1	99.7	96.9	Nephrotic syndrome, type 10, 615861
EMX2	INTELLECTUAL DISABILITY MENDELIOME	118	100	100	Schizencephaly, 269160
ENAM	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.9	100	99.9	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG		128.8	97.4	93.6	Telangiectasia, hereditary hemorrhagic, type 1, 187300

	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME				
ENO3	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	179.3	100	100	?Glycogen storage disease XIII, 612932
ENPP1	SKIN DISORDERS HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	134.8	92.4	83.2	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
ENTPD1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	165.1	100	99.4	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	119.9	79.5	78.4	Adams-Oliver syndrome 4, 615297
EP300	INTELLECTUAL DISABILITY MENDELIOME	199.7	99.6	97.9	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	MENDELIOME	136.7	99.4	97.1	Erythrocytosis, familial, 4, 611783
EPB41	MENDELIOME PRECONCEPTION SCREENING	135.5	98.7	95.6	Elliptocytosis-1, 611804
EPB41L1	MENDELIOME	128.7	99.9	97.7	?Mental retardation, autosomal dominant 11, 614257
EPB42	MENDELIOME PRECONCEPTION SCREENING	160.7	100	99.7	Spherocytosis, type 5, 612690
EPCAM	MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	64.5	93.3	79.8	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	126	99.3	97.7	Vici syndrome, 242840
EPHA2	VISION DISORDERS MENDELIOME	175	99.2	97.8	Cataract 6, multiple types, 116600
EPHB4	MENDELIOME	140.6	99.8	98.6	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300

EPHX1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	122.6	98.8	96.1	?Hypercholanemia, familial, 607748
EPHX2	METABOLIC DISORDERS	114.2	100	99.1	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
EPM2A	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	110.1	86.2	84	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	MENDELIOME	90.2	98.1	96.4	?Diamond-Blackfan anemia-like, 617911 Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623
EPRS	MENDELIOME PRECONCEPTION SCREENING	126.3	100	99.2	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	HEARING IMPAIRMENT MENDELIOME	122.8	96.9	91.4	?Deafness, autosomal recessive 102, 615974
EPS8L2	HEARING IMPAIRMENT MENDELIOME	115.1	94.4	90.9	Deafness autosomal recessive 106, 617637
EPS8L3	SKIN DISORDERS	112.5	99.8	97.7	No OMIM phenotype Marie Unna hereditary hypotrichosis (Zhang (2012) J Med Genet 49,727)
ERAL1	HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS	181.5	100	100	Perrault syndrome 6, 617565
ERBB2	MENDELIOME	135	98.3	97.3	Adenocarcinoma of lung, somatic, 211980 Gastric cancer, somatic, 613659 Glioblastoma, somatic, 137800 Ovarian cancer, somatic, 0
ERBB3	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	139.2	100	99.9	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
ERBB4	ALS MENDELIOME	144.5	99.9	99.1	Amyotrophic lateral sclerosis 19, 615515
ERCC1	MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	76.6	100	97.3	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	123.7	100	99.7	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	SKIN DISORDERS	113.2	99.9	98.9	Trichothiodystrophy 2, photosensitive, 616390

	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER				Xeroderma pigmentosum, group B, 610651
ERCC4	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	139.2	100	99.5	?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	 SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	139.8	100	99.4	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	 SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	191.3	100	99.9	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC6L2	BONE MARROW FAILURE MENDELIOME PRECONCEPTION SCREENING	107.6	99.7	97.7	Bone marrow failure syndrome 2, 615715
ERCC8	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	89.5	92.9	78.4	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERF	CRANIOFACIAL ANOMALIES MENDELIOME	107.9	99.9	97.7	Chitayat syndrome, 617180 Craniosynostosis 4, 600775
ERG		115.7	99.1	98.9	
ERGIC1	MENDELIOME	195.2	95.3	94.7	?Arthrogryposis multiplex congenita, neurogenic type, 208100
ERLIN1	MENDELIOME PRECONCEPTION SCREENING	146.9	100	100	Spastic paraplegia 62, 615681
ERLIN2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	156	100	99.3	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	MENDELIOME	131	99.8	98.1	?Periventricular nodular heterotopia 6, 615544

ESCO2	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105.2	97.3	90.4	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ESPN	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	28.6	44.2	35.3	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 0
ESR1	MENDELIOME PRECONCEPTION SCREENING	127.5	99.9	99.4	Estrogen resistance, 615363 {Atherosclerosis, susceptibility to}, 0 {Breast cancer}, 114480 {HDL response to hormone replacement, augmented}, 0 {Migraine, susceptibility to}, 157300 {Myocardial infarction, susceptibility to}, 608446
ESR2	MENDELIOME HEREDITARY CANCER	133.7	100	99.5	?Ovarian dysgenesis 8, 618187 Medullary thyroid carcinoma (Smith (2016) Hum Mol Genet 25,1836) ?Primary amenorrhea (Asadi (2013) Clin Genet 83,497) ?Breast cancer, increased risk (Pylkas (2012) PLoS Genet 8,e1002734
ESRP1	HEARING IMPAIRMENT MENDELIOME	106.5	99.9	98.5	?Deafness, autosomal recessive 109, 618013
ESRRB	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	119.7	100	99.2	Deafness, autosomal recessive 35, 608565
ETFA	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	143.3	100	99.4	Glutaric acidemia IIA, 231680
ETFB	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	126.6	100	100	Glutaric acidemia IIB, 231680
ETFDH	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	105.4	100	99.3	Glutaric acidemia IIC, 231680
ETHE1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	85.5	99.3	95.8	Ethylmalonic encephalopathy, 602473
ETV6	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME HEREDITARY CANCER	140.1	100	99.9	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216

EVC	CILIO SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	110.4	93.2	89.8	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	CILIO SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	119.3	96.4	94.3	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EWSR1	MENDELIOME	76.6	91.1	80.9	Ewing sarcoma, 612219 Neuroepithelioma, 612219
EXOC6	IRON DISORDERS	87.6	96.9	90.4	No OMIM phenotype ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
EXOC8	CILIO	174.4	100	100	Joubert syndrome
EXOSC2	VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	142	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	88.5	97.3	89.4	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	80.1	91.8	76.9	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.8	97.5	87.4	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	183.3	100	99.9	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME HEREDITARY CANCER	105.4	99.9	98.5	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS	163.5	99.9	99.1	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701

	MENDELIOME HEREDITARY CANCER				
EXTL3	CILIO PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	206.4	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	VISION DISORDERS CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	144.2	100	99.7	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EYA4	HEARING IMPAIRMENT HEART PANEL MENDELIOME	160.6	100	99.5	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.8	98.9	94.9	Retinitis pigmentosa 25, 602772
EZH2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	139.5	99.8	97.6	Weaver syndrome, 277590
F10	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	185.1	99	98.3	Factor X deficiency, 227600
F11	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	155	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	111.4	100	99.5	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.3	100	99.4	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	113.5	96.6	87.6	Factor XIIIB deficiency, 613235
F2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	124.2	99.8	98.1	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050

					{Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	HEMOSTATIC/THROMBOTIC DISORDERS	102.6	100	99.9	No OMIM phenotype Impaired thrombin-induced platelet response (Bianchi et al. (2016) Blood 127(10):1249-1259)
F5	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	173.5	99	97.3	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	166.6	100	98.5	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	118	99.5	98.1	Hemophilia A, 306700
F9	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	144.5	99.2	95.9	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FA2H	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319
FAAP24	PRIMARY IMMUNODEFICIENCIES	118.5	98.5	94.8	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
FADD	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	142.8	100	99.6	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	HEART PANEL METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	151.3	100	99.9	Tyrosinemia, type I, 276700
FAM111A	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	292.2	100	99.9	Gracile bone dysplasia, 602361 Kenny-Caffey syndrome, type 2, 127000
FAM111B	SKIN DISORDERS	152.9	100	99.8	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and

	MENDELIOME MUSCLE DISORDERS				pulmonary fibrosis, 615704
FAM126A	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.2	97.3	95.2	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.2	98.5	95	Retinitis pigmentosa 28, 606068
FAM20A	CRANIOFACIAL ANOMALIES SKIN DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	105.4	98.4	92.1	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	101.3	100	98.9	Raine syndrome, 259775
FAM46A	MENDELIOME	146.1	99.9	99.7	Osteogenesis imperfecta, type XVIII, 617952
FAM58A	DSD RENAL DISORDERS MENDELIOME	73.2	82.8	78.8	STAR syndrome, 300707
FAM83G	SKIN DISORDERS	116.3	100	100	No OMIM phenotype Palmoplantar keratoderma with leukonychia and abundant curly hair (Maruthappu et al. (2016) ESDR)
FAM83H	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	76.7	94.9	87.7	Amelogenesis imperfecta, type IIIA, 130900
FAM92A	MENDELIOME	90	84.9	70	?Polydactyly, postaxial, type A9, 618219
FAN1	RENAL DISORDERS MENDELIOME HEREDITARY CANCER	143.9	100	99.9	Interstitial nephritis, karyomegalic, 614817 Colorectal cancer
FANCA	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	123.3	99.8	98.5	Fanconi anemia, complementation group A, 227650
FANCB	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	68.4	96.7	87.9	Fanconi anemia, complementation group B, 300514

	HEREDITARY CANCER				
FANCC	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	121.6	99.4	97.1	Fanconi anemia, complementation group C, 227645
FANCD2	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	127.6	98.7	95.5	Fanconi anemia, complementation group D2, 227646
FANCE	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	108	85.9	84.6	Fanconi anemia, complementation group E, 600901
FANCF	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	166.8	100	100	Fanconi anemia, complementation group F, 603467
FANCG	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	147.7	100	100	Fanconi anemia, complementation group G, 614082
FANCI	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	152.1	99.5	97.5	Fanconi anemia, complementation group I, 609053
FANCL	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	87.8	99.4	94.7	Fanconi anemia, complementation group L, 614083
FANCM	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME HEREDITARY CANCER	96.8	99.2	94.3	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAR1	MOVEMENT DISORDERS	80.4	96.3	92.4	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154

	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
FARS2	MOVEMENT DISORDERS EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	207.7	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	76.6	96	93.4	?Neurodevelopmental disorder with brain, liver, and lung abnormalities, 618007
FAS	PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	272.1	100	99.3	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	PRIMARY IMMUNODEFICIENCIES MENDELIOME	86.2	100	98.5	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FASTKD2	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	118.9	99.5	96.8	?Mitochondrial complex IV deficiency, 220110
FAT1	RENAL DISORDERS	204.6	100	100	No OMIM phenotype Nephrotic syndrome, tubular ectasia and haematuria (Gee (2016) Nat Commun 7,10822) Facioscapulohumeral dystrophy-like phenotype (Puppo (2015) Hum Mutat 36,443) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (201
FAT2	MENDELIOME	166.3	100	99.9	Spinocerebellar ataxia 45, 617769
FAT4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	224.5	100	99.9	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	SKIN DISORDERS NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	119.6	91.8	91.1	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895
FBN1	CONGENITAL HEART DISEASE HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	159.8	99.9	99.5	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308

					Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FBN2	HEART PANEL MENDELIOME	161.7	100	99.5	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FBP1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	127	100	98.8	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL3	INTELLECTUAL DISABILITY	208.9	100	100	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30481285
FBXL4	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	189.8	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	INTELLECTUAL DISABILITY MENDELIOME	75.9	93.7	84.9	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	108.8	93.5	89	?Mental retardation, autosomal recessive 45, 615979
FBXO32	HEART PANEL	150.6	100	100	No OMIM phenotype
FBXO38	MENDELIOME	179.7	99.7	98.6	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	MOVEMENT DISORDERS MENDELIOME PARK PRECONCEPTION SCREENING	189.4	98.5	93.3	Parkinson disease 15, autosomal recessive, 260300
FCGR1A	PRIMARY IMMUNODEFICIENCIES	82.3	47.5	46.5	[IgG receptor I, phagocytic, familial deficiency of], 0
FCGR2A	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES	244.9	100	100	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES	176.9	99.9	97.2	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	HEMOSTATIC/THROMBOTIC DISORDERS	206.8	98.1	97.6	Thrombocytopenic purpura, autoimmune, 188030
FCGR3A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	225	99.6	97.7	Immunodeficiency 20, 615707
FCGR3B	PRIMARY IMMUNODEFICIENCIES MENDELIOME	176.6	99.1	98.2	Neutropenia, alloimmune neonatal, 0
FCN3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	127.8	100	99.4	Immunodeficiency due to ficolin 3 deficiency, 613860

	PRECONCEPTION SCREENING				
FDCSP		204.4	98.2	92.9	
FDFT1	METABOLIC DISORDERS MENDELIOME	153.6	98.1	95.7	Squalene synthase deficiency, 618156
FDPS	SKIN DISORDERS MENDELIOME	72.8	99.7	95	Porokeratosis 9, multiple types, 616631
FDX2	MITOCHONDRIAL DISORDERS	114.3	99.9	99.3	No OMIM phenotype Mitochondrial muscle myopathy (Spiegel (2014) Eur J Hum Genet 22,902)
FDXR	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	93.4	100	99.1	Auditory neuropathy and optic atrophy, 617717
FECH	SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.9	99.9	99.4	Protoporphyria, erythropoietic, 1, 177000
FERMT1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	104.9	98.9	96.3	Kindler syndrome, 173650
FERMT3	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	122.4	100	98.9	Leukocyte adhesion deficiency, type III, 612840
FEZF1	HH MENDELIOME PRECONCEPTION SCREENING	158.8	99.9	99.3	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	157	99	96.6	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	190.8	99.7	97.9	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGD1	CRANIOFACIAL ANOMALIES EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	85.7	92.7	86.5	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGD4	NEUROPATHIES MENDELIOME	111.9	99.3	97.3	Charcot-Marie-Tooth disease, type 4H, 609311

	PRECONCEPTION SCREENING				
FGF10	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	142.2	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	EPILEPSY HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	95.5	99.6	96.3	Epileptic encephalopathy, early infantile, 47, 617166
FGF14	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	190.1	100	99.7	Spinocerebellar ataxia 27, 609307
FGF16	MENDELIOME	125.5	99.9	96.7	Metacarpal 4-5 fusion, 309630
FGF17	HH MENDELIOME	138.6	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	MENDELIOME	99.2	96.9	84.4	?Renal hypodysplasia/aplasia 2, 615721
FGF23	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	106	99.9	97.8	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF3	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	73.9	92	75.7	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	SKIN DISORDERS MENDELIOME	109.2	99.7	97.8	Trichomegaly, 190330
FGF8	CRANIOFACIAL ANOMALIES SKIN DISORDERS HH SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	111.4	90.2	79.7	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	165	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	CRANIOFACIAL ANOMALIES SKIN DISORDERS HH SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	148	99.7	98.3	Encephalocraniocutaneous lipomatosis, 613001 HEART PANELsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600

					Trigonocephaly 1, 190440
FGFR2	CRANIOFACIAL ANOMALIES SKIN DISORDERS DSD SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	140.1	97.4	96.4	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	110.2	99.6	97	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FGG	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	137	99.3	96.5	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FH	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING HEREDITARY CANCER	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800

FHL1	CARDIO HEART PANEL MENDELIOME MUSCLE DISORDERS	87.2	98.8	93	?Uruguay faciocardiomusculoskeletal syndrome, 300280 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 Myopathy, X-linked, with postural muscle atrophy, 300696 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Scapuloperoneal myopathy, X-linked dominant, 300695
FHL2	HEART PANEL	149.6	99.6	98.4	No OMIM phenotype Cardiomyopathy,hypertrophic (Friedrich (2014) Basic Res Cardiol 109,451) ?Distal myopathy (Evila (2016) Neuromuscul Disord 26,7)
FHOD3	HEART PANEL	135.2	99.9	98.5	No OMIM phenotype
FIBP	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123.3	100	100	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	ALS NEUROPATHIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	154.9	99.8	98.4	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FIGLA	MENDELIOME	81.8	94.3	89.4	Premature ovarian failure 6, 612310
FIGN	INTELLECTUAL DISABILITY	169.6	100	99.9	No OMIM phenotype
FKBP10	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	158.6	96.9	92.8	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	74.3	100	99.4	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	94.5	100	99.7	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	120	99.2	94.2	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152

	MUSCLE DISORDERS				Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLAD1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	191.6	100	98.9	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	160.5	100	99.5	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	234.1	100	99.9	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	SKIN DISORDERS MENDELIOME	691.5	100	100	Peeling skin syndrome 6, 618084
FLI1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	189.6	99.4	97.7	Bleeding disorder, platelet-type, 21, 617443
FLNA	EPILEPSY HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FLNB	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	149.9	99.8	99.2	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia, 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
FLNC	HEART PANEL MENDELIOME MUSCLE DISORDERS	165	100	99.7	Cardiomyopathy, familial hypertrophic, 26, 0 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	HH MENDELIOME	225.7	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT1		143.2	99.8	99	
FLT3	MENDELIOME	134.7	99.7	98.2	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	CONGENITAL HEART DISEASE	155.9	98.6	97.9	Hemangioma, capillary infantile, somatic, 602089

	SKIN DISORDERS HEART PANEL MENDELIOME				Lymphatic malformation 1, 153100
FLVCR1	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.5	99.2	95.8	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	 INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	159.7	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	86.6	83.1	77.7	Mental retardation, autosomal recessive 47, 616193
FMO3	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	153.5	99.9	99.2	Trimethylaminuria, 602079
FMR1	INTELLECTUAL DISABILITY MENDELIOME	78.9	94	84.7	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FN1	SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	145.4	100	99.5	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FNIP1	SKIN DISORDERS	153.4	99.7	98.1	No OMIM phenotype Multiple discoid fibromas (Claessens (2013) J Invest Dermatol 133 S136)
FOLR1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	150.4	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME	32.7	86	68.5	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482
FOXC2	SKIN DISORDERS RENAL DISORDERS MENDELIOME	44.3	95.2	78.8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXD4	HEART PANEL	3.1	25	13.3	No OMIM phenotype
FOXE1	CRANIOFACIAL ANOMALIES	29.3	72.3	56.2	Bamforth-Lazarus syndrome, 241850

	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING				{Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	VISION DISORDERS HEART PANEL MENDELIOME	20.6	69	47.8	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968 {Aortic , familial thoracic 11, susceptibility to}, 617349
FOXF1	MENDELIOME	93.5	98.9	93.7	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXF2	HEARING IMPAIRMENT	67.4	85	76.3	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30561639profound sensorineural hearing loss associated with incomplete partition type I anomaly of the cochlea
FOXG1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	157.5	84.8	81	Rett syndrome, congenital variant, 613454
FOXH1	CONGENITAL HEART DISEASE HEART PANEL	47.2	98.5	85	No OMIM phenotype Congenital heart defects (Roessler (2008) Am J Hum Genet 83,18) Ventricular septal defect (Wang (2010) Int J Cardiol 145,83)
FOXI1	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	152.5	100	100	Enlarged vestibular aqueduct, 600791
FOXL2	DSD MENDELIOME	39.5	92.8	71	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FOXN1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	112.5	100	99.5	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	MENDELIOME	139	92.8	85.7	Rhabdomyosarcoma, alveolar, 268220
FOXP1	INTELLECTUAL DISABILITY MENDELIOME	129.6	100	99.9	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	INTELLECTUAL DISABILITY MENDELIOME	160.2	98.9	96.9	Speech-language disorder-1, 602081
FOXP3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	124.6	98.7	91.6	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FOXRED1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	136.6	100	99.6	Mitochondrial complex I deficiency, nuclear type 19, 618241

FPR1	PRIMARY IMMUNODEFICIENCIES	216.9	100	100	No OMIM phenotype Congenital defects of Phagocyte number,function or both Nanamori M et al.,2004
FRAS1	DSD INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.8	100	99.7	Fraser syndrome 1, 219000
FREM1	VISION DISORDERS SKIN DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.4	99.9	99.1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	DSD INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	182.4	100	99.5	Fraser syndrome 2, 617666
FRMD4A	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	116.7	91.4	90.3	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	MOVEMENT DISORDERS VISION DISORDERS MENDELIOME	114.2	99.9	98.8	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FRMPD4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	114.1	99.7	98	Mental retardation, X-linked 104, 300983
FRRS1L	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.1	68.3	63.4	Epileptic encephalopathy, early infantile, 37, 616981
FSCN2	MENDELIOME	145.1	100	100	Retinitis pigmentosa 30, 607921
FSHB	HH MENDELIOME PRECONCEPTION SCREENING	149.1	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	MENDELIOME PRECONCEPTION SCREENING	140.9	100	98.8	Ovarian dysgenesis 1, 233300 Ovarian hyperstimulation syndrome, 608115 Ovarian response to FSH stimulation, 276400
FSIP2	MENDELIOME	95.6	99.3	97	Spermatogenic failure 34, 618153
FTCD	METABOLIC DISORDERS INTELLECTUAL DISABILITY	89.8	94.6	89.8	Glutamate formiminotransferase deficiency, 229100

	MENDELIOME PRECONCEPTION SCREENING				
FTH1	IRON DISORDERS MENDELIOME	96.4	95.4	84.2	?Hemochromatosis, type 5, 615517
FTL	MOVEMENT DISORDERS VISION DISORDERS IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PARK	147.7	99	93.2	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	118.9	83.7	82.5	Growth retardation, developmental delay, facial dysmorphism, 612938 {Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	INTELLECTUAL DISABILITY MENDELIOME	128.3	98.1	94.5	Mental retardation, X-linked 9/44, 309549
FUCA1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135	100	99.5	Fucosidosis, 230000
FUK	METABOLIC DISORDERS	98.2	98.4	96.7	No OMIM phenotype Ng et al Am J Hum Gen 2018
FUS	ALS MENDELIOME	137.4	97.6	94.7	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
FUT2	METABOLIC DISORDERS	173.7	100	100	[Bombay phenotype, digenic], 616754 {Norwalk virus infection, resistance to}, 0 {Vitamin B12 plasma level QTL1}, 612542
FUT6	METABOLIC DISORDERS MENDELIOME	165.8	100	100	Fucosyltransferase 6 deficiency, 613852
FUT8	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	166.1	99.9	98.8	Congenital disorder of glycosylation with defective fucosylation, 618005
FUZ	CILIO MENDELIOME	118.8	100	100	Neural tube defects, 182940
FXN	IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	75.2	85.7	75.9	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
FXYD2	EPILEPSY RENAL DISORDERS	96.7	99.8	99.2	Hypomagnesemia 2, renal, 154020

	MENDELIOME				
FYB1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	92.3	97.7	91.9	Thrombocytopenia 3, 273900
FYCO1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.7	100	100	Cataract 18, autosomal recessive, 610019
FZD1		175.1	87.1	85.8	
FZD2	SHORT STATURE/SKELETAL DYSPLASIA	176.6	98.3	94.8	No OMIM phenotype Omodysplasia, autosomal dominant (Saal (2015) Hum Mol Genet 24,3399)
FZD4	VISION DISORDERS MENDELIOME	224.2	100	99.8	Exudative vitreoretinopathy 1, 133780 Retinopathy of prematurity, 133780
FZD6	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	208.6	100	100	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
G6PC	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	180.7	100	100	Glycogen storage disease Ia, 232200
G6PC3	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	123.7	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	118.3	99.5	97.5	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GAA	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	128.5	100	99.9	Glycogen storage disease II, 232300
GAB1	HEARING IMPAIRMENT MENDELIOME	170	100	99.7	?Deafness, autosomal recessive 26, 605428
GABBR2	INTELLECTUAL DISABILITY MENDELIOME	136	95.2	92.4	Epileptic encephalopathy, early infantile, 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903 {Nicotine dependence, protection against}, 188890 {Nicotine dependence, susceptibility to}, 188890
GABRA1	EPILEPSY INTELLECTUAL DISABILITY	179.5	100	100	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136

	MENDELIOME				{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRA3	INTELLECTUAL DISABILITY	110.3	99.6	97.7	No OMIM phenotype
GABRB1	INTELLECTUAL DISABILITY MENDELIOME	174.1	100	99.9	Epileptic encephalopathy, early infantile, 45, 617153
GABRB2	INTELLECTUAL DISABILITY MENDELIOME	148.9	100	100	Epileptic encephalopathy, infantile or early childhood, 2, 617829
GABRB3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	140.5	98.1	93.7	Epileptic encephalopathy, early infantile, 43, 617113 {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	EPILEPSY MENDELIOME	137.9	91.1	90.3	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAD1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128.7	99.9	98.4	?Cerebral palsy, spastic quadriplegic, 1, 603513
GAL	MENDELIOME	164.2	100	98.4	?Epilepsy, familial temporal lobe, 8, 616461
GALC	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	100.6	98.9	94.6	Krabbe disease, 245200
GALE	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	154.8	100	100	Galactose epimerase deficiency, 230350
GALK1	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.4	100	99.7	Galactokinase deficiency with cataracts, 230200
GALM	METABOLIC DISORDERS	103	100	99.9	No OMIM phenotype Wada et al Genet Med 2018 Oct
GALNS	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.2	99	95.6	Mucopolysaccharidosis IVA, 253000
GALNT3	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS	128.2	99.2	96	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900

	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
GALT	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	168.7	100	100	Galactosemia, 230400
GAMT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	93.5	90.9	80.7	Cerebral creatine deficiency syndrome 2, 612736
GAN	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	190	100	99.9	Giant axonal neuropathy-1, 256850
GANAB	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME	120.1	99.9	98.9	Polycystic kidney disease 3, 600666
GARS	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	125.7	99.9	98.5	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GAS8	CILIO MENDELIOME PRECONCEPTION SCREENING	150.7	99.8	99.4	Ciliary dyskinesia, primary, 33, 616726
GATA1	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME	83.5	99.6	95.7	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367
GATA2	BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	119.6	99.9	98.5	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	186.8	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	CONGENITAL HEART DISEASE DSD	87.4	68.6	60.7	?Testicular anomalies with or without congenital heart disease, 615542 Atrial septal defect 2, 607941

	HEART PANEL MENDELIOME				Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429
GATA5	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	44.2	98.3	84.4	Congenital heart defects, multiple types, 5, 617912
GATA6	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	61.7	83.7	72.1	Atrial septal defect 9, 614475 Atrioventricular septal defect 5, 614474 Pancreatic agenesis and congenital heart defects, 600001 Persistent truncus arteriosus, 217095 Tetralogy of Fallot, 187500
GATAD1	HEART PANEL MENDELIOME	126.6	97	92	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	INTELLECTUAL DISABILITY MENDELIOME	129.8	100	99.8	Mental retardation, autosomal dominant 18, 615074
GATB	MITOCHONDRIAL DISORDERS	101.5	99.9	98.9	No OMIM phenotype
GATC	MITOCHONDRIAL DISORDERS	126.2	100	100	No OMIM phenotype
GATM	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	150.6	100	100	Cerebral creatine deficiency syndrome 3, 612718
GBA	MOVEMENT DISORDERS BONE MARROW FAILURE METABOLIC DISORDERS MENDELIOME PARK PRECONCEPTION SCREENING	240.3	100	100	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	176.2	99.9	99.3	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	145.5	99.6	97.2	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	MOVEMENT DISORDERS	147.6	99.9	99.1	Glutaricaciduria, type I, 231670

	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
GCH1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARK PRECONCEPTION SCREENING	74.4	97	86.5	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	141.4	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCLC	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	133.2	99.9	98.9	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCLM	METABOLIC DISORDERS	106	98	88.2	{Myocardial infarction, susceptibility to}, 608446
GCM2	RENAL DISORDERS MENDELIOME	161.8	100	100	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GCNT2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	166.8	99.5	99.5	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	34.2	83.1	67.8	?Glycine encephalopathy, 605899
GDAP1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	163.1	99.3	96.1	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400
GDAP2	MOVEMENT DISORDERS	131.1	99.7	97.2	No OMIM phenotype Cerebellar ataxia (Eidhof et al, Brain, vol 141, Issue 9)
GDF1	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME PRECONCEPTION SCREENING	19.5	65	48.4	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF2	SKIN DISORDERS HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS	163.2	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506

	MENDELIOME				
GDF3	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	134.9	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	141.8	100	100	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GDF6	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	75.2	98.7	89	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GDF9	MENDELIOME	169.5	100	100	?Premature ovarian failure 14, 618014
GDI1	INTELLECTUAL DISABILITY MENDELIOME	152	98.9	97.3	Mental retardation, X-linked 41, 300849
GDNF	MENDELIOME HEREDITARY CANCER	185.9	99.9	98.8	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GEMIN4	MENDELIOME	167.1	100	99.9	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFAP	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	102.6	91.7	90.3	Alexander disease, 203450
GFER	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	76.1	92.9	75.4	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFI1	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	83.1	99	92.9	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	170.2	98.2	95.7	Bleeding disorder, platelet-type, 17, 187900
GFM1	INTELLECTUAL DISABILITY	100.3	99.2	95.3	Combined oxidative phosphorylation deficiency 1, 609060

	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
GFM2	INTELLECTUAL DISABILITY MITOCHONDRIAL DISORDERS	118.6	98.7	93.4	No OMIM phenotype Leigh syndrome with arthrogryposis multiplex congenita (Fukumura (2015) J Hum Genet 60,509) Wolcott-Rallison syndrome (Dixon-Salazar (2012) Sci Transl Med 4,138ra78) {Atorvastatin sensitivity} (Callegari (2012) PLoS Genet 8,e100
GFPT1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	144.4	99.9	97.6	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.3	100	99.7	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GGT1	MENDELIOME PRECONCEPTION SCREENING	12.9	19.8	17.2	?Glutathioninuria, 231950
GH1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	175.2	100	100	Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100 Kowarski syndrome, 262650
GHR	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	212.3	99.8	99.5	Growth hormone insensitivity, partial, 604271 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 {Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	116.4	95.3	94.7	Growth hormone deficiency, isolated, type IV, 618157
GHSR	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	206	99.9	98.4	Growth hormone deficiency, isolated partial, 615925
GIF	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	141.2	100	99.9	Intrinsic factor deficiency, 261000
GINS1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	124.3	96.6	83.2	Immunodeficiency 55, 617827
GIPC3	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	103.3	91.8	85.5	Deafness, autosomal recessive 15, 601869
GJA1	VISION DISORDERS	246.4	100	100	Atrioventricular septal defect 3, 600309

	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJA3	VISION DISORDERS MENDELIOME	164.7	100	99.5	Cataract 14, multiple types, 601885
GJA5	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME RITME	268.4	100	100	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	VISION DISORDERS MENDELIOME	153.6	100	100	Cataract 1, multiple types, 116200
GJB1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	229.8	100	99.8	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	205.1	100	100	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB3	SKIN DISORDERS HEARING IMPAIRMENT NEUROPATHIES MENDELIOME	308.9	100	100	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy, 0 Deafness, autosomal recessive, 0 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200
GJB4	SKIN DISORDERS MENDELIOME	369.9	100	100	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	185.4	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	MOVEMENT DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY	41.9	68.9	58.6	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206

	MENDELIOME PRECONCEPTION SCREENING				
GK	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	43.6	74	54.7	Glycerol kinase deficiency, 307030
GLA	CARDIO SKIN DISORDERS HEART PANEL NEUROPATHIES METABOLIC DISORDERS RENAL DISORDERS MENDELIOME	81.3	99.7	97.6	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	MOVEMENT DISORDERS SKIN DISORDERS HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	78.9	90.6	82.7	Glycine encephalopathy, 605899
GLDN	MENDELIOME	130.4	98.2	91.8	Lethal congenital contracture syndrome 11, 617194
GLE1	MENDELIOME PRECONCEPTION SCREENING	110.8	100	99.7	Arthrogryposis, lethal, with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLI1	MENDELIOME	129.7	100	99.9	Polydactyly, postaxial, type A8, 618123
GLI2	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	138.5	99.4	97.4	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	154.2	100	99.7	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GLIS2	CILIO	109	99.9	98.2	Nephronophthisis 7, 611498

	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
GLIS3	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	133.4	99.9	99.3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	SKIN DISORDERS MENDELIOME	66.8	97.8	86.4	Glomuvenous malformations, 138000
GLRA1	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.3	100	100	Hyperekplexia 1, 149400
GLRB	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	98.6	96.6	88.8	Hyperekplexia 2, 614619
GLRX5	IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	108.2	92.6	83.8	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	74.5	94.4	84.3	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	108.8	99.9	98.2	Glutamine deficiency, congenital, 610015
GLYCTK	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	202.6	100	99.5	D-glyceric aciduria, 220120
GM2A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.6	100	100	GM2-gangliosidosis, AB variant, 272750
GMNN	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	101.2	92.6	83.8	Meier-Gorlin syndrome 6, 616835
GMPPA	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY	136.8	100	99.9	Alacrima, achalasia, and mental retardation syndrome, 615510

	MENDELIOME PRECONCEPTION SCREENING				
GMPPB	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	228.5	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GMPS	METABOLIC DISORDERS	112.9	96.9	89.2	No OMIM phenotype Leukemia, acute myelogenous, 601626
GNA11	SKIN DISORDERS RENAL DISORDERS MENDELIOME	149.5	99.5	96.4	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNA14	SKIN DISORDERS	158.1	100	100	No OMIM phenotype
GNAI2	MENDELIOME	138.5	100	99.9	Pituitary ACTH-secreting adenoma, 0 Ventricular tachycardia, idiopathic, 192605
GNAI3	CRANIOFACIAL ANOMALIES MENDELIOME	110.6	99	92	Auriculocondylar syndrome 1, 602483
GNAL	MOVEMENT DISORDERS MENDELIOME	134.9	94.5	91.6	Dystonia 25, 615073
GNAO1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	167.8	93.8	93.8	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	SKIN DISORDERS MENDELIOME	82.6	81.1	69.5	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	141	98.5	95.9	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GNAS-AS1	MENDELIOME	123456	123456	123456	Pseudohypoparathyroidism, type IB, 603233
GNAT1	VISION DISORDERS MENDELIOME	153.9	100	100	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.7	99.9	99.1	Achromatopsia 4, 613856
GNB1	INTELLECTUAL DISABILITY	189	100	100	Leukemia, acute lymphoblastic, somatic, 613065

	MENDELIOME				Mental retardation, autosomal dominant 42, 616973
GNB3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	179	100	100	Night blindness, congenital stationary, type 1H, 617024 {Hypertension, essential, susceptibility to}, 145500
GNB4	NEUROPATHIES MENDELIOME	152.6	100	99.7	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.9	99.9	98.3	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	153.7	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GNMT	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.2	99.4	97	Glycine N-methyltransferase deficiency, 606664
GNPAT	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	133.6	99.4	96.4	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	167.7	98.3	97.4	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	151.6	96.1	89.7	Mucolipidosis III gamma, 252605
GNRH1	HH MENDELIOME	86.7	99.5	91.5	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	HH MENDELIOME PRECONCEPTION SCREENING	161.9	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY	107.9	96.9	92	Mucopolysaccharidosis type IIID, 252940

	MENDELIOME PRECONCEPTION SCREENING				
GORAB	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	176.3	99.7	97.8	Geroderma osteodysplasticum, 231070
GOSR2	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRECONCEPTION SCREENING	127.2	95.9	95	Epilepsy, progressive myoclonic 6, 614018
GOT1	METABOLIC DISORDERS MENDELIOME	127	100	99.5	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	METABOLIC DISORDERS	102.7	94.5	89.2	No OMIM phenotype
GP1BA	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	153	97	94.3	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	34.5	74.2	64.3	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	136.5	100	100	Bleeding disorder, platelet-type, 11, 614201
GP9	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.3	96.6	89.3	Bernard-Soulier syndrome, type C, 231200
GPAA1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130	96.4	95.2	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	85.1	98.6	92.6	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPC6	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	142	100	100	Omodysplasia 1, 258315
GPD1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.5	99.9	99.1	Hypertriglyceridemia, transient infantile, 614480

GPD1L	HEART PANEL METABOLIC DISORDERS MENDELIOME RITME	138.3	100	98.5	Brugada syndrome 2, 611777
GPHN	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	167.2	98.4	96.9	Molybdenum cofactor deficiency C, 615501
GPI	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	142.6	100	99.5	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	MENDELIOME PRECONCEPTION SCREENING	119.7	99.9	99.2	Hyperlipoproteinemia, type 1D, 615947
GPNMB	MENDELIOME PRECONCEPTION SCREENING	177.8	100	100	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	MENDELIOME	159	100	100	Pituitary adenoma 2, GH-secreting, 300943
GPR143	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS MENDELIOME	61.5	85.3	75.5	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR161	SHORT STATURE/SKELETAL DYSPLASIA	196.8	100	100	No OMIM phenotype Pituitary stalk interuption syndrome (Karaca (2015) J Clin Endocrinol Metab 100,E140)
GPR179	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	133.6	100	99.7	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	CRANIOFACIAL ANOMALIES MENDELIOME	186.4	96.5	90.2	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	MENDELIOME	89.9	98.8	94.1	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	MENDELIOME	87.3	100	99.5	?Deafness, X-linked 7, 301018
GPSM2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	112.9	99.8	97	Chudley-McCullough syndrome, 604213
GPT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	138	98.2	90.9	Mental retardation, autosomal recessive 49, 616281
GPX1	METABOLIC DISORDERS	46.2	94.9	83.2	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GPX4	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	119.2	85.2	76.6	Spondylometaphyseal dysplasia, Sedaghatian type, 250220

	PRECONCEPTION SCREENING				
GRAP	HEARING IMPAIRMENT	64.4	82.1	77.6	No OMIM phenotype early onset HI severe. Li et al. Www.pnas.org/cgi/doi/10.1073/pnas.1810951116
GREB1L	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	148.6	100	99.5	Renal hypodysplasia/aplasia 3, 617805
GREM1	HEREDITARY CANCER	95.1	100	100	No OMIM phenotype {Colorectal cancer, increased risk, association with}(Peters (2012) Hum Genet 131,217) Oligosyndactyly of the hands, Cenani-Linz-like (Dimitrov (2010) J Med Genet 47,569) Mixed polyposis syndrome (Jaeger (2012) Nat Genet 44,699)
GREM2	MENDELIOME	133.7	100	100	Tooth agenesis, selective, 9, 617275
GRHL2	VISION DISORDERS BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	140.7	100	99.9	Van der Woude syndrome 2, 606713
GRHPR	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	112.5	85.1	78.2	Hyperoxaluria, primary, type II, 260000
GRIA3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	98.2	99.3	94.7	Mental retardation, X-linked 94, 300699
GRIA4	INTELLECTUAL DISABILITY MENDELIOME	148.3	99.8	98.1	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	175.4	100	99.9	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	133.2	96	94.7	Mental retardation, autosomal recessive, 6, 611092
GRIN1	MOVEMENT DISORDERS EPILEPSY	150.7	100	99.5	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254

	INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS				Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	159.1	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	189.4	99.9	99.3	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	73.1	69.1	62.1	Epileptic encephalopathy, early infantile, 46, 617162
GRIP1	DSD INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.8	100	99.9	Fraser syndrome 3, 617667
GRK1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	126.4	100	99.9	Oguchi disease-2, 613411
GRM1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	185.8	100	99.9	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	151.7	93.3	86.7	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PARK PRECONCEPTION SCREENING	184.5	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	183.7	100	99.8	Deafness, autosomal recessive 25, 613285
GRXCR2	HEARING IMPAIRMENT MENDELIOME	114.9	100	100	?Deafness, autosomal recessive 101, 615837
GSC	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	85.4	86.9	74.5	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471

GSE1	INTELLECTUAL DISABILITY	101.5	99.9	98.8	No OMIM phenotype ?Autism (Sanders (2012) Nature 485,237)
GSN	VISION DISORDERS SKIN DISORDERS RENAL DISORDERS MENDELIOME	119.2	94.2	89	Amyloidosis, Finnish type, 105120
GSS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	104	100	99.8	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2E2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	83.5	96.5	91	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	113.6	100	99.1	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	156.8	96.5	94.7	Jaberi-Elahi syndrome, 617988
GTPBP3	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	137.4	100	99.7	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	VISION DISORDERS MENDELIOME	160.6	100	100	Cone dystrophy-3, 602093 Cone-rod dystrophy 14, 602093
GUCA1B	VISION DISORDERS MENDELIOME	144.2	100	99.9	Retinitis pigmentosa 48, 613827
GUCY1A3	MENDELIOME PRECONCEPTION SCREENING	171.3	99.4	98	Moyamoya 6 with achalasia, 615750
GUCY2C	MENDELIOME PRECONCEPTION SCREENING	135.2	100	99.7	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	91.3	98.3	91.1	?Choroidal dystrophy, central areolar 1, 215500 Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000
GUF1	MENDELIOME	70.5	98.7	92.3	?Epileptic encephalopathy, early infantile, 40, 617065
GULOP	MENDELIOME	123456	123456	123456	Scurvy, 0
GUSB	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY	116.1	92.2	89.4	Mucopolysaccharidosis VII, 253220

	MENDELIOME PRECONCEPTION SCREENING				
GYG1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	157.8	100	99.6	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	110.8	100	98.5	Glycogen storage disease 0, muscle, 611556
GYS2	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	150.2	98.5	93.9	Glycogen storage disease 0, liver, 240600
GZF1	MENDELIOME	229	100	100	Joint laxity, short stature, and myopia, 617662
H19	MENDELIOME	123456	123456	123456	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071
H6PD	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	169.5	99	99	Cortisone reductase deficiency 1, 604931
HAAO	MENDELIOME	100.8	100	99.9	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HABP2	HEMOSTATIC/THROMBOTIC DISORDERS HEREDITARY CANCER	136.9	100	99.6	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HACE1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.3	99.2	95	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.8	98	95.1	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HADHA	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	84.4	96.5	90.3	Fatty liver, acute, of pregnancy, 609016 HELLP syndrome, maternal, of pregnancy, 609016 LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015
HADHB	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	80.5	92.5	79.5	Trifunctional protein deficiency, 609015
HAGH	METABOLIC DISORDERS	130.5	99.8	97.8	[Glyoxalase II deficiency], 614033
HAMP	IRON DISORDERS	175.6	100	100	Hemochromatosis, type 2B, 613313

	MENDELIOME PRECONCEPTION SCREENING				
HAND1	CONGENITAL HEART DISEASE HEART PANEL	84.8	100	98.9	No OMIM phenotype Ventricular septal defect (Cheng (2011) Clin Chim Acta) Cardiac malformations (Reamon-Buettner (2009) Hum Mol Genet 18,3567) Cardiomyopathy, dilated (Zhou (2015) Clin Chem Lab Med Epub, epub)
HAND2	CONGENITAL HEART DISEASE HEART PANEL	32.2	87.8	67.9	No OMIM phenotype Tetralogy of Fallot (Topf (2014) PLoS One 9,e95453) Ventricular septal defect (Sun (2016) G3 (Bethesda) epub,epub) ?Congenital heart disease (Shen (2010) Chin Med J (Engl) 123,1623)
HARS	VISION DISORDERS HEARING IMPAIRMENT NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	159.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS	169.7	99.9	99.2	?Perrault syndrome 2, 614926
HAX1	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	MENDELIOME	141.9	100	100	Erythrocytosis, 7, 617981 Heinz body anemias, alpha-, 140700 Hemoglobin H disease, nondeletional, 613978 Methemoglobinemia, alpha type, 617973 Thalassemias, alpha-, 604131
HBA2	MENDELIOME	133.7	97.8	91.1	Erythrocytosis 7, 617981 Heinz body anemia, 140700 Hemoglobin H disease, deletional and nondeletional, 613978 Thalassemia, alpha-, 604131
HBB	MENDELIOME PRECONCEPTION SCREENING	176.7	100	100	Delta-beta thalassemia, 141749 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Hereditary persistence of fetal hemoglobin, 141749 Methmoglobinemia, beta type, 617971 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Thalassemia-beta, dominant inclusion-body, 603902

					{Malaria, resistance to}, 611162
HBD	MENDELIOME	242.4	100	100	Thalassemia due to Hb Lepore, 0 Thalassemia, delta-, 0
HBG1	MENDELIOME	174.4	98.3	94.7	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	MENDELIOME	329.4	100	100	Cyanosis, transient neonatal, 613977 Fetal hemoglobin quantitative trait locus 1, 141749
HCCS	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	106.6	99.9	99.2	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	105.8	99.3	96.1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	122.4	99.9	97.8	Epileptic encephalopathy, early infantile, 24, 615871
HCN2	HEART PANEL	53.2	58.3	50.7	No OMIM phenotype
HCN3	HEART PANEL	146.4	99.9	99.2	No OMIM phenotype
HCN4	HEART PANEL MENDELIOME RITME	79.4	98.3	91.8	Brugada syndrome 8, 613123 Sick sinus syndrome 2, 163800
HCRT	MENDELIOME	54.2	77.4	66.8	?Narcolepsy 1, 161400
HDAC4	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY	111.9	99.9	99.3	No OMIM phenotype Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706) Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219) ?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HDAC6	INTELLECTUAL DISABILITY MENDELIOME	118.7	99.7	97	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	131.9	100	99.8	Cornelia de Lange syndrome 5, 300882
HECW2	INTELLECTUAL DISABILITY MENDELIOME	133.1	99.9	98.6	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	93.7	94.1	86.8	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	INTELLECTUAL DISABILITY MENDELIOME	142.3	81.4	76.1	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or

	PRECONCEPTION SCREENING				without mental retardation, 613926
HEPH	IRON DISORDERS	86.4	98.9	93.7	No OMIM phenotype ?anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004)).
HERC1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	173.7	99.9	99.4	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.4	80.9	77.9	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	29.1	64.9	42.6	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	HH SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	57.6	99.2	92.6	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HEXA	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	118.3	93.8	92.2	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.7	99.4	94	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HEY2	CONGENITAL HEART DISEASE HEART PANEL	146.1	99.2	92.8	No OMIM phenotype Congenital heart defects and cognitive impairment (Jordan (2015) Am J Med Genet A 167,2145)
HFE	HEART PANEL IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	142	100	99.7	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	HEART PANEL IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	116.8	100	100	Hemochromatosis, type 2A, 602390
HFM1	MENDELIOME	42.8	90.3	76.7	Premature ovarian failure 9, 615724

	PRECONCEPTION SCREENING				
HGD	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.8	100	99.8	Alkaptonuria, 203500
HGF	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	146.9	99.4	96.9	Deafness, autosomal recessive 39, 608265
HGSNAT	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101	86.4	85.7	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544
HIBADH	METABOLIC DISORDERS	105.2	92.1	91	No OMIM phenotype
HIBCH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	67.7	92.7	69.5	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	MENDELIOME PRECONCEPTION SCREENING	61.7	90.9	79.7	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	60	98.5	88	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIST1H1E	INTELLECTUAL DISABILITY MENDELIOME	60.9	99.9	94.7	Rahman syndrome, 617537
HIST1H4C	INTELLECTUAL DISABILITY	108.2	100	100	No OMIM phenotype
HIVEP2	INTELLECTUAL DISABILITY MENDELIOME	191.2	100	100	Mental retardation, autosomal dominant 43, 616977
HK1	VISION DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	143.4	100	99.9	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HLCS	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	172.8	100	100	Holocarboxylase synthetase deficiency, 253270

HMBS	SKIN DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME	109	100	99.8	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGA2	SHORT STATURE/SKELETAL DYSPLASIA	78.8	84	76.5	Leiomyoma, uterine, somatic, 150699
HMGB3	SKIN DISORDERS MENDELIOME	38	88.1	68.6	?Microphthalmia, syndromic 13, 300915
HMGCL	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.3	100	99.9	HMG-CoA lyase deficiency, 246450
HMGCS2	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.5	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	IRON DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	128.7	95.8	89.5	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	23	56.8	42	Oculoauricular syndrome, 612109
HNF1A	MENDELIOME HEREDITARY CANCER	156.7	100	99.4	Diabetes mellitus, insulin-dependent, 20, 612520 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700 {Diabetes mellitus, insulin-dependent}, 222100 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HNF1B	RENAL DISORDERS MENDELIOME	123.7	99.9	98.9	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	RENAL DISORDERS MENDELIOME	140.2	99.9	99.1	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HNMT	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135	100	99.7	Mental retardation, autosomal recessive 51, 616739 {Asthma, susceptibility to}, 600807
HNRNPA1	MENDELIOME	72.7	97.4	87.2	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426

HNRNPA2B1	MENDELIOME	134.7	98.3	96.9	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPDL	MENDELIOME	59	90.6	79.8	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH2	INTELLECTUAL DISABILITY MENDELIOME	170.8	100	100	Mental retardation, X-linked, syndromic, Bain type, 300986
HNRNPK	INTELLECTUAL DISABILITY MENDELIOME	71.7	86.9	78.4	Au-Kline syndrome, 616580
HNRNPU	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	126.6	99.3	97.9	Epileptic encephalopathy, early infantile, 54, 617391
HOGA1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.5	99.8	98.1	Hyperoxaluria, primary, type III, 613616
HOMER2	HEARING IMPAIRMENT MENDELIOME	133.5	99.5	99.4	?Deafness, autosomal dominant 68, 616707
HOOK1		76.7	95.6	87	
HOXA1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	165.3	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA11	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	86.3	88	78.5	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	DSD SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	49	69.2	61.7	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HOXA2	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	74.1	99.5	95.6	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HOXB1	MENDELIOME PRECONCEPTION SCREENING	104.5	100	100	Facial paresis, hereditary congenital, 3, 614744
HOXB13	HEREDITARY CANCER	127.8	99.9	97.7	{Prostate cancer, hereditary, 9}, 610997
HOXC13	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	104.9	97.5	91.1	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	NEUROPATHIES MENDELIOME	137.7	100	99.8	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HOXD13	MENDELIOME	142.3	93.1	89.6	?Brachydactyly-syndactyly syndrome, 610713 Brachydactyly, type D, 113200 Brachydactyly, type E, 113300 Syndactyly, type V, 186300

					Synpolydactyly 1, 186000
HPCA	MENDELIOME PRECONCEPTION SCREENING	283.4	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	137.8	100	100	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPGD	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	88	100	98.5	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPRT1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	58.2	96	84.8	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HPS1	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.8	100	99.3	Hermansky-Pudlak syndrome 1, 203300
HPS3	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.2	99.6	96.4	Hermansky-Pudlak syndrome 3, 614072
HPS4	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	141.9	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	133	99.9	98.7	Hermansky-Pudlak syndrome 5, 614074
HPS6	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.1	91	84.3	Hermansky-Pudlak syndrome 6, 614075

HPSE2	MENDELIOME PRECONCEPTION SCREENING	110.2	98.8	95.4	Urofacial syndrome 1, 236730
HR	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	94.9	97.3	94.2	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	VISION DISORDERS SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	164.7	99.8	98.1	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HRG	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	178.3	95	94.3	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
HS6ST1	HH METABOLIC DISORDERS	75.3	94.8	85	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSCB	IRON DISORDERS	90.1	99.1	95.1	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
HSD11B1	METABOLIC DISORDERS MENDELIOME	147.1	100	99.9	Cortisone reductase deficiency 2, 614662
HSD11B2	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	165.2	85.7	82.5	Apparent mineralocorticoid excess, 218030
HSD17B10	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	117.1	100	99.2	HSD10 mitochondrial disease, 300438
HSD17B3	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	156.4	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	95.1	93.9	90.8	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	DSD	189.2	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2

	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				deficiency, 201810
HSD3B7	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	136.3	98.2	91	Bile acid synthesis defect, congenital, 1, 607765
HSF4	VISION DISORDERS MENDELIOME	103	97.6	94.9	Cataract 5, multiple types, 116800
HSPA9	IRON DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	91.6	91.1	85.9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPB1	NEUROPATHIES MENDELIOME	39.7	93.7	81.8	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	NEUROPATHIES MENDELIOME	290.9	100	100	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB6	HEART PANEL	63.1	89.4	77.4	No OMIM phenotype
HSPB8	NEUROPATHIES MENDELIOME	156.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
HSPD1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	96.5	98.3	93.2	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	121.3	99.4	98.2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	MENDELIOME	220.4	100	100	Periodic fever, menstrual cycle dependent, 614674
HTRA1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	98.2	84.5	81.4	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149
HTRA2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	122.1	100	99.7	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297

	MITOCHONDRIAL DISORDERS				
HTT	MENDELIOME	143.7	98.5	97.1	Huntington disease, 143100 Lopes-Maciel-Rodan syndrome, 617435
HUWE1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	98.4	99.2	97	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.3	100	100	?Mucopolysaccharidosis type IX, 601492
HYAL2	CRANIOFACIAL ANOMALIES	223.2	100	100	No OMIM phenotype Orofacial clefting (Muggenthaler (2017) PLoS Genet 13,e1006470) ?Hypertelorism and high myopia (Shaheen (2016) Genet Med 18,686)
HYDIN	CILIO MENDELIOME PRECONCEPTION SCREENING	133.6	99.9	99.5	Ciliary dyskinesia, primary, 5, 608647
HYLS1	CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	171.1	100	100	Hydrolethalus syndrome, 236680
HYOU1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	134	99.9	99.7	?Immunodeficiency 59 and hypoglycemia, 233600
IARS	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	148.8	99.8	98.6	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093
IARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	131.5	100	99.9	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	113.3	93.3	89.5	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICK	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	131.6	99.9	98.2	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
ICOS	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	160.2	100	100	Immunodeficiency, common variable, 1, 607594
IDH1	SHORT STATURE/SKELETAL DYSPLASIA	97.9	92.5	81.7	{Glioma, susceptibility to, somatic}, 137800
IDH2	EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA	103.5	99.6	96.9	D-2-hydroxyglutaric aciduria 2, 613657

	METABOLIC DISORDERS MENDELIOME				
IDH3B	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	165.5	95.9	95.4	Retinitis pigmentosa 46, 612572
IDS	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	111.3	99.6	98.3	Mucopolysaccharidosis II, 309900
IDUA	SKIN DISORDERS HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123	88.1	80	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IER3IP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	73	93.2	82.2	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	113.5	99.6	97.1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	63.4	99.4	94.9	Osteogenesis imperfecta, type V, 610967
IFNAR2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	138.8	98.5	95.4	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNG	RENAL DISORDERS	131	100	99.8	{AIDS, rapid progression to}, 609423 {Aplastic anemia}, 609135 {Hepatitis C virus, response to therapy of}, 609532 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Tuberculosis, protection against}, 607948
IFNGR1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	138.5	99.2	97.3	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948

IFNGR2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	142.3	93.2	93.1	Immunodeficiency 28, mycobacteriosis, 614889
IFNLR1	HEARING IMPAIRMENT	90.6	97.6	96.5	No OMIM phenotype http://jmg.bmj.com/content/early/2018/02/16/jmedgenet-2017-104954
IFRD1	NEUROPATHIES	133.9	98.7	96	No OMIM phenotype (Lin et al.J. Hum. Genet. 2018Echeveste et al. Parkinsonism Relat Disord. 2017Brkanac et al.Am J Hum Genet. 2009)
IFT122	CRANIOFACIAL ANOMALIES CILIO SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT140	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	114.7	99.9	99	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME	131.7	100	99.6	?Bardet-Biedl syndrome 19, 615996
IFT43	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIO SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	CILIO	123.6	100	99.3	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102

	MENDELIOME				
IFT57	MENDELIOME	109.9	99	94	?Orofaciodigital syndrome XVIII, 617927
IFT74	VISION DISORDERS MENDELIOME	80.1	97.8	88.2	?Bardet-Biedl syndrome 20, 617119
IFT80	CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	57.8	87.6	70.7	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	VISION DISORDERS CILIO INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	92.9	88.3	81.2	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IFT88	CRANIOFACIAL ANOMALIES	78.1	98.1	90.7	No OMIM phenotype ?Cleft lip and palate (Tian (2017) Hum Mol Genet 26,860)
IGBP1	INTELLECTUAL DISABILITY MENDELIOME	118.3	99.8	97.7	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	122.5	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	144	100	99.8	Insulin-like growth factor I, resistance to, 270450
IGF2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	100	100	100	?Growth restriction, severe, with distinctive facies, 616489
IGF2R	MENDELIOME	139.5	98.3	96.7	Hepatocellular carcinoma, somatic, 114550
IGFALS	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	79.7	99.9	96.8	Acid-labile subunit, deficiency of, 615961
IGFBP7	MENDELIOME PRECONCEPTION SCREENING	71.6	91.7	83.5	Retinal arterial macro with supravalvular pulmonic stenosis, 614224
IGHG2	MENDELIOME	34.5	79.6	60	IgG2 deficiency, selective, 0
IGHM	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	185.2	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	107.8	99.3	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320

IGKC	MENDELIOME PRECONCEPTION SCREENING	157.9	100	100	Kappa light chain deficiency, 614102
IGLL1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	86.2	99.3	94.9	Agammaglobulinemia 2, 613500
IGSF1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	84.8	99.5	96.6	Hypothyroidism, central, and testicular enlargement, 300888
IGSF10	HH	246.8	100	99.9	No OMIM phenotype Delayed puberty,Howard et al. EMBO Mol Med. 2016 Jun 1 8(6):626-42
IGSF3	MENDELIOME	115.9	96.7	95	?Lacrimal duct defect, 149700
IHH	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	129.3	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	123.5	98.5	94.2	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	52.5	84.6	73.2	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	183.4	100	100	Immunodeficiency, common variable, 13, 616873
IL10	PRIMARY IMMUNODEFICIENCIES	125.3	100	99.9	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300
IL10RA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	141.9	100	99.9	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	168.8	98.7	96.1	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL11RA	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	139.9	100	99.5	Craniosynostosis and dental anomalies, 614188
IL12B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	121.1	100	99.9	Immunodeficiency 29, mycobacteriosis, 614890

IL12RB1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	124.3	97	94.7	Immunodeficiency 30, 614891
IL17F	PRIMARY IMMUNODEFICIENCIES MENDELIOME	85.8	99.1	94.4	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	140.5	99.8	96.9	Immunodeficiency 51, 613953
IL17RC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	96.1	99.8	99	Candidiasis, familial, 9, 616445
IL17RD	SKIN DISORDERS HH MENDELIOME	135.6	99.3	97.7	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1B		149.8	100	100	{Gastric cancer risk after H. pylori infection}, 137215
IL1RAPL1	INTELLECTUAL DISABILITY MENDELIOME	116.8	99.7	98	Mental retardation, X-linked 21/34, 300143
IL1RN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	162.8	100	100	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL2	PRIMARY IMMUNODEFICIENCIES	65.3	93.4	76.9	No OMIM phenotype Severe combined immunodeficiency due to IL2 deficiency,Combined T-cell and B-cell immunodeficiencies
IL21	PRIMARY IMMUNODEFICIENCIES MENDELIOME	94.7	97.8	87.5	?Immunodeficiency, common variable, 11, 615767
IL21R	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	128.5	100	100	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050
IL2RA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	116.4	100	99.5	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME SCID	65.2	99.8	97.3	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	SKIN DISORDERS MENDELIOME	124.2	99.9	99.6	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	99	100	100	Psoriasis 14, pustular, 614204

	PRECONCEPTION SCREENING				
IL6R		133.4	96.4	92.5	[Interleukin 6, serum level of, QTL], 614752 [Interleukin-6 receptor, soluble, serum level of, QTL], 614689
IL7R	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	129.5	99.9	99.4	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	107.4	100	99.9	Deafness, autosomal recessive 42, 609646
ILK	HEART PANEL	171.6	100	100	No OMIM phenotype Cardiomyopathy, dilated (Knoll (2007) Circulation 116,515) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (2015) Kidney Int 89, 476)
IMPA1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	71.4	97.1	85.8	Mental retardation, autosomal recessive 59, 617323
IMPAD1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.2	99.9	99.4	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPDH1	VISION DISORDERS METABOLIC DISORDERS MENDELIOME	61.3	87.8	83.5	Leber congenital amaurosis 11, 613837 Retinitis pigmentosa 10, 180105
IMPG1	VISION DISORDERS MENDELIOME	103.4	99.6	97.3	Macular dystrophy, vitelliform, 4, 616151
IMPG2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	154.3	99.5	97.8	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INF2	NEUROPATHIES RENAL DISORDERS MENDELIOME	79.2	84.1	81.1	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
ING1	MENDELIOME	127.8	99.5	95.4	Squamous cell carcinoma, head and neck, somatic, 275355
INO80	PRIMARY IMMUNODEFICIENCIES	105.1	99.2	97.1	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
INPP5E	VISION DISORDERS CILIO METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156

	PRECONCEPTION SCREENING				
INPP5K	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	108.3	100	99.6	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.6	96.7	93.7	Opsismodysplasia, 258480
INS	MENDELIOME	87.7	100	99.7	Diabetes mellitus, insulin-dependent, 2, 125852 Diabetes mellitus, permanent neonatal, 606176 Hyperproinsulinemia, 616214 Maturity-onset diabetes of the young, type 10, 613370
INSL3	MENDELIOME	67.5	81	77.9	Cryptorchidism, 219050
INSR	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	141.1	97.1	94.5	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
INTU	CRANIOFACIAL ANOMALIES CILIO RENAL DISORDERS MENDELIOME	122	99.7	96.6	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	160.5	100	100	Nephronophthisis 2, infantile, 602088
IPMK	HEREDITARY CANCER	95.5	96.9	83.9	No OMIM phenotype Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)
IQCB1	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	92.2	89.3	75.4	Senior-Loken syndrome 5, 609254
IQCE	MENDELIOME	132.2	98.8	96.6	?Polydactyly, postaxial, type A7, 617642
IQSEC2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	61.2	92.1	82.5	Mental retardation, X-linked 1/78, 309530
IRAK1	PRIMARY IMMUNODEFICIENCIES	59.7	92.8	80.4	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
IRAK4	PRIMARY IMMUNODEFICIENCIES	95.4	98.3	90.1	Invasive pneumococcal disease, recurrent isolated, 1, 610799

	MENDELIOME PRECONCEPTION SCREENING				IRAK4 deficiency, 607676
IRF1	MENDELIOME	185.4	100	100	Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic, 0 Myelogenous leukemia, acute, 0 Nonsmall cell lung cancer, somatic, 211980
IRF2BP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	63.2	88.2	70.8	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	INTELLECTUAL DISABILITY MENDELIOME	112.1	95.8	87.4	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF3	PRIMARY IMMUNODEFICIENCIES	116.5	99.9	99.1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	SKIN DISORDERS	182.4	100	99.9	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF6	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	113.7	99.9	97.9	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
IRF7	PRIMARY IMMUNODEFICIENCIES MENDELIOME	89.8	99.9	99.1	?Immunodeficiency 39, 616345
IRF8	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	114.7	99.6	97.4	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRX1	VISION DISORDERS	105	83.9	81.1	No OMIM phenotype ?Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123,9)
IRX2		41.4	79.7	64.3	
IRX3	HEART PANEL	73.8	82.9	66.2	No OMIM phenotype
IRX4	CONGENITAL HEART DISEASE HEART PANEL	90	95.8	92.3	No OMIM phenotype Congenital heart defect (Cheng (2014) BMC Genomics 15,1127) {Prostate cancer,susceptibility to} (Nguyen (2012) Hum Mol Genet 21,2076)
IRX5	MENDELIOME PRECONCEPTION SCREENING	74.7	94.2	86.1	Hamamy syndrome, 611174
ISCA1	MENDELIOME MITOCHONDRIAL DISORDERS	50.8	92.5	80.5	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	92	99.7	96.9	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	111.2	100	99.7	Myopathy with lactic acidosis, hereditary, 255125
ISG15	PRIMARY IMMUNODEFICIENCIES	160.1	100	100	Immunodeficiency 38, 616126

	MENDELIOME PRECONCEPTION SCREENING				
ISPD	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	104.4	95.2	84.8	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
ITCH	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	124.7	95.4	94.8	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2	HEMOSTATIC/THROMBOTIC DISORDERS	136.5	97.7	95.4	?Glycoprotein Ia deficiency, 614200
ITGA2B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	107.2	99.6	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	141.5	99.8	98.3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	146.5	99.8	99	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	129.6	99.6	97.6	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	122.4	99.6	98.1	Renal hypodysplasia/aplasia 1, 191830
ITGB2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	152.1	100	99.8	Leukocyte adhesion deficiency, 116920
ITGB3	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	142.2	99.3	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
ITGB4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	150.1	97.4	94.8	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730

ITGB6	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	137	96.5	95.2	Amelogenesis imperfecta, type IH, 616221
ITK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	125.2	100	99.6	Lymphoproliferative syndrome 1, 613011
ITM2B	MENDELIOME	110	99.7	97.9	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPA	EPILEPSY HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	161.4	100	99.9	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
ITPR2	MENDELIOME	137.3	99	95.9	?Anhidrosis, isolated, with normal sweat glands, 106190
IVD	BONE MARROW FAILURE METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.9	100	100	Isovaleric acidemia, 243500
IYD	MENDELIOME PRECONCEPTION SCREENING	117.3	99.7	97.8	Thyroid dyshormonogenesis 4, 274800
JAG1	VISION DISORDERS CONGENITAL HEART DISEASE HEART PANEL INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	148.4	98.1	97.5	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAGN1	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	PRIMARY IMMUNODEFICIENCIES	122.8	99.8	98.7	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)

JAK2	HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	90.6	95.9	94.1	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
JAK3	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	104.2	98.2	95.2	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM3	VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	158.6	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JMJD1C	INTELLECTUAL DISABILITY	144.5	99.8	97.7	No OMIM phenotype ?Rett syndrome (Saez (2016) Genet Med 18,378) ?Congenital heart disease in 22q11.2 deletion syndrome patients (Guo (2015) Am J Hum Genet 97,869) ?Autism spectrum disorder (Saez (2016) Genet Med 18,378) ?Intellectual disabilit
JPH1	MENDELIOME	173.9	100	99.8	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	CARDIO HEART PANEL MENDELIOME	87.5	90.4	75.1	Cardiomyopathy, hypertrophic, 17, 613873
JPH3	MENDELIOME	156.6	100	99.7	Huntington disease-like 2, 606438
JUP	CARDIO SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	145.1	100	99.6	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KALRN	INTELLECTUAL DISABILITY PRECONCEPTION SCREENING	149	100	99.5	{Coronary heart disease, susceptibility to, 5}, 608901
KANK1	INTELLECTUAL DISABILITY MENDELIOME	166.2	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	SKIN DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	151.4	99.9	99.5	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	EPILEPSY INTELLECTUAL DISABILITY	172.3	99.9	99.2	Koolen-De Vries syndrome, 610443

	MENDELIOME				
KARS	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	122.6	100	99.3	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KAT6A	INTELLECTUAL DISABILITY MENDELIOME	169.3	100	99.8	Mental retardation, autosomal dominant 32, 616268
KAT6B	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	192.3	99.6	98.5	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KATNB1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	141.7	100	100	Lissencephaly 6, with microcephaly, 616212
KBTBD13	MENDELIOME MUSCLE DISORDERS	107.1	99.8	96.8	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	MOVEMENT DISORDERS EPILEPSY MENDELIOME	167	100	99.7	Episodic ataxia/myokymia syndrome, 160120
KCNA2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	157.7	100	99.9	Epileptic encephalopathy, early infantile, 32, 616366
KCNA4	INTELLECTUAL DISABILITY	153.1	100	100	No OMIM phenotype Abnormal striatum, congenital cataract and intellectual disability (Kaya (2016) J Med Genet 53,786)
KCNA5	HEART PANEL MENDELIOME RITME	143.3	99.4	96	Atrial fibrillation, familial, 7, 612240
KCNB1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	145	100	99.9	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	199.2	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	MOVEMENT DISORDERS INTELLECTUAL DISABILITY	144	68.5	59	Spinocerebellar ataxia 13, 605259

	MENDELIOME				
KCND2	HEART PANEL	183.4	100	100	No OMIM phenotype Autism and epilepsy (Lee (2014) Hum Mol Genet 23,3481) J-wave syndrome with sudden cardiac death (Perrin (2014) Circ Cardiovasc Genet 7,782) Epilepsy,temporal lobe (Singh (2006) Neurobiol Dis 24,245)
KCND3	MOVEMENT DISORDERS HEART PANEL MENDELIOME RITME	182.5	99.9	99.1	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNE1	HEARING IMPAIRMENT HEART PANEL MENDELIOME PRECONCEPTION SCREENING RITME	462.6	100	100	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	HEART PANEL MENDELIOME RITME	181.5	100	100	Atrial fibrillation, familial, 4, 611493 Long QT syndrome 6, 613693
KCNE3	HEART PANEL MENDELIOME RITME	177.1	100	100	?Brugada syndrome 6, 613119
KCNE4	HEART PANEL	84.8	79.9	77.6	No OMIM phenotype ?Periodic paralysis (Silva (2004) Arq Bras Endocrinol Metabol 48,196) {Atrial fibrillation, association with} (Zeng (2007) Cardiology 108,97)
KCNE5	HEART PANEL RITME	88	97.7	90.3	No OMIM phenotype Atrial fibrillation (Ravn (2008) Heart Rhythm 5,427 Idiopathic ventricular fibrillation (Ohno (2011) Circ Arrhythm Electrophysiol 4,352) Atrial fibrillation,lone,early-onset (Olesen (2014) Heart Rhythm 11,246)
KCNH1	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	185.8	98.7	98.7	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNH2	HEART PANEL MENDELIOME RITME	102.6	92.3	84.8	Long QT syndrome 2, 613688 Short QT syndrome 1, 609620 {Long QT syndrome 2, acquired, susceptibility to}, 613688
KCNJ1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	233.9	100	100	Bartter syndrome, type 2, 241200
KCNJ10	MOVEMENT DISORDERS HEARING IMPAIRMENT EPILEPSY	213.4	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780

	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
KCNJ11	EPILEPSY HEART PANEL INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	299.5	100	100	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNJ13	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	210.4	100	99.9	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNJ2	HEART PANEL MENDELIOME RITME MUSCLE DISORDERS	229.3	100	100	Andersen syndrome, 170390 Atrial fibrillation, familial, 9, 613980 Short QT syndrome 3, 609622
KCNJ5	HEART PANEL RENAL DISORDERS MENDELIOME RITME	193.9	100	99.8	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KCNJ6	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	156.6	100	99.9	Keppen-Lubinsky syndrome, 614098
KCNJ8	HEART PANEL RITME	177.1	100	100	No OMIM phenotype Cantu syndrome (Brownstein (2013) Eur J Med Genet 56,678) Sudden infant death syndrome (Klaver (2011) Int J Cardiol 152,162) ?Ventricular fibrillation (Haissaguerre (2009) J Cardiovasc Electrophysiol 20,93)
KCNK3	HEART PANEL MENDELIOME	165.3	98.9	96.1	Pulmonary hypertension, primary, 4, 615344
KCNK9	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	193.7	100	100	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNMA1	MOVEMENT DISORDERS EPILEPSY MENDELIOME	120.4	94.4	93.2	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNN3	HEART PANEL	151.1	100	99.9	No OMIM phenotype
KCNN4	MENDELIOME	136.1	100	99.8	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	HEARING IMPAIRMENT HEART PANEL MENDELIOME PRECONCEPTION SCREENING	114.7	93	90.3	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 Short QT syndrome 2, 609621

	RITME				{Long QT syndrome 1, acquired, susceptibility to}, 192500
KCNQ1OT1	MENDELIOME	123456	123456	123456	Beckwith-Wiedemann syndrome, 130650
KCNQ2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	85.4	90.1	86.5	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	110.7	98.9	95.5	Seizures, benign neonatal, 2, 121201
KCNQ4	HEARING IMPAIRMENT MENDELIOME	135.2	93.7	89.1	Deafness, autosomal dominant 2A, 600101
KCNQ5	INTELLECTUAL DISABILITY MENDELIOME	158	96.2	94.2	Mental retardation, autosomal dominant 46, 617601
KCNT1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	112	95.3	92.3	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCNT2	MENDELIOME	105.1	97.6	89.9	?Epileptic encephalopathy, early infantile, 57, 617771
KCNV2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.7	100	100	Retinal cone dystrophy 3B, 610356
KCTD1	MENDELIOME	140.5	100	100	Scalp-ear-nipple syndrome, 181270
KCTD17	MENDELIOME	93.7	95.3	88	Dystonia 26, myoclonic, 616398
KCTD3	CILIO	137.8	99.7	97.9	Joubert syndrome
KCTD7	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	166.7	95	95	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDF1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	103.9	100	99.6	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	129.7	96.2	93.4	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM5B	INTELLECTUAL DISABILITY MENDELIOME	138	98.8	96.9	Mental retardation, autosomal recessive 65, 618109
KDM5C	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	112.6	97.9	95.1	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY	109	93.2	84.3	Kabuki syndrome 2, 300867

	MENDELIOME				
KDR	MENDELIOME	137.9	100	99.4	Hemangioma, capillary infantile, somatic, 602089 {Hemangioma, capillary infantile, susceptibility to}, 602089
KDSR	SKIN DISORDERS MENDELIOME	173.7	100	100	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	191.6	100	100	Cornea plana 2, autosomal recessive, 217300
KHDC3L	MENDELIOME PRECONCEPTION SCREENING	110.3	99.9	99.2	Hydatidiform mole, recurrent, 2, 614293
KIAA0556	CILIO RENAL DISORDERS MENDELIOME	134.4	99.9	99.4	Joubert syndrome 26, 616784
KIAA0586	CILIO INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.7	98.2	92.7	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	123.9	99.9	98.7	?Orofaciodigital syndrome XV, 617127
KIAA1109	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	144.1	99.1	97.2	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	VISION DISORDERS	148.6	97.1	96	No OMIM phenotype Retinitis Pigmentosa
KIDINS220	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	155.4	99.9	99.5	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF11	VISION DISORDERS SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	83.8	97.2	94.2	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	111.3	98.1	89.9	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF1A	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	114	99.2	96.1	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357

	PRECONCEPTION SCREENING				
KIF1B	NEUROPATHIES MENDELIOME HEREDITARY CANCER	154.8	100	99.5	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF1BP	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	159.7	96.2	96.1	Goldberg-Shprintzen megacolon syndrome, 609460
KIF1C	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.3	99.9	99.1	Spastic ataxia 2, autosomal recessive, 611302
KIF21A	VISION DISORDERS MENDELIOME	123.5	99.4	96.1	Fibrosis of extraocular muscles, congenital, 1, 135700 Fibrosis of extraocular muscles, congenital, 3B, 135700
KIF22	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	163.1	100	99.9	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF23	IRON DISORDERS	170.1	96.2	94.5	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013)).
KIF2A	INTELLECTUAL DISABILITY MENDELIOME	105.2	97.7	88.7	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF4A	INTELLECTUAL DISABILITY MENDELIOME	92.4	98.2	93.7	?Mental retardation, X-linked 100, 300923
KIF5A	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	136.1	100	99.9	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KIF5C	INTELLECTUAL DISABILITY MENDELIOME	116.3	99.9	99.1	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	INTELLECTUAL DISABILITY	136.3	100	99.7	Mental retardation, autosomal dominant 4, 612581
KISS1	HH MENDELIOME	41.4	98.5	91.2	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	HH MENDELIOME PRECONCEPTION SCREENING	106.4	99.5	95.3	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
KIT	SKIN DISORDERS MENDELIOME	153	100	99.7	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, somatic, 273300

	HEREDITARY CANCER				Leukemia, acute myeloid, 601626 Mastocytosis, cutaneous, 154800 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME	81.8	97	91.6	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	174.2	98.3	96	Retinitis pigmentosa 69, 615780
KL	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	179.9	97.2	96	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLB	HH	213.4	100	100	No OMIM phenotype Xu et al. EMBO Mol Med. 2017 Oct 9(10):1379-1397
KLC2	MENDELIOME PRECONCEPTION SCREENING	115.9	99.8	98.2	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	BONE MARROW FAILURE IRON DISORDERS MENDELIOME	52.1	90.8	81.7	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
KLF10	HEART PANEL	141.4	100	99.7	No OMIM phenotype
KLF11	MENDELIOME	181.3	99.9	99.4	Maturity-onset diabetes of the young, type VII, 610508
KLF6	MENDELIOME	154	100	100	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLF7	INTELLECTUAL DISABILITY	144.3	100	99.5	No OMIM phenotype
KLHL10	MENDELIOME	209.7	100	100	Spermatogenic failure 11, 615081
KLHL15	INTELLECTUAL DISABILITY MENDELIOME	178.1	100	99.9	Mental retardation, X-linked 103, 300982
KLHL24	SKIN DISORDERS MENDELIOME	192.9	100	100	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLHL3	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	141.1	100	99.8	Pseudohypoaldosteronism, type IID, 614495
KLHL40	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	157.9	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	MENDELIOME PRECONCEPTION SCREENING	203.5	100	99.6	Nemaline myopathy 9, 615731

	MUSCLE DISORDERS				
KLHL7	VISION DISORDERS MENDELIOME	123.5	100	99.5	Cold-induced sweating syndrome 3, 617055 Retinitis pigmentosa 42, 612943
KLHL9	MUSCLE DISORDERS	260.8	100	100	No OMIM phenotype Myopathy, distal, early-onset (Cirak (2010) Brain 133, 2123)
KLK4	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	185.1	100	98.8	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	143.4	99.6	96.7	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	117.1	100	100	Cowden syndrome 4, 615107
KMT2A	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	152.5	99.3	98.6	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2B	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	120.3	94	91.2	Dystonia 28, childhood-onset, 617284
KMT2C	INTELLECTUAL DISABILITY MENDELIOME	170.2	91	88.6	Kleefstra syndrome 2, 617768
KMT2D	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	142.1	99.9	99	Kabuki syndrome 1, 147920
KMT5B	INTELLECTUAL DISABILITY MENDELIOME	202.3	100	99.6	Mental retardation, autosomal dominant 51, 617788
KNG1	HEMOSTATIC/THROMBOTIC DISORDERS	193.3	100	100	[High molecular weight kininogen deficiency], 228960 [Kininogen deficiency], 228960
KNL1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	113.7	98.3	95.2	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	EPILEPSY INTELLECTUAL DISABILITY	112.1	100	99.9	Mental retardation, autosomal recessive 41, 615637

	MENDELIOME PRECONCEPTION SCREENING				
KRAS	SKIN DISORDERS HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
KREMEN1	CRANIOFACIAL ANOMALIES MENDELIOME	149.1	94.7	93.6	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	MENDELIOME	85.1	99.9	97.7	Cavernous malformations of CNS and retina, 116860 Cerebral cavernous malformations-1, 116860 Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860
KRT1	SKIN DISORDERS MENDELIOME	120.1	99.9	96.7	Epidermolytic hyperkeratosis, 113800 Ichthyosis histrix, Curth-Macklin type, 146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962
KRT10	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	103	98.7	93.9	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	VISION DISORDERS MENDELIOME	126.1	98.6	95.4	Meesmann corneal dystrophy, 122100
KRT13	SKIN DISORDERS MENDELIOME	140.4	99.9	98.9	White sponge nevus 2, 615785
KRT14	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	59.3	89	82.1	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000
KRT16	SKIN DISORDERS MENDELIOME	38.5	72.4	53.4	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	SKIN DISORDERS	21.5	47.2	31.9	Pachyonychia congenita 2, 167210

	MENDELIOME				Steatocystoma multiplex, 184500
KRT18	MENDELIOME PRECONCEPTION SCREENING	40.2	84.8	67.8	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	SKIN DISORDERS MENDELIOME	140.9	100	99.8	Ichthyosis bullosa of Siemens, 146800
KRT25	MENDELIOME	142.9	100	100	Woolly hair, autosomal recessive 3, 616760
KRT3	VISION DISORDERS MENDELIOME	105.1	100	99.7	Meesmann corneal dystrophy, 122100
KRT4	SKIN DISORDERS MENDELIOME	130.7	100	99.9	White sponge nevus 1, 193900
KRT5	SKIN DISORDERS MENDELIOME	133.5	100	100	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960
KRT6A	SKIN DISORDERS MENDELIOME	197.8	94.8	87.9	Pachyonychia congenita 3, 615726
KRT6B	SKIN DISORDERS MENDELIOME	194.4	95.9	89.3	Pachyonychia congenita 4, 615728
KRT6C	SKIN DISORDERS MENDELIOME	174.5	87.3	79.7	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	SKIN DISORDERS MENDELIOME	139.8	100	100	?Hypotrichosis 13, 615896
KRT74	SKIN DISORDERS MENDELIOME	150.2	99.9	98.9	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT75	SKIN DISORDERS	135.3	100	100	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT8	MENDELIOME PRECONCEPTION SCREENING	39.2	91.7	73.4	Cirrhosis, cryptogenic, 215600 {Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT81	SKIN DISORDERS MENDELIOME	93.4	99.7	96	Monilethrix, 158000
KRT83	SKIN DISORDERS MENDELIOME	81.3	98.4	89.5	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	108.3	98.8	95.3	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	SKIN DISORDERS MENDELIOME	101.4	100	98	Monilethrix, 158000
KRT9	SKIN DISORDERS	84.8	98.2	96	Palmoplantar keratoderma, epidermolytic, 144200

	MENDELIOME				
KY	MENDELIOME	135.5	100	100	Myopathy, myofibrillar, 7, 617114
KYNU	MENDELIOME	116.9	98.5	92.2	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L1CAM	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	133.3	99.8	98.4	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
L2HGDH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.1	98.4	97	L-2-hydroxyglutaric aciduria, 236792
LACTB	MITOCHONDRIAL DISORDERS	117.2	93	80.8	No OMIM phenotype
LAGE3	RENAL DISORDERS MENDELIOME	50.3	95	81.2	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	MOVEMENT DISORDERS VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	137.5	100	99.6	Poretti-Boltshauser syndrome, 615960
LAMA2	HEART PANEL INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	143.5	99.9	99.5	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138
LAMA3	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.6	99.7	99.2	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMA4	CARDIO HEART PANEL MENDELIOME	132.6	100	99.9	Cardiomyopathy, dilated, 1JJ, 615235
LAMB1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	169.8	100	99.6	Lissencephaly 5, 615191
LAMB2	VISION DISORDERS RENAL DISORDERS MENDELIOME	201.7	100	99.8	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049

	PRECONCEPTION SCREENING				
LAMB3	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.4	100	99.6	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.6	99.9	98.8	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC3	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.5	98.3	96.1	Cortical malformations, occipital, 614115
LAMP2	CARDIO HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME RITME MUSCLE DISORDERS	106.1	92.7	91.2	Danon disease, 300257
LAMTOR2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	167	100	99.9	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	143	100	99.6	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	56.3	80.5	63.7	Alazami syndrome, 615071
LARS	MENDELIOME	133.5	99.5	97.1	?Infantile liver failure syndrome 1, 615438
LARS2	HEARING IMPAIRMENT IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	143	100	100	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300
LAS1L	INTELLECTUAL DISABILITY MENDELIOME	90.2	99.7	97.7	Wilson-Turner syndrome, 309585
LAT	PRIMARY IMMUNODEFICIENCIES MENDELIOME SCID	94.6	99.8	98.4	Immunodeficiency 52, 617514

LBR	CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	87.8	93.3	83.9	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	VISION DISORDERS CILIO MENDELIOME PRECONCEPTION SCREENING	127.8	97.3	95.7	Leber congenital amaurosis 5, 604537
LCAT	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	145.6	97.8	91.7	Fish-eye disease, 136120 Norum disease, 245900
LCK	PRIMARY IMMUNODEFICIENCIES MENDELIOME SCID	161.4	98.2	95.5	?Immunodeficiency 22, 615758
LCT	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	142	99.9	99.1	Lactase deficiency, congenital, 223000
LDB3	CARDIO HEART PANEL MENDELIOME MUSCLE DISORDERS	127.3	95.5	93.7	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Left ventricular noncompaction 3, 601493 Myopathy, myofibrillar, 4, 609452
LDHA	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	59.8	94.1	87.1	Glycogen storage disease XI, 612933
LDHB	METABOLIC DISORDERS	106.8	97.3	85.6	[Lactate dehydrogenase-B deficiency], 614128
LDLR	MENDELIOME	171	99.6	97.9	Hypercholesterolemia, familial, 143890 LDL cholesterol level QTL2, 143890
LDLRAP1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	156.1	95.7	91.2	Hypercholesterolemia, familial, autosomal recessive, 603813
LEF1	MENDELIOME	115.9	100	99.9	Sebaceous tumors, somatic, 0
LEFTY2	CONGENITAL HEART DISEASE HEART PANEL	42.3	91.3	77.1	Left-right axis malformations (Koas (1999) Am J Hum Genet 64, 712)
LEMD2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	68.6	89.1	80.6	Cataract 46, juvenile-onset, 212500
LEMD3	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA	96.7	95.4	88.8	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700

	MENDELIOME				
LEP	HH MENDELIOME PRECONCEPTION SCREENING	188.8	100	99.6	Obesity, morbid, due to leptin deficiency, 614962
LEPR	HH MENDELIOME PRECONCEPTION SCREENING	109.6	93.8	90.2	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME	91.6	85.4	83.3	?Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	EPILEPSY MENDELIOME	163.8	97.8	94.8	Epilepsy, familial temporal lobe, 1, 600512
LGI4	MENDELIOME	73.8	99	95.8	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LHB	HH MENDELIOME PRECONCEPTION SCREENING	29	97.2	73.2	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	DSD MENDELIOME PRECONCEPTION SCREENING	154.5	95.4	92.8	Leydig cell adenoma, somatic, with precocious puberty, 176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Luteinizing hormone resistance, female, 238320 Precocious puberty, male, 176410
LHFPL5	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	287.7	100	100	Deafness, autosomal recessive 67, 610265
LHX3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	84.7	94	80.6	Pituitary hormone deficiency, combined, 3, 221750
LHX4	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	144.9	100	99.8	Pituitary hormone deficiency, combined, 4, 262700
LIAS	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	133.7	99.5	97.1	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	123.4	97.2	92.1	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	PRIMARY IMMUNODEFICIENCIES	95.4	100	99	No OMIM phenotype

					DNA ligase I deficiency Combined T-cell and B-cell immunodeficiencies 1 studie Barnes DE et al.,1992
LIG4	BONE MARROW FAILURE DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING SCID HEREDITARY CANCER	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LIM2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	103.2	100	98.6	Cataract 19, multiple types, 615277
LIMS2	HEART PANEL MENDELIOME PRECONCEPTION SCREENING	110.8	93	92.3	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	INTELLECTUAL DISABILITY MENDELIOME	230.8	100	99.8	Mental retardation, autosomal recessive 64, 618103
LINS1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147.8	99.9	98	Mental retardation, autosomal recessive 27, 614340
LIPA	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.9	98.8	95.8	Cholesteryl ester storage disease, 278000 Wolman disease, 278000
LIPC	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.2	100	99.8	Hepatic lipase deficiency, 614025 [High density lipoprotein cholesterol level QTL 12], 612797 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	MENDELIOME PRECONCEPTION SCREENING	113.4	99.9	98.4	Lipodystrophy, familial partial, type 6, 615980
LIPH	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	140.6	100	100	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.2	99.2	95.8	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	227.4	100	100	Lipoyltransferase 1 deficiency, 616299

LIPT2	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	92.2	97.3	83.2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	NEUROPATHIES MENDELIOME	126.3	94.8	91.5	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	129.8	99.3	94.4	Combined factor V and VIII deficiency, 227300
LMAN2L	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	127.9	100	99.8	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	MENDELIOME PRECONCEPTION SCREENING	94	95.9	88.1	Acheiropody, 200500 Hypoplastic or aplastic tibia with polydactyly, 188740 Laurin-Sandrow syndrome, 135750 Polydactyly, preaxial type II, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	80.2	91.9	83	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	MENDELIOME PRECONCEPTION SCREENING	132.4	99.6	97.8	Lipase deficiency, combined, 246650
LMNA	CARDIO SKIN DISORDERS HEART PANEL NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING RITME MUSCLE DISORDERS	89.2	97.9	91.3	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMNB1	MOVEMENT DISORDERS MENDELIOME	123.3	99.9	99.1	Leukodystrophy, adult-onset, autosomal dominant, 169500
LMNB2	MENDELIOME	121.2	97.1	93.2	?Epilepsy, progressive myoclonic, 9, 616540 {Lipodystrophy, partial, acquired, susceptibility to}, 608709
LMOD1		177.4	100	100	No OMIM phenotype

	HEART PANEL				Megacystis-microcolon-intestinal hypoperistalsis syndrome (Halim (2017) Proc Natl Acad Sci USA 114)
LMOD3	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	141.5	99.9	98.5	Nemaline myopathy 10, 616165
LMX1A	HEARING IMPAIRMENT	109.2	100	99.9	No OMIM phenotype Wesdorp et al (submitted) nonsyndromic with vestibular involvement
LMX1B	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	111.4	97	92.3	Nail-patella syndrome, 161200
LNPK	MENDELIOME	84.7	92	84.1	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	141.5	97.9	96.4	CODAS syndrome, 600373
LOR	SKIN DISORDERS MENDELIOME	13.5	62.6	33.1	Vohwinkel syndrome with ichthyosis, 604117
LOX	HEART PANEL MENDELIOME	104.4	99.8	97.6	Aortic , familial thoracic 10, 617168
LOXHD1	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	136.5	100	99.6	Deafness, autosomal recessive 77, 613079
LPA		126.3	99	98.1	[LPA deficiency, congenital], 0 {Coronary artery disease, susceptibility to}, 0
LPAR6	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	104.2	99.7	98.2	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	134.6	99.8	97.8	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	SKIN DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCIES	111.5	100	99.6	Majeed syndrome, 609628

	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				
LPL	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.2	100	100	Combined hyperlipidemia, familial, 144250 Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 0
LPP	MENDELIOME	132.1	100	100	Leukemia, acute myeloid, 601626 Lipoma, 0
LRAT	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	298.3	100	100	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	134.4	99.3	97.8	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.5	99.4	97.5	Urofacial syndrome 2, 615112
LRIT3	VISION DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	142.4	94.4	94.1	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	142.1	97.2	95.6	Albinism, oculocutaneous, type VII, 615179
LRP1	MENDELIOME	196	99.7	99.1	?Keratosis pilaris atrophicans, 604093
LRP2	VISION DISORDERS CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	176.3	100	99.8	Donnai-Barrow syndrome, 222448
LRP4	SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	166.6	99.1	98.9	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	VISION DISORDERS HEARING IMPAIRMENT	189.8	98.2	97.9	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750

	SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LRP6	CRANIOFACIAL ANOMALIES MENDELIOME	169.3	100	99.7	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LRPAP1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.3	99.5	97.2	Myopia 23, autosomal recessive, 615431
LRPPRC	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	127.3	99.4	97.2	Leigh syndrome, French-Canadian type, 220111
LRRC10	HEART PANEL	193.6	100	100	No OMIM phenotype Cardiomyopathy,dilated (Qu (2015) Mol Med Rep 12,3718)
LRRC56	CILIO MENDELIOME	106.6	99	95.8	Ciliary dyskinesia, primary, 39, 618254
LRRC6	CILIO MENDELIOME PRECONCEPTION SCREENING	137.9	94.7	91	Ciliary dyskinesia, primary, 19, 614935
LRRC8A	PRIMARY IMMUNODEFICIENCIES MENDELIOME	273	100	99.9	?Agammaglobulinemia 5, 613506
LRRK1	SHORT STATURE/SKELETAL DYSPLASIA	154.1	98.9	97.1	No OMIM phenotype Osteosclerotic metaphyseal dysplasia (Iida (2016) J Med Genet 53,568) ?Parkinson disease (Schulte (2013) Neurogenetics epub,epub)
LRRK2	PARK	117	98.2	93.1	{Parkinson disease 8}, 607060
LRSAM1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	130.4	100	99.7	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	125.9	99.8	96.6	Deafness, autosomal recessive 63, 611451
LSS	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	127.8	100	99.1	Cataract 44, 616509
LTBP1		138.1	95.3	93.9	
LTBP2		104.6	99.6	97.1	?Weill-Marchesani syndrome 3, recessive, 614819

	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING				Glaucoma 3, primary congenital, D, 613086 Microspheropha and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	113.5	98.7	94.7	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.1	98.6	95	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	METABOLIC DISORDERS PRECONCEPTION SCREENING	54.3	71.8	61.4	Leukotriene C4 synthase deficiency, 614037
LYRM4	MENDELIOME MITOCHONDRIAL DISORDERS	60.1	63.2	54.3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	49	87.6	72.4	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	VISION DISORDERS SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	134.6	97.8	93.9	Chediak-Higashi syndrome, 214500
LYZ	SKIN DISORDERS RENAL DISORDERS MENDELIOME	165.3	100	100	Amyloidosis, renal, 105200
LZTFL1	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	109.1	99.1	95.3	Bardet-Biedl syndrome 17, 615994
LZTR1	HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA	134	100	99.4	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670

	INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER				
LZTS1	MENDELIOME	105.5	100	100	Esophageal squamous cell carcinoma, 133239
MAB21L1	INTELLECTUAL DISABILITY	170.4	100	100	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30487245
MAB21L2	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	245.6	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	INTELLECTUAL DISABILITY	150.3	99.8	99	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30471716
MAD1L1	MENDELIOME	97.8	99.8	97.1	Lymphoma, somatic, 0 Prostate cancer, somatic, 176807
MAD2L2	BONE MARROW FAILURE MENDELIOME	125.9	100	100	?Fanconi anemia, complementation group V, 617243
MAF	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	60.2	77.5	72.7	Ayme-Gripp syndrome, 601088 Cataract 21, multiple types, 610202
MAFA	MENDELIOME	28.6	79	59.6	Insulinomatosis and diabetes mellitus, 147630
MAFB	RENAL DISORDERS MENDELIOME	93.8	99.7	97.8	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	146.5	100	99.5	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	RENAL DISORDERS MENDELIOME	80	99.7	97.4	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	INTELLECTUAL DISABILITY MENDELIOME	83.4	90.6	81.2	Schaaf-Yang syndrome, 615547
MAGI2	RENAL DISORDERS MENDELIOME	98	91.8	88.3	Nephrotic syndrome, type 15, 617609
MAGT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	101.8	98.4	95.8	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAK	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.1	95.6	94.1	Retinitis pigmentosa 62, 614181
MAL	PRIMARY IMMUNODEFICIENCIES	138.6	100	99.9	No OMIM phenotype
MALT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	136.6	89.1	85.4	Immunodeficiency 12, 615468
MAML2	MENDELIOME	122.9	100	100	Mucoepidermoid salivary gland carcinoma, 0

MAMLD1	DSD MENDELIOME	131.4	99.9	98.3	Hypospadias 2, X-linked, 300758
MAN1B1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128.9	100	99.7	Mental retardation, autosomal recessive 15, 614202
MAN2B1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAOA	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	113.8	99.9	98.7	Brunner syndrome, 300615 {Antisocial behavior}, 300615
MAP1B	INTELLECTUAL DISABILITY	142	100	99.5	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30150678
MAP2K1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	92.3	99.8	95.6	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	107.9	97.6	89.2	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	DSD MENDELIOME	166	92.9	89.2	46XY sex reversal 6, 613762
MAP3K14	PRIMARY IMMUNODEFICIENCIES	111.3	99.3	98.4	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
MAP3K20	MENDELIOME	125.3	99.8	98	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	114.6	99.7	98.1	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137

MAP3K8	MENDELIOME	143.2	100	99.8	Lung cancer, somatic, 211980
MAPK8		143.8	99.9	99.1	
MAPK8IP3	INTELLECTUAL DISABILITY	150.2	100	99.8	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30612693
MAPKAPK3	VISION DISORDERS MENDELIOME	92.8	98.9	96.3	?Macular dystrophy, patterned, 3, 617111
MAPKBP1	CILIO RENAL DISORDERS MENDELIOME	144.1	100	100	Nephronophthisis 20, 617271
MAPRE2	INTELLECTUAL DISABILITY MENDELIOME	205.7	100	99.5	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	MENDELIOME PARK PRECONCEPTION SCREENING	127.2	100	98.6	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600
MARS	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	125.2	99.7	97.3	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	173.2	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	159.4	97.5	94	Deafness, autosomal recessive 49, 610153
MASP1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	148.6	100	99.6	3MC syndrome 1, 257920
MASP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	139.8	100	99.3	MASP2 deficiency, 613791
MASTL	HEMOSTATIC/THROMBOTIC DISORDERS	130.5	100	99.3	?Thrombocytopenia-2, 188000
MAT1A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	185.4	99.7	97.5	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MAT2A	 HEART PANEL	115.4	99.7	96.9	No OMIM phenotype Thoracic aortic s (Guo (2015) Am J Hum Genet 96, 170)
MATN3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	116.6	84.7	84.7	?Spondyloepimetaphyseal dysplasia, 608728 Epiphyseal dysplasia, multiple, 5, 607078 {Osteoarthritis susceptibility 2}, 140600

MATR3	ALS MENDELIOME	92.5	95.7	88.8	Amyotrophic lateral sclerosis 21, 606070
MAX	HEREDITARY CANCER	86.6	99.4	96.4	{Pheochromocytoma, susceptibility to}, 171300
MB	MUSCLE DISORDERS	153.8	100	100	No OMIM phenotype
MBD5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	196.2	99.9	99.6	Mental retardation, autosomal dominant 1, 156200
MBL2	PRIMARY IMMUNODEFICIENCIES	109.8	100	99.6	{Chronic infections, due to MBL deficiency}, 614372
MBOAT7	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	91.8	99.3	94.7	Mental retardation, autosomal recessive 57, 617188
MBTPS2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	113.4	99.6	97.6	?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014
MC2R	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	213.1	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	MENDELIOME	272	100	100	Obesity, autosomal dominant, 601665
MCC	MENDELIOME	139	100	99.6	Colorectal cancer, somatic, 114500
MCCC1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	151.7	100	99.4	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	131.1	99.9	98.9	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	99.8	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.7	99.9	98.8	Factor V and factor VIII, combined deficiency of, 613625
MCM2	HEARING IMPAIRMENT MENDELIOME	176.8	100	100	?Deafness, autosomal dominant 70, 616968
MCM3AP	MENDELIOME	137.9	100	99.3	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	164.3	99.9	98.8	Immunodeficiency 54, 609981

MCM5	MENDELIOME	131.5	100	100	?Meier-Gorlin syndrome 8, 617564
MCM6	MENDELIOME	154.1	100	99.7	Lactase persistence/nonpersistence, 223100
MCM8	MENDELIOME	123.6	99.9	98.5	?Premature ovarian failure 10, 612885
MCM9	MENDELIOME PRECONCEPTION SCREENING	151.6	100	99.9	Ovarian dysgenesis 4, 616185
MCOLN1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	150.2	98.8	97	Mucolipidosis IV, 252650
MCPH1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	148.6	99.9	98.1	Microcephaly 1, primary, autosomal recessive, 251200
MCUR1	MITOCHONDRIAL DISORDERS	65.3	93.5	77.4	No OMIM phenotype
MDH2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING HEREDITARY CANCER	123.3	98	97.9	Epileptic encephalopathy, early infantile, 51, 617339
MECOM	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	143.4	100	99.6	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	87.3	99.1	93.1	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MECR	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	108.1	98.8	96.1	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	 CRANIOFACIAL ANOMALIES SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	105.7	98	94.8	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED13	INTELLECTUAL DISABILITY	167.6	99.9	99.4	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/29740699

MED13L	CONGENITAL HEART DISEASE HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	134.6	100	99.6	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808
MED17	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	118	95.2	91.7	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	131.2	98.7	96.9	Mental retardation, autosomal recessive 18, 614249
MED25	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.9	99.1	95.7	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEF2C	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	137.7	97.9	93.5	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MEFV	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	108.8	94.9	91	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MEGF10	 MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	154.3	100	99.8	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MEGF8	CRANIOFACIAL ANOMALIES MENDELIOME PRECONCEPTION SCREENING	127.6	99.9	98.6	Carpenter syndrome 2, 614976
MEIOB	MENDELIOME	109.3	98.8	92.5	?Spermatogenic failure 22, 617706
MEIS2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	138.2	100	99.9	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	MENDELIOME HEREDITARY CANCER	123.2	99.5	96.3	Adrenal adenoma, somatic, 0 Angiofibroma, somatic, 0 Carcinoid tumor of lung, 0 Lipoma, somatic, 0 Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic, 0
MEOX1	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	76.8	96.6	91.2	Klippel-Feil syndrome 2, 214300

	PRECONCEPTION SCREENING				
MERTK	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	161.4	99.4	97.7	Retinitis pigmentosa 38, 613862
MESP2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	81.5	93.1	87.9	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	HEARING IMPAIRMENT MENDELIOME HEREDITARY CANCER	184.7	100	99.6	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
METTL23	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	145	100	100	Mental retardation, autosomal recessive 44, 615942
MFAP5	 HEART PANEL MENDELIOME	126.8	100	99.5	Aortic , familial thoracic 9, 616166
MFF	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	93.7	90.4	87.6	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	150.6	100	99.9	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MFRP	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.3	100	100	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	115.1	98.8	95.9	Microcephaly 15, primary, autosomal recessive, 616486
MFSD8	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.1	99.9	98.4	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MGAT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY	157.2	100	99.9	Congenital disorder of glycosylation, type IIa, 212066

	MENDELIOME PRECONCEPTION SCREENING				
MGME1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	151.1	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	132	92.7	91.6	Keutel syndrome, 245150
MIB1	 CARDIO HEART PANEL MENDELIOME	141.7	100	99.6	Left ventricular noncompaction 7, 615092
MICU1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	134.2	96	88.8	Myopathy with extrapyramidal signs, 615673
MICU2	MITOCHONDRIAL DISORDERS	43	94.5	86	No OMIM phenotype Shamseldin et al,Brain 2017
MID1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	164.6	99.8	98.4	Opitz GBBB syndrome, type I, 300000
MID2	INTELLECTUAL DISABILITY MENDELIOME	141	99.6	97.3	?Mental retardation, X-linked 101, 300928
MIEF2	MITOCHONDRIAL DISORDERS	122.9	100	99.9	No OMIM phenotype mitochondrial medicine meeting 2016,Cambridge
MINPP1	METABOLIC DISORDERS MENDELIOME	147.8	98.8	94.8	Thyroid carcinoma, follicular, 188470
MIP	VISION DISORDERS MENDELIOME	132.2	99.8	96.1	Cataract 15, multiple types, 615274
MIPEP	MENDELIOME MITOCHONDRIAL DISORDERS	102.1	95.3	88.6	Combined oxidative phosphorylation deficiency 31, 617228
MIR17HG	MENDELIOME	123456	123456	123456	Feingold syndrome 2, 614326
MIR184	VISION DISORDERS MENDELIOME	123456	123456	123456	EDICT syndrome, 614303
MIR204	MENDELIOME	123456	123456	123456	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96	HEARING IMPAIRMENT MENDELIOME	123456	123456	123456	Deafness, autosomal dominant 50, 613074
MITF	VISION DISORDERS	155.5	100	99.9	COMMAD syndrome, 617306

	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME HEREDITARY CANCER				Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MKKS	VISION DISORDERS CILIO DSD INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKL1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	101	96.2	91.1	Megakaryoblastic leukemia, acute, 0
MKRN3	MENDELIOME	162	100	99.9	Precocious puberty, central, 2, 615346
MKS1	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	114.5	99.9	98.5	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.4	100	99.8	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	162	100	99.7	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLH3	MENDELIOME	151.7	100	100	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089
MLLT10	MENDELIOME	142	94.8	91.4	Leukemia, acute myeloid, 601626
MLPH	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	99.1	99.6	95.9	Griscelli syndrome, type 3, 609227
MLYCD	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	75.6	91.3	86.9	Malonyl-CoA decarboxylase deficiency, 248360

	PRECONCEPTION SCREENING				
MMAA	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	183.2	100	99.6	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMAB	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101.2	100	99.9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110
MMACHC	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	205.8	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	77	89.3	75	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	101.1	98.2	93.6	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP1	MENDELIOME	163.6	100	98.2	COPD, rate of decline of lung function in, 606963 {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600
MMP10		122.3	99.8	98.1	
MMP12		152	99.9	98.4	
MMP13	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	124.1	93.5	91.5	Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400 Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	152.1	100	99.5	?Winchester syndrome, 277950
MMP19	MENDELIOME	129.1	99.6	97.7	Cavitary optic disc anomalies, 611543
MMP2	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	164.4	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	100.5	100	98.6	Amelogenesis imperfecta, type IIA2, 612529

	PRECONCEPTION SCREENING				
MMP21	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME PRECONCEPTION SCREENING	93.3	90.2	84.6	Heterotaxy, visceral, 7, autosomal, 616749
MMP3		101.6	98.5	95.3	{Coronary heart disease, susceptibility to, 6}, 614466
MMP7		180.4	100	99.9	
MMP8		148.7	100	99.9	
MMP9	 SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	125.4	96.2	91.8	Metaphyseal anadysplasia 2, 613073
MN1	MENDELIOME	93.6	98.6	93	Meningioma, 607174
MNX1	MENDELIOME	29.3	63	51.9	Currarino syndrome, 176450
MOCOS	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	169	99.1	96.6	Xanthinuria, type II, 603592
MOCS1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	87.4	98.4	93	Molybdenum cofactor deficiency A, 252150
MOCS2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.5	99.6	99.6	Molybdenum cofactor deficiency B, 252160
MOG	MENDELIOME	105.3	100	99.9	?Narcolepsy 7, 614250
MOGS	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	121.6	99.8	99.1	Congenital disorder of glycosylation, type IIb, 606056
MORC2	NEUROPATHIES MENDELIOME	135.9	100	99.7	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPC1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	121.8	100	99.5	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY	111.8	100	99.7	Congenital disorder of glycosylation, type If, 609180

	MENDELIOME PRECONCEPTION SCREENING				
MPDZ	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	149	98.7	96.6	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	146.2	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	85.4	100	99.3	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	136.7	99.6	97.5	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	72.5	97.1	79.3	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	MENDELIOME PRECONCEPTION SCREENING	155	100	99.9	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	108.5	100	99.4	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	NEUROPATHIES MENDELIOME	123.4	100	99.3	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Roussy-Levy syndrome, 180800
MPZL2	HEARING IMPAIRMENT MENDELIOME	96.9	100	100	Deafness, autosomal recessive 111, 618145
MRAP	MENDELIOME PRECONCEPTION SCREENING	161.3	100	100	Glucocorticoid deficiency 2, 607398
MRE11	MOVEMENT DISORDERS BRSTKNK SKIN DISORDERS	51.2	95.3	82.3	Ataxia-telangiectasia-like disorder 1, 604391

	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER				
MRM2	MITOCHONDRIAL DISORDERS	122.9	99.7	96.9	No OMIM phenotype Encephalomyopathy, childhood-onset and stroke-like episodes (Garone (2017) Hum Mol Genet 26,4257)
MRPL12	MITOCHONDRIAL DISORDERS	107.3	99.4	93.5	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	66.3	91.2	77.9	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	MITOCHONDRIAL DISORDERS	101.6	99.8	96.5	No OMIM phenotype Paper Rotig
MRPL44	MENDELIOME MITOCHONDRIAL DISORDERS	110.5	99.7	97.6	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	MITOCHONDRIAL DISORDERS	155	100	99.5	No OMIM phenotype SSIEM 2016
MRPS14	MITOCHONDRIAL DISORDERS	215.9	100	100	No OMIM phenotype Jackson et al ESHG 2018
MRPS16	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	161.1	100	99.1	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	166.4	99.7	97.9	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	138.8	95.3	91.8	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS23	MITOCHONDRIAL DISORDERS	156.7	100	99.7	No OMIM phenotype Kohda et al,PLoS Genet 2016
MRPS28	MITOCHONDRIAL DISORDERS	122.3	87.9	86.2	No OMIM phenotype Pulman et alHum Mol Gen 2018
MRPS34	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	132.7	99.9	98.3	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	MENDELIOME MITOCHONDRIAL DISORDERS	173.5	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	MITOCHONDRIAL DISORDERS	190.2	100	100	No OMIM phenotype

					?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
MS4A1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	123.7	99.4	96.2	Immunodeficiency, common variable, 5, 613495
MSH2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	113.4	98.6	93.1	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	MENDELIOME HEREDITARY CANCER	113.4	99	95	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4, 617100
MSH5	MENDELIOME	116.5	99.5	96.6	?Premature ovarian failure 13, 617442
MSH6	MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	171.1	100	99.5	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MSL3	INTELLECTUAL DISABILITY	69.8	94.5	82.7	No OMIM phenotype
MSMO1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	45.8	92.6	78.5	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	PRIMARY IMMUNODEFICIENCIES MENDELIOME	88.6	99	95	Immunodeficiency 50, 300988
MSR1	MENDELIOME	170.8	100	99.4	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	155.6	99.7	98.5	Deafness, autosomal recessive 74, 613718
MSTN	MENDELIOME MUSCLE DISORDERS	161.5	100	99.5	Muscle hypertrophy, 614160
MSTO1	MENDELIOME MITOCHONDRIAL DISORDERS	140	99.8	97	Myopathy, mitochondrial, and ataxia, 617675
MSX1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	75.2	95.4	87.5	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	CRANIOFACIAL ANOMALIES MENDELIOME	94.2	98	85.5	Craniosynostosis 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
MTA1		129.3	98	95.6	
MTAP	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	109.2	93.6	86.5	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	124.6	99.3	96.2	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248

	PRECONCEPTION SCREENING				
MTHFD1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	139.6	99.8	98.4	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	 MOVEMENT DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	126.1	98.4	97.2	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTM1	 METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS	93.7	99.2	93	Myotubular myopathy, X-linked, 310400
MTMR2	NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	106.6	100	99.2	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	173.7	89.5	87.3	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	140	100	99.8	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTPAP	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	109.6	98.9	93.5	?Spastic ataxia 4, autosomal recessive, 613672
MTR	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	140.9	99.8	98.8	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.1	100	99.2	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	MOVEMENT DISORDERS	132.4	99.9	98.8	Abetalipoproteinemia, 200100

	MENDELIOME PRECONCEPTION SCREENING				{Metabolic syndrome, protection against}, 605552
MUC1	MENDELIOME	55.6	77.1	66.4	Medullary cystic kidney disease 1, 174000
MUC5B	HEREDITARY CANCER	98.5	87.7	82.4	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUSK	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	159.4	100	99.9	Fetal nesia deformation sequence, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUT	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	121.8	99.2	95.1	Methylmalonic aciduria, mut(0) type, 251000
MUTYH	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	165	100	99.9	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	SKIN DISORDERS MENDELIOME	101.2	100	99	Porokeratosis 7, multiple types, 614714
MVK	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.3	92.1	90.4	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MXI1	MENDELIOME	108.3	90.6	83.6	Neurofibrosarcoma, somatic, 0 {Prostate cancer, susceptibility to, somatic}, 176807
MYBPC1	MENDELIOME PRECONCEPTION SCREENING	150.7	99.9	99.4	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYBPC3	CARDIO HEART PANEL MENDELIOME	142.5	98.5	95.7	Cardiomyopathy, dilated, 1MM, 615396 Cardiomyopathy, hypertrophic, 4, 115197 Left ventricular noncompaction 10, 615396
MYBPHL	HEART PANEL	99.8	99.2	94.2	No OMIM phenotype
MYC	MENDELIOME	114.3	64.8	63.1	Burkitt lymphoma, 113970
MYCN	INTELLECTUAL DISABILITY MENDELIOME	94.1	94.9	84.8	Feingold syndrome 1, 164280
MYD88	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	186.5	100	99.9	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260

MYF5	MENDELIOME	180.4	100	100	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYF6	MENDELIOME MUSCLE DISORDERS	121.6	100	100	Centronuclear myopathy 3, 614408
MYH11	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	132.6	100	99.3	Aortic , familial thoracic 4, 132900
MYH14	HEARING IMPAIRMENT NEUROPATHIES MENDELIOME	102	97.7	91.5	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	129.4	99.9	98.8	Proximal myopathy and ophthalmoplegia, 605637
MYH3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME MUSCLE DISORDERS	110.4	99.9	98.6	Arthrogryposis, distal, type 2A, 193700 Arthrogryposis, distal, type 2B, 601680 Arthrogryposis, distal, type 8, 178110
MYH6	CARDIO CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	113.3	99	96.1	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251 {Sick sinus syndrome 3}, 614090
MYH7	CARDIO CONGENITAL HEART DISEASE HEART PANEL MENDELIOME MUSCLE DISORDERS	111.4	99.4	96.8	Cardiomyopathy, dilated, 1S, 613426 Cardiomyopathy, hypertrophic, 1, 192600 Laing distal myopathy, 160500 Left ventricular noncompaction 5, 613426 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160 Scapuloperoneal syndrome, myopathic type, 181430
MYH7B	HEART PANEL	113.2	97.6	94.5	No OMIM phenotype ?Cardiomyopathy,left ventricular noncompaction (Esposito (2013) Orphanet J Rare Dis 8) ?Hearing loss (Haraksingh (2014) BMC Genomics 15,1155)
MYH8	SKIN DISORDERS MENDELIOME	134.9	100	99.4	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	BONE MARROW FAILURE HEARING IMPAIRMENT HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100

	MENDELIOME				
MYL2	CARDIO HEART PANEL MENDELIOME	134.6	98.7	90.1	Cardiomyopathy, hypertrophic, 10, 608758
MYL3	CARDIO HEART PANEL MENDELIOME PRECONCEPTION SCREENING	103.1	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	HEART PANEL MENDELIOME	158.5	100	100	?Atrial fibrillation, familial, 18, 617280
MYL7	HEART PANEL	130.8	100	100	No OMIM phenotype
MYLK	 HEART PANEL MENDELIOME	148.4	99.9	99.3	Aortic , familial thoracic 7, 613780
MYLK2	HEART PANEL MENDELIOME	120	100	100	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYMK	MENDELIOME PRECONCEPTION SCREENING	141	100	100	Carey-Fineman-Ziter syndrome, 254940
MYO15A	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	116.5	97.3	94	Deafness, autosomal recessive 3, 600316
MYO18B	MENDELIOME PRECONCEPTION SCREENING	132.4	100	99.1	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.9	98.6	97	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	119.8	98.5	93	Deafness, autosomal recessive 30, 607101
MYO5A	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.3	99.5	97.4	Griscelli syndrome, type 1, 214450
MYO5B	MENDELIOME PRECONCEPTION SCREENING	137.6	98.2	95.5	Microvillus inclusion disease, 251850
MYO6	HEARING IMPAIRMENT HEART PANEL MENDELIOME PRECONCEPTION SCREENING	89.7	98.1	92.3	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	VISION DISORDERS	134.1	99.7	98.1	Deafness, autosomal dominant 11, 601317

	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING				Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900
MYO9A	MENDELIOME	142.5	99.5	97.8	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	VISION DISORDERS MENDELIOME	172.5	100	99.3	Glaucoma 1A, primary open angle, 137750
MYOM1	HEART PANEL	149.5	99.8	98.4	No OMIM phenotype
MYOT	HEART PANEL MENDELIOME MUSCLE DISORDERS	139.4	99.3	95.5	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	CARDIO HEART PANEL MENDELIOME	145.9	100	100	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	CARDIO HEART PANEL MENDELIOME PRECONCEPTION SCREENING	142.4	99.3	98.4	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, hypertrophic, 22, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	MENDELIOME	116.1	96.9	95.9	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
MYSM1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	96	97.6	92.5	Bone marrow failure syndrome 4, 618116
MYT1L	INTELLECTUAL DISABILITY MENDELIOME	178.3	100	99.7	Mental retardation, autosomal dominant 39, 616521
MZB1		118.6	100	100	
NAA10	VISION DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	102.4	98.7	96.7	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	INTELLECTUAL DISABILITY MENDELIOME	86.6	96.5	90.9	Mental retardation, autosomal dominant 50, 617787
NACC1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	167.7	100	99.9	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	METABOLIC DISORDERS MENDELIOME	143	96.2	89.3	?2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.4	100	100	Kanz disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NAGLU	NEUROPATHIES	108.7	92.4	90.4	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491

	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	81	98	91.6	N-acetylglutamate synthase deficiency, 237310
NALCN	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.5	99.8	97.5	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	MENDELIOME	28.1	87.1	64.7	Spermatogenic failure 12, 615413
NANS	MOVEMENT DISORDERS EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	106.1	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	120	97.4	97.1	Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	MENDELIOME	70.1	89.3	79.9	?N-acetylaspartate deficiency, 614063
NAXD	MITOCHONDRIAL DISORDERS	138.7	99.9	99	No OMIM phenotype van Bergen et al Brain 2019 142 50-58
NAXE	MENDELIOME MITOCHONDRIAL DISORDERS	81.1	99.7	95.9	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	145.3	99.5	97.6	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEA	HEMOSTATIC/THROMBOTIC DISORDERS INTELLECTUAL DISABILITY	128.4	90.8	89.4	No OMIM phenotype Lentaigne et al.,Blood 2016 127:2814-2823 Inherited platelet disorders: toward DNA-based diagnosis
NBEAL2	BONE MARROW FAILURE	172.7	99.5	99.3	Gray platelet syndrome, 139090

	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				
NBN	BRSTKNK PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	80.6	99.1	94.6	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCAPD2	MENDELIOME	138.5	100	99.1	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	MENDELIOME	120.7	99.4	98	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	CILIO RENAL DISORDERS	127.3	99.3	96.8	nephronophthisis syndrome
NCAPH	MENDELIOME	141.2	100	100	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	23.9	25.8	22.1	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	124.3	100	99.4	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	158.7	100	100	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCOA4	IRON DISORDERS	130.6	94.8	91.3	No OMIM phenotype Bellili R. etal. Cell Reports 2016: combination of iron overload and mild microcytic,hypochromic anemia in mice.
NCOA6	HEART PANEL	150.2	100	99.6	No OMIM phenotype
NCSTN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	111.9	100	99.8	Acne inversa, familial, 1, 142690
NDE1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	100.9	100	99.5	?Microhydranencephaly, 605013 Lissencephaly 4 (with microcephaly), 614019
NDN	MENDELIOME	74.2	92.7	79.7	Prader-Willi syndrome, 176270
NDP	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME	116.8	100	100	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	128.4	99.9	98.8	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	INTELLECTUAL DISABILITY	201.4	100	100	Mental retardation, autosomal recessive 46, 616116

	MENDELIOME PRECONCEPTION SCREENING				
NDUFA1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	166.8	100	99.6	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	MENDELIOME MITOCHONDRIAL DISORDERS	136.7	98.9	96.8	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	86.9	99.5	95.8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	160.2	100	100	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	MENDELIOME MITOCHONDRIAL DISORDERS	91.4	92.3	91.6	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	133.9	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	MITOCHONDRIAL DISORDERS	129.7	91.4	87.4	No OMIM phenotype
NDUFA4	MITOCHONDRIAL DISORDERS	79.9	98.8	84.7	No OMIM phenotype Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795) ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA5	MITOCHONDRIAL DISORDERS	71	87.1	61.3	No OMIM phenotype
NDUFA6	MENDELIOME MITOCHONDRIAL DISORDERS	212.4	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	MITOCHONDRIAL DISORDERS	114.8	100	99.5	No OMIM phenotype
NDUFA8	MITOCHONDRIAL DISORDERS	138.4	100	99.7	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	124.7	98.6	93.2	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	MITOCHONDRIAL DISORDERS	116.3	99.9	96.2	No OMIM phenotype
NDUFAF1	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	115.6	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234

NDUFAF2	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	58.6	85.7	70.9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	120.8	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	79.4	98.9	91.8	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	95.7	98.8	94.5	Mitochondrial complex I deficiency, nuclear type 16, 616238
NDUFAF6	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	79.3	97.1	85.6	Mitochondrial complex I deficiency, nuclear type 17, 612392
NDUFAF7	MITOCHONDRIAL DISORDERS	101.2	100	98.8	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFAF8	MITOCHONDRIAL DISORDERS	30	60.2	47.4	No OMIM phenotype Alston et al ESHG 2018
NDUFB1	MITOCHONDRIAL DISORDERS	29.3	60.5	53	No OMIM phenotype ?Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	MITOCHONDRIAL DISORDERS	120.2	99.4	95.8	No OMIM phenotype Complex I deficiency (Friederich (2016) Hum Mol Genet)
NDUFB11	SKIN DISORDERS IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	109.6	94.4	88	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB2	MITOCHONDRIAL DISORDERS	102.4	100	100	
NDUFB3	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	22.6	91.9	59.2	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	MITOCHONDRIAL DISORDERS	100.6	85	82.9	
NDUFB5	MITOCHONDRIAL DISORDERS	88.5	100	100	

NDUFB6	MITOCHONDRIAL DISORDERS	39.7	99.9	91.4	
NDUFB7	MITOCHONDRIAL DISORDERS	50.4	100	97.4	
NDUFB8	MENDELIOME MITOCHONDRIAL DISORDERS	116.6	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS	120.1	99.8	97.4	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	MITOCHONDRIAL DISORDERS	87.2	100	98.3	
NDUFC2	MITOCHONDRIAL DISORDERS	39.9	98	84	
NDUFS1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	132.2	99.8	98.6	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	VISION DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	117.8	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	142.4	90.7	90.6	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	147.3	100	99.1	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	MITOCHONDRIAL DISORDERS	196.7	100	100	
NDUFS6	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	119.1	99.9	99.4	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	118.4	100	99.7	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	INTELLECTUAL DISABILITY	141.4	100	99.9	Mitochondrial complex I deficiency, nuclear type 2, 618222

	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
NDUFV1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	136.7	99.7	97.8	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	69.5	78.7	53.9	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	MITOCHONDRIAL DISORDERS	119.3	99.9	98.8	
NEB	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	124	82.9	81.9	Nemaline myopathy 2, autosomal recessive, 256030
NEBL	HEART PANEL	102.2	96.7	92.9	No OMIM phenotype Cardiomyopathy,dilated (Purejav (2010) J Am Coll Cardiol 56,1493)
NECAP1	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	116.9	100	99.9	?Epileptic encephalopathy, early infantile, 21, 615833
NECTIN1	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	145.4	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.8	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	105.2	71.7	70.5	Periventricular nodular heterotopia 7, 617201
NEFH	NEUROPATHIES MENDELIOME	111.6	96.6	87.5	?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	164.6	99.7	98.1	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	CILIO DSD SHORT STATURE/SKELETAL DYSPLASIA	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892

	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
NEK11	SKIN DISORDERS	122.7	99.7	95.2	No OMIM phenotype Familial melanoma
NEK2	VISION DISORDERS MENDELIOME	110.4	98.8	93.3	?Retinitis pigmentosa 67, 615565
NEK8	CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	171.4	100	99.9	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	136.8	99.7	98.5	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NEU1	MOVEMENT DISORDERS EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	148.1	99.4	97.1	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEUROD1	VISION DISORDERS MENDELIOME	166.5	100	100	Maturity-onset diabetes of the young 6, 606394 {Diabetes mellitus, noninsulin-dependent}, 125853
NEUROG3	MENDELIOME PRECONCEPTION SCREENING	119.4	100	99.3	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	139.2	99.9	99	Mental retardation, X-linked 98, 300912
NEXN	CARDIO HEART PANEL MENDELIOME	79.8	94.2	79.9	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	125.9	92.3	89.3	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	MOVEMENT DISORDERS MENDELIOME HEREDITARY CANCER	100.2	100	99.9	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091

NFAT5	PRIMARY IMMUNODEFICIENCIES	216.5	99.2	97.8	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
NFE2L2	MENDELIOME	180.4	100	99.6	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	INTELLECTUAL DISABILITY MENDELIOME	145.1	100	99.3	Brain malformations with or without urinary tract defects, 613735
NFIX	INTELLECTUAL DISABILITY MENDELIOME	165.3	97.7	94.9	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	105.3	99.3	96.8	Immunodeficiency, common variable, 12, 616576
NFKB2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	123.3	97.5	92.6	Immunodeficiency, common variable, 10, 615577
NFKBIA	CRANIOFACIAL ANOMALIES SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	116.3	98.5	93.8	Ectodermal dysplasia and immunodeficiency, 612132
NFS1	MITOCHONDRIAL DISORDERS	82.1	86.4	83.5	
NFU1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	47.7	94.9	77.2	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	257.6	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128	100	99.5	Congenital disorder of deglycosylation, 615273
NHEJ1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	80.3	100	99.1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	174.2	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHP2	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	VISION DISORDERS	127.1	94.3	93.3	Cataract 40, X-linked, 302200

	INTELLECTUAL DISABILITY MENDELIOME				Nance-Horan syndrome, 302350
NIN	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	143.5	99.7	98.6	?Seckel syndrome 7, 614851
NIPA1	MOVEMENT DISORDERS MENDELIOME	174.3	99.9	99.1	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	157.8	99.4	93.2	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	116.1	96.5	94.5	Cornelia de Lange syndrome 1, 122470
NKX2-1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	52	96.6	83.3	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NKX2-5	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	83.2	100	99.5	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetralogy of Fallot, 187500 Ventricular septal defect 3, 614432
NKX2-6	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME PRECONCEPTION SCREENING	104.4	100	99.7	Conotruncal heart malformations, 217095 Persistent truncus arteriosus, 217095
NKX3-2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	55.8	92.4	73.9	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	MOVEMENT DISORDERS MENDELIOME	52	79	74.5	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN3	INTELLECTUAL DISABILITY	128.8	100	99	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	INTELLECTUAL DISABILITY MENDELIOME	193.6	99.4	97.1	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497 {Autism susceptibility, X-linked 2}, 300495
NLRC4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	179.9	100	99.7	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES	126.1	99	96.5	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225

	MENDELIOME				{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	PRIMARY IMMUNODEFICIENCIES MENDELIOME	165.9	100	99.9	Familial cold autoinflammatory syndrome 2, 611762
NLRP2	PRECONCEPTION SCREENING	119	100	99.8	No OMIM phenotype Beckwith-Wiedemann syndrome (Meyer (2009) PLoS Genet 5) Nijmegen breakage syndrome
NLRP3	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES MENDELIOME	150.4	100	100	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
NLRP7	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	135.9	99.8	98.7	Hydatidiform mole, recurrent, 1, 231090
NME1	SKIN DISORDERS MENDELIOME	104.2	99.9	99.2	Neuroblastoma, 256700
NME8	CILIO MENDELIOME PRECONCEPTION SCREENING	105.6	97.5	91	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.9	100	99.7	Leber congenital amaurosis 9, 608553
NNT	HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	136.9	98.6	97.1	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	MENDELIOME	79.9	99.5	97.4	Premature ovarian failure 5, 611548
NOD2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	135.8	100	99.7	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321
NODAL	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	160.7	100	99.9	Heterotaxy, visceral, 5, 270100
NOG	CRANIOFACIAL ANOMALIES MENDELIOME	191.9	100	100	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumbs and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NOL3	MOVEMENT DISORDERS	76.3	93.5	83.9	Myoclonus, familial cortical, 614937

	MENDELIOME				
NOMO3		40.2	16.7	16.3	
NONO	INTELLECTUAL DISABILITY MENDELIOME	93.8	99.7	96.4	Mental retardation, X-linked, syndromic 34, 300967
NOP10	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	MENDELIOME	137.3	99.9	98.6	Spinocerebellar ataxia 36, 614153
NOS1AP	HEART PANEL	192.2	100	100	No OMIM phenotype Long QT syndrome (Shigemizu (2015) PLoS One 10,e0130329) ?Obsessive-compulsive disorder (Delorme (2010) BMC Med Genet 11,108) {Cardiac repolarisation, association with} (Arking (2006) Nat Genet 38,644)
NOS3		107.6	95.3	91	{Alzheimer disease, late-onset, susceptibility to}, 104300 {Coronary artery spasm 1, susceptibility to}, 0 {Hypertension, pregnancy-induced}, 189800 {Hypertension, susceptibility to}, 145500 {Ischemic stroke, susceptibility to}, 601367 {Placental abruption}, 0
NOTCH1	CONGENITAL HEART DISEASE SKIN DISORDERS HEART PANEL MENDELIOME	137.5	99.1	98	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	CONGENITAL HEART DISEASE HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	172.4	100	99.9	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH3	MENDELIOME	110.6	93	88.2	?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
NOTCH4		108.4	99.9	98.9	
NPC1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147.9	99.2	97.8	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220

NPC2	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	140.7	100	99.9	Niemann-pick disease, type C2, 607625
NPHP1	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	117.6	98.8	96.4	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.6	99.4	96.1	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	136.7	99.9	99.3	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	102	99.9	98.9	Nephrotic syndrome, type 1, 256300
NPHS2	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	116.9	99.8	95.9	Nephrotic syndrome, type 2, 600995
NPL	METABOLIC DISORDERS	119	100	100	No OMIM phenotype SIEMM (sialic aciduria)
NPM1	MENDELIOME HEREDITARY CANCER	70.3	88.7	77.9	Leukemia, acute myeloid, somatic, 601626
NPPA	HEART PANEL MENDELIOME PRECONCEPTION SCREENING RITME	115.4	100	100	Atrial fibrillation, familial, 6, 612201 Atrial standstill 2, 615745
NPPB	HEART PANEL	160.4	100	100	No OMIM phenotype ?Hypertension (Zeng (2013) J Hum Hypertens 27,271) {Diabetes type 2,reduced risk,association with} (Meirhaeghe (2007) Hum Mol Genet 16,1343)
NPPC	SHORT STATURE/SKELETAL DYSPLASIA	87.8	99.8	96.7	No OMIM phenotype autosomal dominant short stature and brachydactyly (Hisado-

					Olivaetal.GenetMed.2017Jun29.)
NPR2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	164.8	100	100	Acromesomelic dysplasia, Maroteaux type, 602875 Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255
NPRL2	EPILEPSY MENDELIOME	168.5	100	100	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	EPILEPSY MENDELIOME	127.5	100	99.7	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	DSD HH MENDELIOME	119.3	99.9	98.6	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NR0B2	MENDELIOME PRECONCEPTION SCREENING	96.8	100	99.8	Obesity, mild, early-onset, 601665
NR1H4	MENDELIOME PRECONCEPTION SCREENING	141.9	96.6	92.2	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.4	99.9	98.9	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NR2F1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	201.6	99.9	98.4	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	246.1	98.7	94.3	Congenital heart defects, multiple types, 4, 615779
NR3C1	DSD MENDELIOME	137.4	100	99.8	Glucocorticoid resistance, 615962
NR3C2	RENAL DISORDERS MENDELIOME	159.5	99.4	95.9	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NR4A2	INTELLECTUAL DISABILITY	149.2	100	100	No OMIM phenotype
NR4A3	MENDELIOME	112.6	99.8	98	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	DSD MENDELIOME	79.9	100	98.3	46, XX sex reversal 4, 617480 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
NRAS	SKIN DISORDERS HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550

	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER				Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NRL	VISION DISORDERS MENDELIOME	68.7	99.7	95.8	Retinal degeneration, autosomal recessive, clumped pigment type, 0 Retinitis pigmentosa 27, 613750
NRXN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	160.9	96.8	95.7	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	CRANIOFACIAL ANOMALIES SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	155.2	100	99.9	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSD2	INTELLECTUAL DISABILITY	136.9	99.7	97.8	No OMIM phenotype
NSDHL	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	169.2	99.9	98.2	CHILD syndrome, 308050 CK syndrome, 300831
NSMCE2	MENDELIOME	100.3	99	92.5	Seckel syndrome 10, 617253
NSMCE3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	130	99.9	98.5	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	HH MENDELIOME	78.4	95.7	95.2	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.7	95.3	92.2	Mental retardation, autosomal recessive 5, 611091
NSUN3	MITOCHONDRIAL DISORDERS	187.1	100	100	
NT5C2	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.3	97.1	92.7	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	62	89.3	78.8	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	166.3	100	99.8	Calcification of joints and arteries, 211800
NTF4	MENDELIOME	98.8	99.7	92.1	Glaucoma 1, open angle, 1O, 613100
NTHL1	MENDELIOME	98.1	99.1	95.7	Familial adenomatous polyposis 3, 616415

	PRECONCEPTION SCREENING HEREDITARY CANCER				
NTM		200	99.9	98.5	
NTRK1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.6	99.7	97.7	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
NTRK2	INTELLECTUAL DISABILITY MENDELIOME	170.8	100	100	Epileptic encephalopathy, early infantile, 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUBPL	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	89.8	92.9	85.9	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUMA1	MENDELIOME	127.1	100	99.5	Leukemia, acute promyelocytic, somatic, 612376
NUP107	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	122.9	99.2	94.1	?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	RENAL DISORDERS MENDELIOME	127.1	96.9	94.3	Nephrotic syndrome, type 18, 618177
NUP155	HEART PANEL MENDELIOME	115.1	97.6	92.3	?Atrial fibrillation 15, 615770
NUP160	RENAL DISORDERS MENDELIOME	161.1	100	99.8	?Nephrotic syndrome, type 19, 618178
NUP205	RENAL DISORDERS MENDELIOME	133.7	98.9	98	?Nephrotic syndrome, type 13, 616893
NUP214	MENDELIOME	167.3	99.8	99.2	Leukemia, acute myeloid, somatic, 601626 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP37	MENDELIOME	168.6	98.5	93.5	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	111.6	100	99.9	Striatonigral degeneration, infantile, 271930
NUP85	RENAL DISORDERS MENDELIOME	135.3	100	100	Nephrotic syndrome, type 17, 618176
NUP93	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	140.7	97.9	94.9	Nephrotic syndrome, type 12, 616892
NUS1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	69.6	62	40.7	?Congenital disorder of glycosylation, type 1aa, 617082 Mental retardation, autosomal dominant 55, with seizures, 617831

NXN	SHORT STATURE/SKELETAL DYSPLASIA	86	99.7	96.5	No OMIM phenotype Robinow syndrome (PMID: 29575616PMID: 29276006)
NYX	VISION DISORDERS MENDELIOME	94.9	98.1	96	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAT	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	89.2	77.7	70.5	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSCN	HEART PANEL	159.3	99.3	98.2	No OMIM phenotype Cardiomyopathy,dilated (Marston (2015) PLoS One 10,e138568) Glioblastoma (Balakrishnan (2007) Cancer Res 67,3545) ?Breast cancer (Aloraifi (2015) FEBS J epub,epub) ?Schizophrenia (Fromer (2014) Nature 506,179) ?Cardiomyopa
OBSL1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	140.5	99.8	98.5	3-M syndrome 2, 612921
OCA2	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.9	99.5	97.9	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
OCLN	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	220.7	100	100	Pseudo-TORCH syndrome 1, 251290
OCRL	VISION DISORDERS CILIO METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	122.2	98.8	96.3	Dent disease 2, 300555 Lowe syndrome, 309000
ODAM	SKIN DISORDERS	130.5	97.5	90.4	No OMIM phenotype Amelogenesis imperfecta,hypomaturation type,IIA4
ODC1	INTELLECTUAL DISABILITY	146.7	100	99.7	{Colonic adenoma recurrence, reduced risk of}, 114500
OFD1	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIO SKIN DISORDERS EPILEPSY	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209

	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME				
OGDH	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	201.3	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OGG1	MENDELIOME HEREDITARY CANCER	128.5	100	99.6	Renal cell carcinoma, clear cell, somatic, 144700
OGT	INTELLECTUAL DISABILITY MENDELIOME	125.9	100	99.4	Mental retardation, X-linked 106, 300997
OPA1	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	122.5	99.1	94.1	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	VISION DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	128	99.5	97.4	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPHN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	89	99.1	96.2	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
OPLAH	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	109.4	99.4	97.9	5-oxoprolinase deficiency, 260005
OPN1LW	VISION DISORDERS MENDELIOME	76.6	67.4	61.9	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	VISION DISORDERS MENDELIOME	67.2	68.2	60.4	Blue cone monochromacy, 303700 Colorblindness, deutan, 303800
OPN1SW	MENDELIOME	130.2	100	100	Colorblindness, tritan, 190900
OPTN	ALS MENDELIOME	113.8	100	99.4	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	237.3	93.8	89.8	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	SHORT STATURE/SKELETAL DYSPLASIA	106.7	99.9	98.9	Meier-Gorlin syndrome 1, 224690

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
ORC4	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	57.6	95.8	82	Meier-Gorlin syndrome 2, 613800
ORC6	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	126.8	100	100	Meier-Gorlin syndrome 3, 613803
OSBPL2	HEARING IMPAIRMENT MENDELIOME	145.8	100	100	Deafness, autosomal dominant 67, 616340
OSGEP	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	120.5	100	99.6	Galloway-Mowat syndrome 3, 617729
OSMR	SKIN DISORDERS MENDELIOME	145.9	100	99.9	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	80.7	90.8	88.4	Osteopetrosis, autosomal recessive 5, 259720
OTC	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	123.3	99.9	99.4	Ornithine transcarbamylase deficiency, 311250
OTOA	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	117.3	99	96.6	Deafness, autosomal recessive 22, 607039
OTOF	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	131.2	100	99.7	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	145.1	99.4	98.2	Deafness, autosomal recessive 18B, 614945
OTOGL	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	113.3	98.4	93.9	Deafness, autosomal recessive 84B, 614944
OTUD6B	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123.4	99.9	98.2	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	PRIMARY IMMUNODEFICIENCIES MENDELIOME	149.5	90.5	86.3	Autoinflammation, panniculitis, and dermatosis syndrome, 617099

	PRECONCEPTION SCREENING				
OTX2	VISION DISORDERS CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	154.8	100	99.8	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OVOL2	VISION DISORDERS MENDELIOME	119.1	96.9	90.6	Corneal dystrophy, posterior polymorphous, 1, 122000
OXA1L	MITOCHONDRIAL DISORDERS	162	100	100	
OXCT1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.4	99.6	97.8	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
P2RX2	HEARING IMPAIRMENT MENDELIOME	132.8	99	95.5	Deafness, autosomal dominant 41, 608224
P2RY12	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	186.2	100	100	Bleeding disorder, platelet-type, 8, 609821
P3H1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	137	100	99.9	Osteogenesis imperfecta, type VIII, 610915
P3H2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.2	99.2	93.4	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	VISION DISORDERS MENDELIOME	158.7	100	99.5	Myopia 25, autosomal dominant, 617238
P4HB	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	105.1	94.6	94.4	Cole-Carpenter syndrome 1, 112240
PABPN1	MENDELIOME MUSCLE DISORDERS	66.6	62.4	60.5	Oculopharyngeal muscular dystrophy, 164300
PACS1	INTELLECTUAL DISABILITY MENDELIOME	117.4	97.5	95.7	Schuurs-Hoeijmakers syndrome, 615009
PACS2	INTELLECTUAL DISABILITY MENDELIOME	150.7	98.4	95.9	Epileptic encephalopathy, early infantile, 66, 618067
PADI3	SKIN DISORDERS MENDELIOME	148.5	100	100	Uncombable hair syndrome, 191480
PADI6	MENDELIOME	110.5	100	99	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	105.2	89.1	81.4	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAH	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY	151.7	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600

	MENDELIOME PRECONCEPTION SCREENING				
PAK1	MENDELIOME	119.4	100	99.4	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	82.8	97.6	91.8	Mental retardation, X-linked 30/47, 300558
PALB2	BONE MARROW FAILURE BRSTKNK SKIN DISORDERS MENDELIOME HEREDITARY CANCER	152.6	100	99.7	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAM16	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	50.7	65.2	64.7	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	MOVEMENT DISORDERS VISION DISORDERS IRON DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	146.6	99.3	93.1	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANX1	INTELLECTUAL DISABILITY	169	100	100	No OMIM phenotype Intellectual disability, sensorineural hearing loss, skeletal defects and primary ovarian failure (Shao (2016) J Biol Chem 291,12432)
PAPPA2	SHORT STATURE/SKELETAL DYSPLASIA	175.4	100	99.8	No OMIM phenotype Short stature (Dauber (2016) EMBO Mol Med epub,epub)
PAPSS2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	108.5	99.7	98.5	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	MENDELIOME PARK PRECONCEPTION SCREENING	86.9	100	99	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	BONE MARROW FAILURE DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARS2	MITOCHONDRIAL DISORDERS	219.1	100	100	

PATL2	MENDELIOME PRECONCEPTION SCREENING	107.8	99.9	99.2	Oocyte maturation defect 4, 617743
PAX1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	132.4	87.7	82.4	?Otofaciocervical syndrome 2, 615560
PAX2	VISION DISORDERS RENAL DISORDERS MENDELIOME	168.5	99.9	99.3	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PAX3	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME	118.5	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX4	MENDELIOME	96.6	99.9	98.8	Diabetes mellitus, type 2, 125853 Maturity-onset diabetes of the young, type IX, 612225 {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227
PAX5	PRIMARY IMMUNODEFICIENCIES HEREDITARY CANCER	118.8	98.3	95.4	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PAX6	MOVEMENT DISORDERS VISION DISORDERS CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	119.9	100	99.9	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX7	CRANIOFACIAL ANOMALIES MENDELIOME	117.8	100	100	Rhabdomyosarcoma 2, alveolar, 268220
PAX8	INTELLECTUAL DISABILITY MENDELIOME	94.1	100	99.9	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	238.8	99.6	99.3	Tooth agenesis, selective, 3, 604625
PBX1	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	111.8	99.3	95.2	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	149.3	97.7	94.6	Pyruvate carboxylase deficiency, 266150

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
PCBD1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	113.3	99.5	99.1	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	HEART PANEL PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.1	96.4	89.2	Propionicacidemia, 606054
PCCB	HEART PANEL PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.8	98.7	96.4	Propionicacidemia, 606054
PCDH12	MENDELIOME	206.7	100	100	Microcephaly, seizures, spasticity, and brain calcification, 251280
PCDH15	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	153.9	99	98	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083
PCDH19	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	224.1	100	99.3	Epileptic encephalopathy, early infantile, 9, 300088
PCGF2	INTELLECTUAL DISABILITY	94.5	99.9	98.4	no OMIM phenotype ?Developmental disorder (Fitzgerald (2015) Nature 519,223)
PCIF1		144.7	100	99.9	
PCK1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	143	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	METABOLIC DISORDERS PRECONCEPTION SCREENING	194.5	100	99.8	PEPCK deficiency, mitochondrial, 261650
PCLO	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	165.2	99.7	98.5	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	SKIN DISORDERS MENDELIOME	92.1	100	98.2	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY	117.6	98.9	96	Microcephalic osteodysplastic primordial dwarfism, type II, 210720

	MENDELIOME PRECONCEPTION SCREENING				
PCSK1	HH MENDELIOME PRECONCEPTION SCREENING	147.2	100	99	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PCSK9	MENDELIOME	99.3	94.3	91.3	Hypercholesterolemia, familial, 3, 603776 {Low density lipoprotein cholesterol level QTL 1}, 603776
PCYT1A	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	113.5	98.3	94.7	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDCD10	MENDELIOME	85.1	96.3	88	Cerebral cavernous malformations 3, 603285
PDE10A	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	119.8	81.2	80.8	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE11A	MENDELIOME	168.9	99.9	98.9	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	HEARING IMPAIRMENT MENDELIOME	124	100	99.7	?Deafness, autosomal dominant 74, 618140
PDE3A	MENDELIOME	124.5	99.9	98.8	Hypertension and brachydactyly syndrome, 112410
PDE4D	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	101.2	92.8	88.3	Acrodysostosis 2, with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDE6A	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.8	100	99.8	Retinitis pigmentosa 43, 613810
PDE6B	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.9	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	137	99	96.7	Cone dystrophy 4, 613093
PDE6D	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME	106.1	100	99.9	?Joubert syndrome 22, 615665
PDE6G	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	95.3	99.5	96.3	Retinitis pigmentosa 57, 613582
PDE6H	VISION DISORDERS MENDELIOME	68.4	97.6	77	Achromatopsia 6, 610024 Retinal cone dystrophy 3, 610024

	PRECONCEPTION SCREENING				
PDE8B	MOVEMENT DISORDERS MENDELIOME	111	99.9	98.9	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	MOVEMENT DISORDERS SKIN DISORDERS MENDELIOME PARK HEREDITARY CANCER	95.1	100	100	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	MENDELIOME	148.3	100	100	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	MOVEMENT DISORDERS SKIN DISORDERS MENDELIOME PARK	147.1	99.1	96.5	Basal ganglia calcification, idiopathic, 4, 615007 Kos overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDGFRL	MENDELIOME	158.7	100	100	Colorectal cancer, somatic, 114500 Hepatocellular cancer, somatic, 114550
PDHA1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	109.8	98.1	92.1	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	EPILEPSY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	133	99.3	96.8	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	132.5	98.9	94.6	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	MITOCHONDRIAL DISORDERS	127.4	97.7	94	
PDK2	MITOCHONDRIAL DISORDERS	156.8	100	100	
PDK3	NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS	105.1	96.4	94.3	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	MITOCHONDRIAL DISORDERS	110.1	99.8	97.4	
PDLIM3	HEART PANEL	148.4	100	100	No OMIM phenotype Cardiomyopathy,dilated (Arola (2007) Mol Genet Metab 90,435 ?Cardiomyopathy, hypertrophic (Bagnall (2010) Int J Cardiol 145,601)
PDP1	EPILEPSY	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782

	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
PDSS1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	116.7	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	EPILEPSY MENDELIOME PRECONCEPTION SCREENING	35.4	89	72.1	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PDYN	MOVEMENT DISORDERS MENDELIOME	107.1	100	99.9	Spinocerebellar ataxia 23, 610245
PDZD7	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	80.9	98.4	93.9	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	116	99.6	98.5	Prolidase deficiency, 170100
PER2	MENDELIOME	95.7	100	99.6	Advanced sleep phase syndrome, familial, 1, 604348
PER3	MENDELIOME	170.7	99.2	95.8	?Advanced sleep phase syndrome, familial, 3, 616882
PERP	SKIN DISORDERS	151.3	100	100	No OMIM phenotype Keratoderma
PET100	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	94.5	88.8	74.8	Mitochondrial complex IV deficiency, 220110

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
PET117	MITOCHONDRIAL DISORDERS	95.3	100	99.8	
PEX1	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	115.8	97.7	95.4	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	111.8	96.1	90.1	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105.7	99.7	98.3	?Peroxisome biogenesis disorder 14B, 614920
PEX12	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	168.3	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	197.6	99.8	98.7	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	149	99.7	97.5	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	137	97.1	93.1	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	EPILEPSY	92.9	99.9	99.2	Peroxisome biogenesis disorder 12A (Zellweger), 614886

	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
PEX2	MOVEMENT DISORDERS VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147.1	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	VISION DISORDERS HEARING IMPAIRMENT EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	76.4	100	99.8	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	98.1	99.1	94.3	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	111.7	99.9	98.3	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	HEARING IMPAIRMENT EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	94.5	90.4	86.1	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS NEUROPATHIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY	113.5	89.6	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100

	MENDELIOME PRECONCEPTION SCREENING				
PFKM	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	150.4	100	99.8	Glycogen storage disease VII, 232800
PFN1	ALS MENDELIOME	152	100	100	Amyotrophic lateral sclerosis 18, 614808
PGAM2	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	170.9	100	99.9	Glycogen storage disease X, 261670
PGAP1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	98.1	94.9	88.6	Mental retardation, autosomal recessive 42, 615802
PGAP2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	158.4	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	72.8	62.5	58	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	54.5	93.3	81.6	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	CRANIOFACIAL ANOMALIES HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	133.6	100	99.9	Congenital disorder of glycosylation, type It, 614921
PGM3	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	191.4	99.9	99.7	Immunodeficiency 23, 615816
PHACTR1		116.6	100	99.4	

PHC1	MENDELIOME	234.9	100	99.9	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	125	99.9	98	Hypophosphatemic rickets, X-linked dominant, 307800
PHF21A	INTELLECTUAL DISABILITY	111.1	99.1	97.8	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30487643
PHF6	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	62.8	92.6	83.7	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	INTELLECTUAL DISABILITY MENDELIOME	94	99.8	97.7	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	SKIN DISORDERS EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	115.6	100	99.8	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	INTELLECTUAL DISABILITY MENDELIOME	124.5	95.9	91	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991
PHKA1	HEART PANEL METABOLIC DISORDERS MENDELIOME MUSCLE DISORDERS	106.7	98.9	95.3	Muscle glycogenosis, 300559
PHKA2	METABOLIC DISORDERS MENDELIOME	108.2	100	99.5	Glycogen storage disease, type IXa1, 306000 Glycogen storage disease, type IXa2, 306000
PHKB	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.4	99.8	97.5	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	METABOLIC DISORDERS	115.2	99.8	97.8	No OMIM phenotype
PHKG2	MENDELIOME PRECONCEPTION SCREENING	159.1	100	100	Cirrhosis due to liver phosphorylase kinase deficiency, 0 Glycogen storage disease IXc, 613027
PHOX2A	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	29.9	59.9	32.6	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	MENDELIOME HEREDITARY CANCER	92.9	93	87.2	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013

PHYH	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS HEART PANEL NEUROPATHIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	74.6	97.5	90.8	Refsum disease, 266500
PI4KA	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	112.8	93.6	89.4	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	CILIO MENDELIOME	66	96.6	82.4	Joubert syndrome 33, 617767
PICALM	MENDELIOME	106.1	99.9	96.1	Leukemia, acute myeloid, somatic, 601626
PIEZO1	SKIN DISORDERS MENDELIOME	140.2	99.5	97.4	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphatic malformation 6, 616843
PIEZO2	 MENDELIOME PRECONCEPTION SCREENING	126.1	99.9	99.2	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PIGA	SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	90.5	90.4	81.3	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGC	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129	99.7	96.4	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	167.4	100	99.7	Mental retardation, autosomal recessive 53, 616917
PIGH	MENDELIOME	96.5	77.8	68.5	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGL	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	121.8	99.9	99.3	CHIME syndrome, 280000
PIGM	METABOLIC DISORDERS	165.4	100	100	Glycosylphosphatidylinositol deficiency, 610293

	MENDELIOME PRECONCEPTION SCREENING				
PIGN	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	111.3	92.6	87.1	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147	100	99.9	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	EPILEPSY METABOLIC DISORDERS MENDELIOME	101.4	91.6	83.8	?Epileptic encephalopathy, early infantile, 55, 617599
PIGQ	METABOLIC DISORDERS	125.4	92.6	90.7	No OMIM phenotype Intractable seizure, developmental delay, and optic atrophy (Alazami (2015) Cell Rep 10, 148) Ohtahara syndrome (Martin (2014) Hum Mol Genet 23, 3200)
PIGS	MENDELIOME	108.8	100	99.9	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	171.3	98.1	98	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGV	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	145.5	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	147.6	100	99.8	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	121.4	100	99.9	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIH1D3	CILIO MENDELIOME	70.5	94.3	79.6	Ciliary dyskinesia, primary, 36, X-linked, 300991

PIK3C3		121.7	97.8	95.8	
PIK3CA	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	120.7	99.9	99.1	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PIK3CD	PRIMARY IMMUNODEFICIENCIES MENDELIOME	132.7	99.2	96.8	Immunodeficiency 14, 615513
PIK3R1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	129.3	99.7	97.3	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	86.2	89.1	86.1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.1	100	99.8	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	VISION DISORDERS METABOLIC DISORDERS MENDELIOME	141.6	99.8	98.4	Corneal fleck dystrophy, 121850
PINK1	MENDELIOME PARK PRECONCEPTION SCREENING	90.3	87.2	81.1	Parkinson disease 6, early onset, 605909
PIP5K1C	 METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	107.6	96.3	95.1	Lethal congenital contractural syndrome 3, 611369
PISD	SHORT STATURE/SKELETAL DYSPLASIA MITOCHONDRIAL DISORDERS	163	100	100	No OMIM phenotype Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial

					function (PMID: 30488656)
PITPNM3	MENDELIOME	117.2	99	97.8	Cone-rod dystrophy 5, 600977
PITRM1	MITOCHONDRIAL DISORDERS	117.7	97.5	95.7	
PITX1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	144.6	91.4	86.8	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 Liebenberg syndrome, 186550
PITX2	VISION DISORDERS CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE SKIN DISORDERS HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	147.8	99.7	97.5	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PITX3	VISION DISORDERS MENDELIOME	40.2	95.6	82.8	Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, multiple types, 610623 Cataract 11, syndromic, autosomal recessive, 610623
PKD1	CILIO RENAL DISORDERS MENDELIOME	28.1	42.6	34.5	Polycystic kidney disease 1, 173900
PKD1L1	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	123.8	100	99.6	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	CILIO RENAL DISORDERS MENDELIOME	110.6	89.3	84.2	Polycystic kidney disease 2, 613095
PKDCC	SHORT STATURE/SKELETAL DYSPLASIA	81.3	78.2	72.8	No OMIM phenotype skeletal disorder characterised by rhizomelic shortening of extremities and dysmorphic features (PMID: 30478137)
PKHD1	CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	154.9	100	99.7	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	178.8	100	100	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	122	99.9	98.5	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	CARDIO	99.6	94.6	87.7	Arrhythmogenic right ventricular dysplasia 9, 609040

	HEART PANEL MENDELIOME RITME				
PKP4	HEART PANEL	138.9	99.2	96.2	No OMIM phenotype
PLA2G4A	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	134.3	99.9	98.8	Phospholipase A2, group IV A, deficiency of, 0
PLA2G5	VISION DISORDERS METABOLIC DISORDERS	125.4	100	100	[Fleck retina, familial benign], 228980
PLA2G6	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARK PRECONCEPTION SCREENING	117.5	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLA2G7	HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.6	99.9	97.2	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAA	MENDELIOME	169.4	100	99.1	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	MENDELIOME	219.1	100	100	Adenomas, salivary gland pleomorphic, somatic, 181030
PLAT	HEMOSTATIC/THROMBOTIC DISORDERS	97.8	100	99.8	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	111.4	99.8	98.4	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLAUR		122.9	100	100	
PLCB1	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	142.8	100	99.7	Epileptic encephalopathy, early infantile, 12, 613722
PLCB4	CRANIOFACIAL ANOMALIES METABOLIC DISORDERS MENDELIOME	126.1	99.2	95.7	Auriculocondylar syndrome 2, 614669
PLCD1	SKIN DISORDERS METABOLIC DISORDERS	116.9	99.5	97.1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600

	MENDELIOME PRECONCEPTION SCREENING				
PLCE1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	155.3	99.5	98.9	Nephrotic syndrome, type 3, 610725
PLCG2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME	118.9	100	99.8	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLCZ1	MENDELIOME	67.1	96.8	85.1	?Spermatogenic failure 17, 617214
PLD1	MENDELIOME	125.2	99.9	99.1	Cardiac valvular defect, developmental, 212093
PLD3	MENDELIOME	157.2	100	99.3	?Spinocerebellar ataxia 46, 617770
PLEC	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	114.1	99.7	98.7	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	MENDELIOME PRECONCEPTION SCREENING	131.7	99.8	97.9	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	86.9	96.2	89	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PLEKHM1	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	141.1	100	99.9	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLEKHM2	HEART PANEL	112.7	100	99.7	No OMIM phenotype Cardiomyopathy, dilated with left ventricular noncompaction (Muhammad (2015) Hum Mol Genet 24, 7227)
PLG	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	115.4	87.8	87	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	81	96.2	88.4	Lipodystrophy, familial partial, type 4, 613877
PLK4	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA	145.5	99.5	96.3	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
PLN	CARDIO HEART PANEL MENDELIOME RITME	209.7	100	100	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	SKIN DISORDERS HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.9	99.8	97.5	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	108.6	94.7	88.6	Bruck syndrome 2, 609220
PLOD3	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.1	99	96.3	Lysyl hydroxylase 3 deficiency, 612394
PLP1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	129.2	100	99.4	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	112.6	99.1	92.6	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	MENDELIOME	164.4	96.6	84.4	Phospholipid phosphatase 6, 611666
PLS3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	131.5	96.8	95.2	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	MENDELIOME	158.9	100	100	Diarrhea 10, protein-losing enteropathy type, 618183
PLXND1	INTELLECTUAL DISABILITY	110.8	96.9	93.1	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/26068067
PMFBP1	MENDELIOME	109.6	100	99.5	Spermatogenic failure 31, 618112
PML	MENDELIOME	124.4	100	99.9	Leukemia, acute promyelocytic, PML/RARA type, 0
PMM2	MOVEMENT DISORDERS EPILEPSY HEART PANEL NEUROPATHIES	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065

	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
PMP22	NEUROPATHIES MENDELIOME	111.2	96.7	91.9	?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PMPCA	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	120.8	99.4	96.8	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	121	99.7	97.8	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	95.1	83.5	80.7	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMS2CL	HEREDITARY CANCER				No OMIM phenotype
PMVK	SKIN DISORDERS MENDELIOME	125.3	100	99.9	Porokeratosis 1, multiple types, 175800
PNKD	MOVEMENT DISORDERS MENDELIOME	99.8	100	99.2	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	93	99.8	97.7	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLIP	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	160.5	99.5	95.6	?Pancreatic lipase deficiency, 614338
PNMT	METABOLIC DISORDERS	91.5	99.5	96.6	?Hypertension, essential, 145500
PNP	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS	151.4	100	99.5	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING SCID				
PNPLA1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	192.6	100	100	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	SKIN DISORDERS HEART PANEL METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	113.2	99.7	97.4	Neutral lipid storage disease with myopathy, 610717
PNPLA6	MOVEMENT DISORDERS VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	122.1	99.7	98.5	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-MCRANIOFACIAL ANOMALIESrlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
PNPLA8	MENDELIOME MITOCHONDRIAL DISORDERS	114.4	100	99.7	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	EPILEPSY METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	66.4	100	98.3	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	HEARING IMPAIRMENT MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	53.7	93.3	80.9	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POC1A	CILIO SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	133.8	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	78.7	98	94.2	Cone-rod dystrophy 20, 615973
POC5	VISION DISORDERS	123.3	97.5	91.3	No OMIM phenotype Retinitis Pigmentosa
POF1B	MENDELIOME	74.9	90.9	81	?Premature ovarian failure 2B, 300604
POFUT1	SKIN DISORDERS	139.4	99.9	97.5	Dowling-Degos disease 2, 615327

	METABOLIC DISORDERS MENDELIOME				
POGLUT1	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	117.4	98.2	93.8	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POGZ	INTELLECTUAL DISABILITY MENDELIOME	168.2	99.4	99.2	White-Sutton syndrome, 616364
POLA1	PRIMARY IMMUNODEFICIENCIES MENDELIOME	110.7	98.2	92.8	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLD1	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	101.2	93.9	90.8	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	MENDELIOME HEREDITARY CANCER	144.1	100	99.5	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLE2	PRIMARY IMMUNODEFICIENCIES	59	93.4	74.6	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
POLG	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARK PRECONCEPTION SCREENING	114.4	100	99.5	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	MENDELIOME MITOCHONDRIAL DISORDERS	157.5	98.8	96.8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POLH	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	140.7	100	99.8	Xeroderma pigmentosum, variant type, 278750
POLR1A	MENDELIOME	120.1	99.9	99.1	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	MOVEMENT DISORDERS CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	117	99.7	96.1	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	176.2	91.6	91.6	Treacher Collins syndrome 2, 613717
POLR3A	MOVEMENT DISORDERS SKIN DISORDERS METABOLIC DISORDERS	137.4	100	99.9	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694

	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
POLR3B	MOVEMENT DISORDERS SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	146.4	99.9	98.5	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	116.2	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMGNT1	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	127.6	99.7	97.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123
POMGNT2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	259.6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	205.1	100	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	114.4	95.2	87.5	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POMT1	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	155.7	99.7	98.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	HEART PANEL METABOLIC DISORDERS INTELLECTUAL DISABILITY	111.1	98.9	97.5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B,

	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS				2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
POP1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	114.2	100	99.7	Anauxetic dysplasia 2, 617396
POR	DSD MENDELIOME PRECONCEPTION SCREENING	167.7	99.9	98.7	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	117.7	100	99.3	Focal dermal hypoplasia, 305600
POT1	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES HEREDITARY CANCER	90.7	99.6	96	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU1F1	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	106.3	98.2	94.7	Pituitary hormone deficiency, combined, 1, 613038
POU2AF1		111.6	98.8	94.4	
POU3F3	INTELLECTUAL DISABILITY	30.1	67.7	57.2	No OMIM phenotype ?Intellectual disability (Dheedene (2014) Mol Syndromol 5,32)
POU3F4	HEARING IMPAIRMENT MENDELIOME	151	100	99.9	Deafness, X-linked 2, 304400
POU4F3	HEARING IMPAIRMENT MENDELIOME	298.1	100	100	Deafness, autosomal dominant 15, 602459
POU6F2	HEREDITARY CANCER	142.4	100	99.9	{Wilms tumor susceptibility-5}, 601583
PPA2	HEART PANEL MENDELIOME MITOCHONDRIAL DISORDERS	80.4	94.6	82.5	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	MENDELIOME	153.9	100	99.9	Carotid intimal medial thickness 1, 609338 Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 [Obesity, resistance to], 0 {Diabetes, type 2}, 125853
PPARGC1A		142.6	99.9	99.3	
PPCS	HEART PANEL METABOLIC DISORDERS	108.7	99.9	98.4	Cardiomyopathy, dilated, 2C, 618189

	MENDELIOME MITOCHONDRIAL DISORDERS				
PPIB	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	118.4	100	100	Osteogenesis imperfecta, type IX, 259440
PPIP5K2	HEARING IMPAIRMENT	74.7	96.6	86.6	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/29590114
PPM1D	INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	166.7	100	99.8	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450
PPM1K	METABOLIC DISORDERS MENDELIOME	180	100	100	?Maple syrup urine disease, mild variant, 615135
PPOX	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	96.1	99.7	98.2	Porphyria variegata, 176200
PPP1CB	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	96.6	99.6	98.4	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R15B	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	133.4	99.4	98	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R3A	MENDELIOME	146.5	99.5	98	Insulin resistance, severe, digenic, 125853
PPP2R1A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	134	91.7	91.6	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	MENDELIOME	156	100	99.8	Lung cancer, 211980
PPP2R2B	MENDELIOME	141.9	99.7	97.3	Spinocerebellar ataxia 12, 604326
PPP2R5B	INTELLECTUAL DISABILITY	111.3	99.6	95.1	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24, 4775)
PPP2R5C	INTELLECTUAL DISABILITY	107.6	95.1	88.2	No OMIM phenotype Overgrowth (Loveday (2015) Hum Mol Genet 24,4775)
PPP2R5D	INTELLECTUAL DISABILITY MENDELIOME	143.2	100	99.8	Mental retardation, autosomal dominant 35, 616355
PPP3CA	INTELLECTUAL DISABILITY MENDELIOME	123.8	99	92.9	Epileptic encephalopathy, infantile or early childhood, 1, 617711
PPT1	VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	144.5	90	87.3	Ceroid lipofuscinosis, neuronal, 1, 256730

PQBP1	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	186.1	100	100	Renpenning syndrome, 309500
PRCC	MENDELIOME	146.6	99.8	97.9	Renal cell carcinoma, papillary, 605074
PRCD	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	89.4	100	99.9	Retinitis pigmentosa 36, 610599
PRDM12	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	112.2	91	87.7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	VISION DISORDERS	122.6	92.7	87.8	No OMIM phenotype Macular dystrophy, North Carolina (Small (2016) Ophthalmology 123, 9)
PRDM16	CARDIO HEART PANEL MENDELIOME	161.5	100	99.1	Cardiomyopathy, dilated, 1LL, 615373 Left ventricular noncompaction 8, 615373
PRDM5	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	129.4	99.5	95.5	Brittle cornea syndrome 2, 614170
PRDM6	MENDELIOME	108.3	92.8	79	Patent ductus arteriosus 3, 617039
PRDM8	MENDELIOME	81.2	92	85.1	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	MENDELIOME	115	100	99.6	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	MENDELIOME	107.6	99.5	95.9	Myasthenic syndrome, congenital, 22, 616224
PRF1	MOVEMENT DISORDERS BONE MARROW FAILURE EPILEPSY PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRG4	MENDELIOME PRECONCEPTION SCREENING	144.6	97.6	88.4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRECONCEPTION SCREENING	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRIMPOL	VISION DISORDERS MENDELIOME	110.1	95.7	90.1	Myopia 22, autosomal dominant, 615420
PRKAA1	MITOCHONDRIAL DISORDERS	119.9	100	99.3	
PRKACA	MENDELIOME	97.4	78.9	76.9	Cushing syndrome, ACTH-independent adrenal, somatic, 615830
PRKACG	HEMOSTATIC/THROMBOTIC DISORDERS	217.6	100	99.9	?Bleeding disorder, platelet-type, 19, 616176

	MENDELIOME				
PRKAG2	CARDIO HEART PANEL METABOLIC DISORDERS MENDELIOME RITME	125.6	98.1	91.6	Cardiomyopathy, hypertrophic 6, 600858 Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200
PRKAR1A	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	90.7	99.1	93.9	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKCA	MENDELIOME	150.1	100	100	Pituitary tumor, invasive, 0
PRKCB	HEARING IMPAIRMENT	164.5	100	99.8	No OMIM phenotype Hearing loss (Martin-Sierra (2016) Hum Mol Genet epub,epub)
PRKCD	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	181.2	100	99.9	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	MOVEMENT DISORDERS MENDELIOME	116.1	99	94.5	Spinocerebellar ataxia 14, 605361
PRKCSH	METABOLIC DISORDERS MENDELIOME	135.2	99.7	96.3	Polycystic liver disease 1, 174050
PRKD1	MENDELIOME	145.3	97.4	94.8	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	106.7	98.4	94.8	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	 HEART PANEL MENDELIOME	123.4	98.7	95.4	Aortic , familial thoracic 8, 615436
PRKN	MENDELIOME PARK PRECONCEPTION SCREENING HEREDITARY CANCER	98.6	79.6	78.8	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PRKRA	MOVEMENT DISORDERS MENDELIOME PARK PRECONCEPTION SCREENING	179.6	99.8	98.4	Dystonia 16, 612067
PRLR	MENDELIOME	151.2	100	99.9	?Hyperprolactinemia, 615555 Multiple fibroadenomas of the breast, 615554
PRMT7	INTELLECTUAL DISABILITY MENDELIOME	138.7	100	99.8	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157

	PRECONCEPTION SCREENING				
PRNP	MENDELIOME	153.4	100	100	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Prion disease with protracted course, 606688 {Kuru, susceptibility to}, 245300
PROC	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.5	99.7	97.2	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	83.8	84.9	82.3	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROK2	HH MENDELIOME	105.6	98.4	91.9	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	HH SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	331.8	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	112.3	95.4	92.8	Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786
PROP1	HH SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	76.5	91.6	84.3	Pituitary hormone deficiency, combined, 2, 262600
PROS1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.4	96.8	91.4	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	HEMOSTATIC/THROMBOTIC DISORDERS	131.3	99.9	98.2	[Protein Z deficiency], 614024
PRPF3	VISION DISORDERS MENDELIOME	86	98.8	96	Retinitis pigmentosa 18, 601414
PRPF31	VISION DISORDERS MENDELIOME	115.9	97.5	92	Retinitis pigmentosa 11, 600138
PRPF4	VISION DISORDERS MENDELIOME	149.4	100	99.4	Retinitis pigmentosa 70, 615922
PRPF6	VISION DISORDERS MENDELIOME	130.3	100	100	Retinitis pigmentosa 60, 613983
PRPF8	VISION DISORDERS	139	99.9	99	Retinitis pigmentosa 13, 600059

	MENDELIOME				
PRPH2	VISION DISORDERS MENDELIOME	244.1	100	100	Choroidal dystrophy, central areolar 2, 613105 Leber congenital amaurosis 18, 608133 Macular dystrophy, patterned, 1, 169150 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133 Retinitis punctata albescens, 136880
PRPS1	HEARING IMPAIRMENT NEUROPATHIES PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	149.5	100	100	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRR12	INTELLECTUAL DISABILITY	75	94.6	86.2	No OMIM phenotype PMID 29556724, in house recurrency
PRRT2	MOVEMENT DISORDERS EPILEPSY MENDELIOME	78.9	99.9	98.4	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PRRX1	MENDELIOME	100.2	100	99.9	Agnathia-otocephaly complex, 202650
PRSS1	MENDELIOME HEREDITARY CANCER	190.8	100	99.9	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PRSS12	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	153.3	99.9	98.5	Mental retardation, autosomal recessive 1, 249500
PRSS56	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	51.5	96.6	83.8	Microphthalmia, isolated 6, 613517
PRUNE1	MENDELIOME	136.1	100	100	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	115.6	99.8	98.3	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	MOVEMENT DISORDERS EPILEPSY NEUROPATHIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.4	99.9	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1		53.2	91.4	75.8	?Phosphoserine aminotransferase deficiency, 610992

	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				Neu-Laxova syndrome 2, 616038
PSEN1	SKIN DISORDERS MENDELIOME PARK	160.7	100	99.9	?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
PSEN2	MENDELIOME	123.9	100	100	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	67.6	100	98.4	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	PRIMARY IMMUNODEFICIENCIES	66.5	100	97.8	No OMIM phenotype autoinflammatory disorder (INFEVERS website http://fmf.igh.cnrs.fr/ISSAID/infevers/) and based on literature it underlies a human primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
PSMB4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	122.5	100	99.2	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	118.7	100	99.8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	PRIMARY IMMUNODEFICIENCIES MENDELIOME	85.6	99.5	95.7	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3IP	MENDELIOME PRECONCEPTION SCREENING	113.4	99.9	99.7	Ovarian dysgenesis 3, 614324
PSMD12	INTELLECTUAL DISABILITY MENDELIOME	76.3	98.1	90.9	Stankiewicz-Isidor syndrome, 617516
PSPH	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128.9	98.8	95.4	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	88.2	99.7	97.7	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCD3	MITOCHONDRIAL DISORDERS	93.7	97	93.7	

PTCH1	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	114.6	98.4	95.9	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	120.1	99.4	97.5	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTCONGENITAL HEART DISEASE1	INTELLECTUAL DISABILITY	157.2	100	99.8	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	127.2	100	100	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	BRSTKNK SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	143.2	99.6	96	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTF1A	MENDELIOME PRECONCEPTION SCREENING	74	88.4	78.1	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069
PTGIS	METABOLIC DISORDERS MENDELIOME	126.7	96.7	94.6	Hypertension, essential, 145500
PTGS1	HEMOSTATIC/THROMBOTIC DISORDERS	148	99.6	99	No OMIM phenotype
PTH	MENDELIOME	103.5	99.9	97.6	Hypoparathyroidism, autosomal dominant, 146200 Hypoparathyroidism, autosomal recessive, 146200
PTH1R	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	108.5	99.9	98.8	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTHLH	SKIN DISORDERS MENDELIOME	120.5	99.4	93.2	Brachydactyly, type E2, 613382
PTPN11	CONGENITAL HEART DISEASE SKIN DISORDERS HEART PANEL	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950

	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER				
PTPN12	MENDELIOME	144.8	97.7	94.8	Colon cancer, somatic, 114500
PTPN14	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	175.7	99.4	96.4	?Choanal atresia and lymphedema, 613611
PTPN22	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES	134.5	98	91.9	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	101.6	93.9	86.3	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
PTPRF	SKIN DISORDERS MENDELIOME	170.9	100	99.9	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	MENDELIOME	169	97.2	96.1	Colon cancer, somatic, 114500
PTPRO	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	140.8	99.9	99	Nephrotic syndrome, type 6, 614196
PTPRQ	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	104.7	93.3	89.1	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	279.6	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	INTELLECTUAL DISABILITY	144	100	100	No OMIM phenotype ?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry)
PTS	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	107.2	99.6	94.1	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	INTELLECTUAL DISABILITY MENDELIOME	173.4	99.9	98.3	Verheij syndrome, 615583
PUM1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY	158	100	99.9	Spinocerebellar ataxia 47, 617931

	MENDELIOME				
PURA	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	121.8	94.5	87.2	Mental retardation, autosomal dominant 31, 616158
PUS1	IRON DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	127.2	98.6	93.9	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	192	100	100	?Mental retardation, autosomal recessive 55, 617051
PUS7	INTELLECTUAL DISABILITY PRECONCEPTION SCREENING	136.5	99.8	98	No OMIM phenotype ?Intellectual disability (Riazuddin (2017) Mol Psychiatry 22,1604)
PXDN	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	163.8	99.8	98.5	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	86.3	99.4	94.3	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	127.6	100	97.6	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	158.6	100	100	Glycogen storage disease VI, 232700
PYGM	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	127.1	100	99.9	McArdle disease, 232600
PYROXD1	MENDELIOME MITOCHONDRIAL DISORDERS	48.4	85.5	70.6	Myopathy, myofibrillar, 8, 617258
QARS	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	166.6	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760

	PRECONCEPTION SCREENING				
QDPR	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	92.3	100	99.5	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	INTELLECTUAL DISABILITY MENDELIOME	165.9	99.9	98.9	Ververi-Brady syndrome, 617982
QRSL1	MITOCHONDRIAL DISORDERS	96.7	98.7	93.8	
RAB11B	INTELLECTUAL DISABILITY MENDELIOME	249.8	100	100	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB18	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	82.7	97.1	86.4	Warburg micro syndrome 3, 614222
RAB23	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.3	99.7	98	Carpenter syndrome, 201000
RAB27A	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.9	100	99.9	Griscelli syndrome, type 2, 607624
RAB28	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	52.1	96.5	87.1	Cone-rod dystrophy 18, 615374
RAB33B	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	233.9	100	100	Smith-McCort dysplasia 2, 615222
RAB39B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	113	100	99.7	?Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RAB3GAP1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124.2	99.4	98.8	Warburg micro syndrome 1, 600118
RAB3GAP2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	94.1	98.4	93.9	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225

RAB7A	NEUROPATHIES MENDELIOME	157.9	100	100	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	INTELLECTUAL DISABILITY MENDELIOME	117.9	97.6	92.8	Mental retardation, autosomal dominant 48, 617751
RAC2	PRIMARY IMMUNODEFICIENCIES MENDELIOME SCID	104.1	100	99.4	Neutrophil immunodeficiency syndrome, 608203
RAC3	SHORT STATURE/SKELETAL DYSPLASIA	88.9	94	94	No OMIM phenotype Robinow syndroom
RAD21	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	78.5	98.8	94.7	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	BRSTKNK SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	99	92.6	86.2	Nijmegen breakage syndrome-like disorder, 613078
RAD51	MOVEMENT DISORDERS BONE MARROW FAILURE MENDELIOME	123.2	89.4	89.4	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RAD51C	BONE MARROW FAILURE BRSTKNK MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	143.4	100	98.9	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	BRSTKNK HEREDITARY CANCER	153.9	100	99.5	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAD54B	MENDELIOME	104.6	99	93.9	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	MENDELIOME	123.6	100	99.2	Adenocarcinoma, colonic, somatic, 0 Lymphoma, non-Hodgkin, somatic, 605027 {Breast cancer, invasive ductal}, 114480
RAF1	SKIN DISORDERS HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553

RAG1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	206.9	100	100	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	221	100	100	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	SKIN DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME	146.3	100	99.7	Smith-Magenis syndrome, 182290
RALA	INTELLECTUAL DISABILITY	132.9	83.2	80.1	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30500825
RANBP2	PRIMARY IMMUNODEFICIENCIES	110.9	49.7	48.9	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RANGRF	HEART PANEL RITME	114	99.9	98.4	No OMIM phenotype Brugada syndrome (Selga (2015) PLoS One 10,e0132888 Histiocytoid cardiomyopathy (Cataldo (2014)
RAP1GDS1	MENDELIOME	96.2	99.3	93.5	Lymphocytic leukemia, acute T-cell, 0
RAPGEF2	MENDELIOME	157.3	99	97.5	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	140.5	99.6	96.3	Fetal nesia deformation sequence, 208150 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	118.7	100	100	Microphthalmia, syndromic 12, 615524
RARS	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	86.4	92.7	85.9	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	107.2	100	99.1	Pontocerebellar hypoplasia, type 6, 611523
RASA1	MENDELIOME	96.9	95.6	84.6	Basal cell carcinoma, somatic, 605462 Capillary malformation-arteriovenous malformation 1, 608354

RASGRP1	PRIMARY IMMUNODEFICIENCIES	127.5	99.9	99.5	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
RASGRP2	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	97.5	99.9	98.6	?Bleeding disorder, platelet-type, 18, 615888
RAX	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	82.5	88.7	77.3	Microphthalmia, isolated 3, 611038
RAX2	VISION DISORDERS MENDELIOME	52.2	91	67.8	?Macular degeneration, age-related, 6, 613757 Cone-rod dystrophy 11, 610381
RB1	MENDELIOME HEREDITARY CANCER	88	90.1	76.3	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RB1CC1	MENDELIOME	111.6	96.8	91	Breast cancer, somatic, 114480
RBBP8	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	110.7	99.6	96.4	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744
RBCK1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	104.1	99.2	94.9	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX1	INTELLECTUAL DISABILITY	136.7	89.1	87.8	No OMIM phenotype Epilepsy, rolandic (Lal (2013) PLoS One 8, e73323) Mental retardation (Bhalla (2004) J Hum Genet 49, 308 ?Autism spectrum disorder (Griswold (2015) Mol Autism 6, 43) ?Developmental coordination disorder (Mosca (2016) J Med Ge
RBM10	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	112.1	99.4	95.4	TARP syndrome, 311900
RBM20	CARDIO HEART PANEL MENDELIOME	180.9	99.2	96.6	Cardiomyopathy, dilated, 1DD, 613172
RBM28	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	138.7	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079

	PRECONCEPTION SCREENING				
RBM8A	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	106.6	100	99.4	Thrombocytopenia-absent radius syndrome, 274000
RBMX	MENDELIOME	52.9	94	82.1	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238
RBP3	VISION DISORDERS MENDELIOME	155.2	100	100	?Retinitis pigmentosa 66, 615233
RBP4	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	99.6	99.4	96.1	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	89.2	94.1	86.4	Adams-Oliver syndrome 3, 614814
RCBTB1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123.7	100	99.7	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	161.6	100	99.9	Leber congenital amaurosis 12, 610612
RDH11	VISION DISORDERS MENDELIOME	119.2	100	99.9	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	94.4	100	98.4	Leber congenital amaurosis 13, 612712
RDH5	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	160.2	100	99.7	Fundus albipunctatus, 136880
RDX	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	43.2	84.7	64.8	Deafness, autosomal recessive 24, 611022
RECQL	HEREDITARY CANCER	127.4	99.7	96.1	No OMIM phenotype Breast cancer (Cybulski (2015) Nat Genet 47,643)
RECQL4	CRANIOFACIAL ANOMALIES SKIN DISORDERS	149.6	99.2	96.5	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280

	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER				Rothmund-Thomson syndrome, 268400
REEP1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME	78.3	76.3	75.7	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
REEP2	MENDELIOME	135.1	98.7	96.1	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	VISION DISORDERS MENDELIOME	171.8	99.4	96.4	Retinitis pigmentosa 77, 617304
RELB	PRIMARY IMMUNODEFICIENCIES MENDELIOME	90.6	87.9	75.1	?Immunodeficiency 53, 617585
RELN	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	155.6	100	99.8	Lissencephaly 2 (Norman-Roberts type), 257320 {Epilepsy, familial temporal lobe, 7}, 616436
REN	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.9	100	100	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
REPS1	MENDELIOME	123	96	93.5	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	INTELLECTUAL DISABILITY MENDELIOME	71.4	94.9	88.2	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	HEARING IMPAIRMENT MENDELIOME HEREDITARY CANCER	128.6	98.5	98.4	Fibromatosis, gingival, 5, 617626 {Wilms tumor 6, susceptibility to}, 616806
RET	MENDELIOME HEREDITARY CANCER	141	99.7	97.8	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, protection against}, 142623 {Hirschsprung disease, susceptibility to, 1}, 142623
RETREG1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	126.1	95.6	90.1	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
REV3L	INTELLECTUAL DISABILITY	133.3	97.1	94.5	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/26068067
RFT1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	108.3	99.8	97.3	Congenital disorder of glycosylation, type In, 612015

	PRECONCEPTION SCREENING				
RFWD3	MENDELIOME	139.2	99.8	98.9	?Fanconi anemia, complementation group W, 617784
RFX5	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	116.9	98.7	96.3	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	MENDELIOME PRECONCEPTION SCREENING	153.9	100	99.6	Mitchell-Riley syndrome, 615710
RFXANK	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	105.9	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	84.8	94.4	91.6	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	MENDELIOME PRECONCEPTION SCREENING	126.8	100	99.4	Retinitis pigmentosa 44, 613769
RGS9	VISION DISORDERS MENDELIOME	101.4	98.5	97.2	Bradyopsia, 608415
RGS9BP	VISION DISORDERS MENDELIOME	93.9	100	99.6	Bradyopsia, 608415
RHAG	MENDELIOME	151.4	100	99.2	Anemia, hemolytic, Rh-null, regulator type, 268150 Overhydrated hereditary stomatocytosis, 185000
RHBDF2	SKIN DISORDERS MENDELIOME HEREDITARY CANCER	97.7	99.5	97	Tylosis with esophageal cancer, 148500
RHCE	MENDELIOME	216.6	97.7	97.2	Rh-null disease, amorph type, 617970 [Blood group, Rhesus], 0
RHEB	INTELLECTUAL DISABILITY	39.4	88.7	75.1	No OMIM phenotype
RHO	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	210.1	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RHOA	SKIN DISORDERS	101.4	81.6	80.7	No OMIM phenotype neuroectodermal syndrome
RHOBTB2	INTELLECTUAL DISABILITY MENDELIOME	227.5	100	100	Epileptic encephalopathy, early infantile, 64, 618004
RHOH	PRIMARY IMMUNODEFICIENCIES	134.9	100	100	No OMIM phenotype Combined T-cell and B-cell immunodeficiencies 1 studie Crequer A et al.,2012

RIMS1	VISION DISORDERS MENDELIOME	126.6	98.8	96	Cone-rod dystrophy 7, 603649
RIN2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	113.4	100	99.9	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	HEREDITARY CANCER	171	99.7	97.5	No OMIM phenotype ?Breast cancer (Park (2014) Cancer Discov 4, 804)
RIPK1	MENDELIOME	128.9	100	98.7	Immunodeficiency 57, 618108
RIPK4	CRANIOFACIAL ANOMALIES SKIN DISORDERS DSD MENDELIOME PRECONCEPTION SCREENING	163.3	100	99.6	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RIPOR2	HEARING IMPAIRMENT MENDELIOME	121.4	100	99.9	?Deafness, autosomal recessive 104, 616515
RIPPLY2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	63.5	99	83.8	?Spondylocostal dysostosis 6, 616566
RIT1	HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	165.6	100	100	Noonan syndrome 8, 615355
RLBP1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	144.8	100	100	Bothnia retinal dystrophy, 607475 Fundus albipunctatus, 136880 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880
RLIM	INTELLECTUAL DISABILITY MENDELIOME	149.6	99.6	97.8	Tonne-Kalscheuer syndrome, 300978
RMND1	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	137.2	99.8	97.3	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	123456	123456	123456	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460

	SCID				
RNASEH1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	98.6	99.1	95.6	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	142.1	100	99.9	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.8	93.2	87.5	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	209.2	100	99.9	Aicardi-Goutieres syndrome 3, 610329
RNASEL	MENDELIOME	147.9	100	99.6	Prostate cancer 1, 601518
RNASET2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	96.4	91.9	88.3	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	INTELLECTUAL DISABILITY MENDELIOME	144.8	100	100	?Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	INTELLECTUAL DISABILITY MENDELIOME	177.8	100	99.1	Tenorio syndrome, 616260
RNF135	MENDELIOME	96	94	91.6	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192
RNF139	MENDELIOME	223.5	100	100	Renal cell carcinoma, 144700
RNF168	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	215.3	100	99.1	RIDDLE syndrome, 611943
RNF170	MOVEMENT DISORDERS MENDELIOME	147.1	98.3	91.2	Ataxia, sensory, 1, autosomal dominant, 608984
RNF212	MENDELIOME	118.7	99.1	96.9	Recombination rate QTL 1, 612042

RNF213		157	100	99.5	{Moyamoya disease 2, susceptibility to}, 607151
RNF216	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	137.1	99.8	98.6	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF31	PRIMARY IMMUNODEFICIENCIES	154.7	99.5	98.2	No OMIM phenotype Autoinflammation, immunodeficiency, amylopectinosis and lymphangiectasia (Boisson (2015) J Exp Med 212,939)
RNF43	MENDELIOME HEREDITARY CANCER	126.5	100	99.6	Sessile serrated polyposis cancer syndrome, 617108
RNF6	MENDELIOME	195.4	100	99.4	Esophageal carcinoma, somatic, 133239
RNPC3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	41.3	85.1	61.1	?Growth hormone deficiency, isolated, type V, 618160
RNU4ATAC	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	123456	123456	123456	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROBO2	RENAL DISORDERS MENDELIOME	155.2	97.9	96.7	Vesicoureteral reflux 2, 610878
ROBO3	MENDELIOME PRECONCEPTION SCREENING	99.7	98.7	95.6	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4		92.8	99.9	98.1	
ROGDI	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	112.2	97.9	95.3	Kohlschutter-Tonz syndrome, 226750
ROM1	VISION DISORDERS MENDELIOME	115	100	99.4	Retinitis pigmentosa 7, digenic form, 608133
ROR1	HEARING IMPAIRMENT MENDELIOME	177.5	96.8	96.8	?Deafness, autosomal recessive 108, 617654
ROR2	DSD SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	165.9	99.4	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	INTELLECTUAL DISABILITY MENDELIOME	129.8	92.1	87.7	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	132.3	100	100	Immunodeficiency 42, 616622
RP1	VISION DISORDERS	121.8	91.4	90.3	Retinitis pigmentosa 1, 180100

	MENDELIOME PRECONCEPTION SCREENING				
RP1L1	VISION DISORDERS MENDELIOME	94.8	100	99.8	Occult macular dystrophy, 613587
RP2	VISION DISORDERS MENDELIOME	180	100	98.9	Retinitis pigmentosa 2, 312600
RP9	VISION DISORDERS MENDELIOME	62.9	77.8	76	?Retinitis pigmentosa 9, 180104
RPE65	VISION DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.3	100	99.3	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 20, 613794
RPGR	VISION DISORDERS MENDELIOME	91.8	83.1	73.8	Cone-rod dystrophy, X-linked, 1, 304020 Macular degeneration, X-linked atrophic, 300834 Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455
RPGRIP1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	154.2	100	99.9	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RPIA	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	113.1	94.3	90.9	?Ribose 5-phosphate isomerase deficiency, 608611
RPL10	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	85.9	99.1	93.1	Mental retardation, X-linked, syndromic, 35, 300998 {Autism, susceptibility to, X-linked 5}, 300847
RPL11	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	99.8	100	99.5	Diamond-Blackfan anemia 7, 612562
RPL15	BONE MARROW FAILURE MENDELIOME	41.6	88.3	81.2	?Diamond-Blackfan anemia 12, 615550
RPL18	BONE MARROW FAILURE	88	100	98.6	No OMIM phenotype Dooijeweert et al.2017 Eur J Haematol 2018

					100-163-170
RPL21	SKIN DISORDERS MENDELIOME	64.7	79	57.4	Hypotrichosis 12, 615885
RPL26	BONE MARROW FAILURE MENDELIOME	46.8	94.8	83.7	?Diamond-Blackfan anemia 11, 614900
RPL27	BONE MARROW FAILURE MENDELIOME	39.3	80.4	58.3	?Diamond-Blackfan anemia 16, 617408
RPL31	BONE MARROW FAILURE	84.7	99.7	95.9	No OMIM phenotype Dooijeweert et al.2017 Eur J Haematol 2018 100-163-170
RPL35A	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	83.9	99.2	91	Diamond-Blackfan anemia 5, 612528
RPL5	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	43.8	81.8	69.3	Diamond-Blackfan anemia 6, 612561
RPL9	BONE MARROW FAILURE	78	99	91.8	No OMIM phenotype Dooijeweert et al.2017 Eur J Haematol 2018 100-163-170
RPS10	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	140.1	99.8	97.1	Diamond-Blackfan anemia 9, 613308
RPS14	MENDELIOME	133.3	99.7	96.2	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	BONE MARROW FAILURE	80.9	99.2	92.1	No OMIM phenotype Dooijeweert et al.2017 Eur J Haematol 2018 100-163-170
RPS17	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	52.4	85	73.7	Diamond-Blackfan anemia 4, 612527
RPS19	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	82.5	99.7	95.5	Diamond-Blackfan anemia 1, 105650
RPS20	HEREDITARY CANCER	74.7	99.7	94.1	No OMIM phenotype Colorectal cancer, non-polyposis (Nieminen (2014) Gastroenterology 147,595)
RPS23	MENDELIOME	63.2	90.3	82.2	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	110.4	92.4	87.2	Diamond-blackfan anemia 3, 610629

RPS26	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	106.8	94.8	82.1	Diamond-Blackfan anemia 10, 613309
RPS27	BONE MARROW FAILURE MENDELIOME	39.7	86.7	60.3	?Diamond-Blackfan anemia 17, 617409
RPS28	BONE MARROW FAILURE MENDELIOME	47.7	100	92	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	BONE MARROW FAILURE MENDELIOME	106.2	97.9	96.6	Diamond-Blackfan anemia 13, 615909
RPS6KA3	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	79.3	94.2	83.3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RPS7	BONE MARROW FAILURE MENDELIOME HEREDITARY CANCER	93.7	76.9	63.4	Diamond-Blackfan anemia 8, 612563
RPSA	PRIMARY IMMUNODEFICIENCIES MENDELIOME	88.8	100	99.7	Asplenia, isolated congenital, 271400
RRAS	SHORT STATURE/SKELETAL DYSPLASIA	116.3	89.4	81.6	No OMIM phenotype DD Noonan
RRAS2	MENDELIOME	73.4	86.9	70.3	Ovarian carcinoma, 0
RRM1	MITOCHONDRIAL DISORDERS	132.4	99.8	98.6	
RRM2B	EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	128.6	99.7	97.5	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RS1	VISION DISORDERS MENDELIOME	60	97.8	88.3	Retinoschisis, 312700
RSPH1	CILIO MENDELIOME PRECONCEPTION SCREENING	146.1	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	CILIO MENDELIOME PRECONCEPTION SCREENING	132.5	99.7	97.5	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	CILIO MENDELIOME PRECONCEPTION SCREENING	139	98.3	96.5	Ciliary dyskinesia, primary, 11, 612649
RSPH9	CILIO PRIMARY IMMUNODEFICIENCIES	127.4	100	99.6	Ciliary dyskinesia, primary, 12, 612650

	MENDELIOME PRECONCEPTION SCREENING				
RSPO1	SKIN DISORDERS DSD MENDELIOME PRECONCEPTION SCREENING	109.7	100	100	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	MENDELIOME	127.1	98.9	95.2	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	107.2	100	100	Anonychia congenita, 206800
RSPRY1	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	168.9	100	99.9	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RTEL1	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RTN2	MOVEMENT DISORDERS MENDELIOME	104.8	99.2	96.7	Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	98.1	99.9	99.1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.5	97.2	94.7	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	104.1	98	97.5	?Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME HEREDITARY CANCER	92	97.2	89.7	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399

RUNX2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	106.4	72.3	72.2	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
RUNX3		66.9	97.7	86.7	
RUSC2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	182.3	100	99.9	Mental retardation, autosomal recessive 61, 617773
RYR1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	120.7	96.8	93.7	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
RYR2	HEART PANEL MENDELIOME RITME	142.2	99.7	98.4	Arrhythmogenic right ventricular dysplasia 2, 600996 Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772
S1PR2	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	278	97.4	92.7	Deafness, autosomal recessive 68, 610419
SAA2		140.1	97.1	90.6	
SACS	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	154.5	100	99.7	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.3	100	99.9	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	138.5	99.3	98.4	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL2	MENDELIOME	138.9	100	100	?Coloboma, ocular, autosomal recessive, 216820
SALL4	CRANIOFACIAL ANOMALIES RENAL DISORDERS MENDELIOME	147.5	97.6	96.3	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SAMD11	VISION DISORDERS	63.2	82.8	75.2	No OMIM phenotype ?Autism spectrum disorder (Chapman (2015) Hum Genet 134, 1055) Adult onset Retinitis Pigmentosa
SAMD12	MENDELIOME	161.6	100	99.9	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	BONE MARROW FAILURE	159.1	99.9	99.3	MIRAGE syndrome, 617053

	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER				Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	MOVEMENT DISORDERS BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	165.7	100	99.9	Ataxia-pancytopenia syndrome, 159550
SAMHD1	MOVEMENT DISORDERS SKIN DISORDERS EPILEPSY PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	127.9	99.6	96.6	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	MENDELIOME PRECONCEPTION SCREENING	125	89.8	88.9	Chylomicron retention disease, 246700
SARDH	METABOLIC DISORDERS	120.4	95	92.8	[Sarcosinemia], 268900
SARS	INTELLECTUAL DISABILITY MENDELIOME	113.7	100	99.7	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	104.8	94.8	92.7	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	SKIN DISORDERS	122.3	99.7	98.4	No OMIM phenotype Disseminated superficial actinic porokeratosis (Zhang (2005) Br J Dermatol 152,658)
SASH1	SKIN DISORDERS	143.1	98.9	96.8	No OMIM phenotype Lentiginosis, autosomal dominant (Shellman (2015) J Invest Dermatol 135,3192) Pigmentation defects, palmoplantar keratoderma and skin carcinoma (Courcet (2015) Eur J Hum Genet 23,957)
SASS6	MENDELIOME	62.9	97.6	84.1	?Microcephaly 14, primary, autosomal recessive, 616402
SAT1	SKIN DISORDERS METABOLIC DISORDERS	141.1	100	99.2	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SATB2	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	110.5	98.5	93.4	Glass syndrome, 612313
SBDS	BONE MARROW FAILURE	212.3	100	99.9	Shwachman-Diamond syndrome, 260400

	PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER				{Aplastic anemia, susceptibility to}, 609135
SBF1	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	107.9	98.5	96.5	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	117	99.6	96.8	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	VISION DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	198.4	100	99.2	Lathosterolosis, 607330
SCAPER	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135.9	96	93.6	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	EPILEPSY METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.2	100	99.9	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	82.5	85.2	73.6	Van den Ende-Gupta syndrome, 600920
SCLT1	CILIO	69.2	90.5	80	OFD type IX
SCN10A	SKIN DISORDERS HEART PANEL NEUROPATHIES MENDELIOME	165.3	100	99.5	Episodic pain syndrome, familial, 2, 615551
SCN11A	MOVEMENT DISORDERS SKIN DISORDERS NEUROPATHIES MENDELIOME	138.1	99.2	97.6	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	EPILEPSY INTELLECTUAL DISABILITY	135.2	99.6	98	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208

	MENDELIOME				Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	EPILEPSY HEART PANEL INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING RITME	168.3	97.1	96.1	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350
SCN2A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	156.7	99	96.4	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN2B	HEART PANEL MENDELIOME	185.8	100	100	Atrial fibrillation, familial, 14, 615378
SCN3A	INTELLECTUAL DISABILITY MENDELIOME	166	99.3	97.5	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN3B	HEART PANEL MENDELIOME RITME	147.3	100	100	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4A	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	214	99.9	99.5	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SCN4B	HEART PANEL MENDELIOME RITME	77.5	100	97.9	Atrial fibrillation, familial, 17, 611819 Long QT syndrome-10, 611819
SCN5A	CARDIO HEART PANEL MENDELIOME RITME	169.4	99	99	Atrial fibrillation, familial, 10, 614022 Brugada syndrome 1, 601144 Cardiomyopathy, dilated, 1E, 601154 Heart block, nonprogressive, 113900 Heart block, progressive, type IA, 113900 Long QT syndrome-3, 603830 Sick sinus syndrome 1, 608567 Ventricular fibrillation, familial, 1, 603829 {Sudden infant death syndrome, susceptibility to}, 272120
SCN8A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	198.3	100	99.7	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SCN9A	SKIN DISORDERS NEUROPATHIES	146.5	98.5	97	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020

	MENDELIOME PRECONCEPTION SCREENING				Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder,, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCNN1A	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	134.3	99	96.3	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.9	100	99.8	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.4	99.7	97.1	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	109.6	97.9	94.3	Mitochondrial complex IV deficiency, 220110
SCO2	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	113.1	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	106.9	99.6	96.5	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.2	98.6	96.3	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.9	99.8	97.4	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	HEART PANEL INTELLECTUAL DISABILITY	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000

	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING HEREDITARY CANCER				Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	42.5	100	96.2	Mitochondrial complex II deficiency, 252011
SDHAF2	MENDELIOME HEREDITARY CANCER	144.3	94.7	94.3	Paragangliomas 2, 601650
SDHB	MENDELIOME MITOCHONDRIAL DISORDERS HEREDITARY CANCER	120.3	100	99.3	Gastrointestinal stromal tumor, 606444 Paragangliolas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDHC	MENDELIOME HEREDITARY CANCER	100.1	99.8	96.8	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	MENDELIOME MITOCHONDRIAL DISORDERS HEREDITARY CANCER	48.4	55.2	50.4	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SDR9C7	SKIN DISORDERS MENDELIOME	198.3	100	100	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	MENDELIOME PRECONCEPTION SCREENING	121.8	98	94.6	Craniolenticulosutural dysplasia, 607812
SEC23B	SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	161.1	97.5	96.4	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	136.9	99.9	98.6	Cole-Carpenter syndrome 2, 616294
SEC61A1	RENAL DISORDERS MENDELIOME	132.9	100	100	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SEC63	MENDELIOME	60.8	85.4	75.7	Polycystic liver disease 2, 617004
SECISBP2	MENDELIOME PRECONCEPTION SCREENING	122.5	98.3	95.3	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	METABOLIC DISORDERS MENDELIOME	140.5	100	100	Extraoral halitosis due to MTO deficiency, 618148
SELENON	 MENDELIOME	111.7	85.2	83.3	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310

	PRECONCEPTION SCREENING MUSCLE DISORDERS				
SELENOP		166.5	91.7	86.1	
SEMA3A	HH	182	100	100	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME	142.6	99.9	99	?CHARGE syndrome, 214800
SEMA4A	VISION DISORDERS MENDELIOME HEREDITARY CANCER	127.5	99.9	98.9	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	159.3	100	100	Pontocerebellar hypoplasia type 2D, 613811
SEPT12	MENDELIOME	105	97.4	93.8	Spermatogenic failure 10, 614822
SEPT9	NEUROPATHIES MENDELIOME	118.7	99.7	96.8	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related, 0 Ovarian carcinoma, 0
SERAC1	MOVEMENT DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	MENDELIOME PRECONCEPTION SCREENING	141.1	100	99.9	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
SERPINA3	MENDELIOME	152.8	100	100	Alpha-1-antichymotrypsin deficiency, 0 Cerebrovascular disease, occlusive, 0
SERPINA6	MENDELIOME PRECONCEPTION SCREENING	190.9	100	100	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	164.1	95.9	95.9	?Deafness, autosomal recessive 91, 613453
SERPINB7	SKIN DISORDERS MENDELIOME	127.4	100	99.5	Palmoplantar keratoderma, Nagashima type, 615598

	PRECONCEPTION SCREENING				
SERPINB8	SKIN DISORDERS MENDELIOME	151.6	95	95	Peeling skin syndrome 5, 617115
SERPINC1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	143.1	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	181.8	100	100	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	155	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	101.8	100	99.4	Osteogenesis imperfecta, type VI, 613982
SERPINF2	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	143.8	99.9	99.3	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	97.9	97.3	92.6	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	183.7	100	99.9	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SERPINI1	MENDELIOME	105.2	99.9	96.9	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	INTELLECTUAL DISABILITY MENDELIOME	55.4	89.9	79.4	Mental retardation, autosomal dominant 58, 618106
SETBP1	INTELLECTUAL DISABILITY MENDELIOME	151.7	97.6	96.1	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150
SETD1A	INTELLECTUAL DISABILITY	119.2	98.6	96.6	No OMIM phenotype Schizophrenia (Takata (2014) Neuron 82, 723)
SETD1B	INTELLECTUAL DISABILITY	133.3	96.5	94.6	No OMIM phenotype
SETD2	INTELLECTUAL DISABILITY MENDELIOME	147.8	100	99.6	Luscan-Lumish syndrome, 616831
SETD5	INTELLECTUAL DISABILITY MENDELIOME	184.5	100	99.7	Mental retardation, autosomal dominant 23, 615761
SETX	ALS MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	163.2	99.9	99.1	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002

SF3B1	IRON DISORDERS MENDELIOME	128.7	99.3	97.5	Myelodysplastic syndrome, somatic, 614286
SF3B4	CRANIOFACIAL ANOMALIES MENDELIOME	89.5	99.8	97.7	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	MENDELIOME PRECONCEPTION SCREENING	164.1	100	98.5	Pyle disease, 265900
SFTPA1	HEREDITARY CANCER	175.5	99.8	98.3	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	MENDELIOME HEREDITARY CANCER	167.9	100	99.1	Pulmonary fibrosis, idiopathic, 178500
SFTPB	MENDELIOME PRECONCEPTION SCREENING	99.4	99.9	98.7	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	MENDELIOME	102.6	99.8	95.6	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	131.7	100	99.1	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	HEART PANEL MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	147.3	100	99.7	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	HEART PANEL MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	154.2	96.6	94.2	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	HEART PANEL MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	94.8	100	99.4	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	MOVEMENT DISORDERS MENDELIOME	88.9	93.7	90	Dystonia-11, myoclonic, 159900
SGCG	HEART PANEL MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	138.7	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	MENDELIOME PRECONCEPTION SCREENING	94.8	98	94.3	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	SKIN DISORDERS RENAL DISORDERS MENDELIOME	164.1	100	100	Nephrotic syndrome, type 14, 617575
SGSH	VISION DISORDERS SHORT STATURE/SKELETAL DYSPLASIA	129	95.1	93.6	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900

	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING				
SH2B3	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	97.5	90.7	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME	104.7	89.9	89.4	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	CRANIOFACIAL ANOMALIES PRIMARY IMMUNODEFICIENCIES MENDELIOME	110.9	91.4	91.4	Cherubism, 118400
SH3PXD2B	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	140.2	100	99.8	Frank-ter Haar syndrome, 249420
SH3TC2	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	121.3	100	99.7	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK2	INTELLECTUAL DISABILITY	128.1	100	99.8	{Autism susceptibility 17}, 613436
SHANK3	INTELLECTUAL DISABILITY MENDELIOME	84.8	81.2	73.5	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	VISION DISORDERS CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	117.5	99	94	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SHOC2	SKIN DISORDERS HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SHOX	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	29.1	73.5	61.4	Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300 Short stature, idiopathic familial, 300582
SHROOM3	CONGENITAL HEART DISEASE HEART PANEL	137.3	99.9	98.9	No OMIM phenotype Heterotaxy (Tariq (2011) Genome Biol 12,R91) ?Neural tube defects (Lemay (2015) J Med Genet 52,493) {Leukaemia risk,association with} (Rudd (2006) Blood 108,638)
SHROOM4	INTELLECTUAL DISABILITY	100.8	99.8	98.2	Stocco dos Santos X-linked mental retardation syndrome, 300434

	MENDELIOME				
SI	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	98.1	96.2	87.1	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	ALS MENDELIOME	148.5	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	85.6	97	92.4	Epileptic encephalopathy, early infantile, 30, 616341
SIK3	MENDELIOME	117.2	98	96.8	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	154.4	99.8	98	Marinesco-Sjogren syndrome, 248800
SIM1	MENDELIOME	151.4	100	99.8	Obesity, severe, 601665
SIN3A	INTELLECTUAL DISABILITY MENDELIOME	137.9	100	99.4	Witteveen-Kolk syndrome, 613406
SIPA1L3	VISION DISORDERS MENDELIOME	140.8	99.7	98.6	?Cataract 45, 616851
SIX1	CRANIOFACIAL ANOMALIES HEARING IMPAIRMENT MENDELIOME	117.3	99.7	97.6	Branchiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	145.3	100	98.9	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	HEARING IMPAIRMENT RENAL DISORDERS MENDELIOME	43.8	88.3	76.1	Branchiootorenal syndrome 2, 610896
SIX6	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	228.6	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	 CRANIOFACIAL ANOMALIES SKIN DISORDERS HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	85.3	96.4	90.8	Shprintzen-Goldberg syndrome, 182212
SKIV2L	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	149.1	100	99.8	Trichohepatoenteric syndrome 2, 614602

SLC10A2	MENDELIOME PRECONCEPTION SCREENING	151	100	100	Bile acid malabsorption, primary, 613291
SLC10A7	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS	110	100	98.9	No OMIM phenotype Ashikov et al. (PMID: 29878199) en Dubail et al (PMID: 30082715)
SLC11A2	IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.2	100	99.3	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	172.7	99.8	99.1	Bartter syndrome, type 1, 601678
SLC12A3	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.3	100	100	Gitelman syndrome, 263800
SLC12A5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.9	85.4	81.6	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	141.8	100	99.9	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	164.1	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	EPILEPSY METABOLIC DISORDERS MENDELIOME	157.1	99.9	98.8	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	VISION DISORDERS RENAL DISORDERS MENDELIOME	164.4	100	99.9	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	60.3	92.8	82.1	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	119.6	96.8	92.9	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	HEARING IMPAIRMENT	132.7	100	100	Deafness, autosomal dominant 25, 605583

	MENDELIOME				
SLC17A9	SKIN DISORDERS MENDELIOME	111.5	95.6	95.6	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	MENDELIOME	126.5	100	99.2	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	MENDELIOME	276.6	100	100	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	BONE MARROW FAILURE IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	119.5	99.8	97.8	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	186.4	100	99.9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	174.2	100	99.9	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC1A2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	128	99.9	99.4	Epileptic encephalopathy, early infantile, 41, 617105
SLC1A3	MOVEMENT DISORDERS MENDELIOME	121.9	100	99.8	Episodic ataxia, type 6, 612656
SLC1A4	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	156.5	98.9	94.9	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	MOVEMENT DISORDERS MENDELIOME PARK	119	99.7	97.3	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	105.7	100	99.7	Hypouricemia, renal, 220150
SLC22A18	MENDELIOME	85.1	92.8	81.7	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A4	HEARING IMPAIRMENT	119.1	99.8	98.2	{Rheumatoid arthritis, susceptibility to}, 180300
SLC22A5	HEART PANEL METABOLIC DISORDERS MENDELIOME	153.3	100	100	Carnitine deficiency, systemic primary, 212140

	PRECONCEPTION SCREENING				
SLC24A1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	218.5	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	126.8	99.6	97.3	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	114.5	99.6	97.7	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC25A1	EPILEPSY METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	71	92.2	87	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	MITOCHONDRIAL DISORDERS	70.2	76.5	68.6	
SLC25A12	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	150.5	99.8	98.4	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	110.7	95.7	92.3	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	192.5	98.8	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	88.6	99.9	98.3	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A2	HEARING IMPAIRMENT	219.9	100	100	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/29287867
SLC25A20	HEART PANEL METABOLIC DISORDERS MENDELIOME	110.3	100	99.7	Carnitine-acylcarnitine translocase deficiency, 212138

	PRECONCEPTION SCREENING				
SLC25A21	METABOLIC DISORDERS MITOCHONDRIAL DISORDERS	114	100	98.9	No OMIM phenotype ?Synpolydactyly (Meyertholen (2012) Mol Syndromol 3 25)
SLC25A22	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	108.7	99.5	96.9	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	115.8	98.6	96.3	Fontaine progeroid syndrome, 612289
SLC25A26	MENDELIOME PRECONCEPTION SCREENING	98	98.9	96	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	139	99.8	97.6	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	117	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A37	IRON DISORDERS	194	100	100	No OMIM phenotype ?anemia and disruptions in ISC biogenesis, inhibition protoporphyrin biosynthesis (Shaw et al. (2006) erythropoietic protophyria (Wang et al. (2011))
SLC25A38	BONE MARROW FAILURE IRON DISORDERS METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	111.4	99.8	98.1	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	HEART PANEL MENDELIOME MITOCHONDRIAL DISORDERS MUSCLE DISORDERS	134.1	100	100	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A42	METABOLIC DISORDERS MITOCHONDRIAL DISORDERS	116.9	97.8	94.2	No OMIM phenotype Mitochondrial myopathy (Shamseldin (2016) Hum Genet 135,21)
SLC25A46	VISION DISORDERS NEUROPATHIES MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	205.7	95.9	87.3	Neuropathy, hereditary motor and sensory, type VIB, 616505

SLC26A1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	139.8	100	99.9	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	CRANIOFACIAL ANOMALIES SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	233.2	100	100	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	156.1	99.9	98.9	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	123.3	99.9	99.1	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	150.9	98.7	95.8	?Deafness, autosomal recessive 61, 613865
SLC26A8	MENDELIOME	138.9	100	99.3	Spermatogenic failure 3, 606766
SLC27A4	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	155.8	99.6	97.9	Ichthyosis prematurity syndrome, 608649
SLC29A3	SKIN DISORDERS HEARING IMPAIRMENT PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	203.6	99.9	99.5	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	190.1	92.9	92.8	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	SKIN DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	166.4	97.7	97.6	Arterial tortuosity syndrome, 208050
SLC2A2	METABOLIC DISORDERS RENAL DISORDERS	178.4	100	99.9	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853

	MENDELIOME PRECONCEPTION SCREENING				
SLC2A9	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	119	99.2	96.2	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC30A10	MOVEMENT DISORDERS METABOLIC DISORDERS MENDELIOME PARK PRECONCEPTION SCREENING	164.4	100	100	Hypermanganesemia with dystonia 1, 613280
SLC30A2	MENDELIOME	146.1	100	99.6	Zinc deficiency, transient neonatal, 608118
SLC30A9	MENDELIOME	81.2	96	88.3	?Birk-Landau-Perez syndrome, 617595
SLC33A1	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	140.9	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC34A1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	153.2	100	99.5	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	MENDELIOME PRECONCEPTION SCREENING	169.1	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	105.6	98.9	94.5	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124	99.9	97.7	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	108.8	99.7	96.8	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	50.2	78.1	71.9	?Arthrogryposis, mental retardation, and seizures, 615553

SLC35C1	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	230.2	99.9	98.4	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	115.4	95.7	90.4	Schneckenbecken dysplasia, 269250
SLC36A2	RENAL DISORDERS MENDELIOME	123.9	100	100	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC38A8	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	76.4	99.4	95.8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	114.8	99.8	98	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARK PRECONCEPTION SCREENING	107.7	99.8	97.9	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	81.8	99.2	96	Acrodermatitis enteropathica, 201100
SLC39A5	VISION DISORDERS MENDELIOME	119.3	100	99.2	Myopia 24, autosomal dominant, 615946
SLC39A8	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	128.5	100	99.7	Congenital disorder of glycosylation, type IIn, 616721

	PRECONCEPTION SCREENING				
SLC3A1	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	162.8	100	99.5	Cystinuria, 220100
SLC40A1	IRON DISORDERS MENDELIOME	155.7	99.9	99.4	Hemochromatosis, type 4, 606069
SLC41A1	RENAL DISORDERS	149.8	100	99.9	No OMIM phenotype Nephrolithiasis-like phenotype (Hurd (2013) J Am Soc Nephrol 24,967) ?Parkinson disease (Yan (2011) Int J Neurosci 121,632)
SLC44A4	HEARING IMPAIRMENT MENDELIOME	122.9	100	99.9	?Deafness, autosomal dominant 72, 617606
SLC45A1	MENDELIOME	120.7	100	99.2	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	148.1	100	99.9	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	BONE MARROW FAILURE IRON DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	106	99.4	96.4	Folate malabsorption, hereditary, 229050
SLC4A1	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	140	100	99.9	Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653 [Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Malaria, resistance to], 611162
SLC4A11	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	153	100	99.7	Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400 Corneal endothelial dystrophy, autosomal recessive, 217700
SLC4A4	SKIN DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	122.3	99	97.1	Renal tubular acidosis, proximal, with ocular abnormalities, 604278

	PRECONCEPTION SCREENING				
SLC52A1	METABOLIC DISORDERS MENDELIOME	219.3	100	100	Riboflavin deficiency, 615026
SLC52A2	MOVEMENT DISORDERS VISION DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	MOVEMENT DISORDERS NEUROPATHIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	119.6	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	140	100	100	Glucose/galactose malabsorption, 606824
SLC5A2	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	118.7	100	100	Renal glucosuria, 233100
SLC5A5	MENDELIOME PRECONCEPTION SCREENING	93.9	99.7	96.8	Thyroid dyshormonogenesis 1, 274400
SLC5A7	 NEUROPATHIES MENDELIOME	117.1	100	99.9	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	143.6	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A17	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	189.8	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	149.1	100	99.3	HEART PANELnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600

	PRECONCEPTION SCREENING				
SLC6A2	MENDELIOME	149.9	100	99.8	?Orthostatic intolerance, 604715
SLC6A20	RENAL DISORDERS MENDELIOME	178.6	100	99.9	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARK PRECONCEPTION SCREENING	145.7	100	99.8	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC6A5	MENDELIOME PRECONCEPTION SCREENING	136.5	100	99.7	Hyperekplexia 3, 614618
SLC6A8	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	56.5	89.8	79.1	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	MENDELIOME	161.2	100	99.4	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	191.9	100	100	Retinitis pigmentosa 68, 615725
SLC7A5		89.6	98.3	92.7	
SLC7A7	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.9	100	99.9	Lysinuric protein intolerance, 222700
SLC7A9	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.5	99.9	99	Cystinuria, 220100
SLC8A1		198.2	99.7	99	
SLC9A1	MOVEMENT DISORDERS MENDELIOME	160.9	100	100	?Lichtenstein-Knorr syndrome, 616291
SLC9A3	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	147.1	98.7	96.9	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	RENAL DISORDERS MENDELIOME	111.5	99.5	96	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	EPILEPSY	104.2	97.6	91.3	Mental retardation, X-linked syndromic, Christianson type, 300243

	INTELLECTUAL DISABILITY MENDELIOME				
SLC9A7	INTELLECTUAL DISABILITY	102.9	95.7	88.5	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30335141
SLCO1B1	METABOLIC DISORDERS MENDELIOME	46	92.8	84.4	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	METABOLIC DISORDERS MENDELIOME	48.9	94.9	79.5	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	110.5	100	99.6	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	SHORT STATURE/SKELETAL DYSPLASIA	169.1	99.8	97.7	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95)
SLFN14	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	193.1	100	100	Bleeding disorder, platelet-type, 20, 616913
SLIT3	RENAL DISORDERS	146.8	97.3	95.2	No OMIM phenotype Major depressive disorder (Glessner (2010) PLoS One 5, e15463) ?Autism spectrum disorder (Bi (2012) Hum Mutat 33, 1635) ?Glioma and Hodgkin lymphoma (Ritter (2015) Genet Med 17, 831) ?Schizophrenia (Gulsuner (2013) Cell 154,
SLITRK1	MENDELIOME	132.6	100	100	?Trichotillomania, 613229 Tourette syndrome, 137580
SLITRK6	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	206.9	100	100	Deafness and myopia, 221200
SLMAP	HEART PANEL	121.2	93.5	85.2	No OMIM phenotype Brugada syndrome (Ishikawa (2012) Circ Arrhythm Electrophysiol epub)
SLN		46.3	100	99.2	
SLURP1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	97.1	99.8	96.2	Meleda disease, 248300
SLX4	BONE MARROW FAILURE SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	114.2	100	99.8	Fanconi anemia, complementation group P, 613951
SMAD1	HEART PANEL	184	99.9	99.7	No OMIM phenotype
SMAD2	HEART PANEL	151.7	99.9	99.1	No OMIM phenotype Congenital heart disease (Zaidi (2013) Nature 498,220) Arterial s and dissections (Micha (2015) Hum Mutat 36,1145)

					Holoprosencephaly (Roessler (2008) Am J Hum Genet 83,18)
SMAD3	SKIN DISORDERS HEART PANEL MENDELIOME	131.7	99.9	99.2	Loeys-Dietz syndrome 3, 613795
SMAD4	HEART PANEL SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD6	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	100.5	80	72	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMAD9	HEART PANEL MENDELIOME HEREDITARY CANCER	132.8	100	100	Pulmonary hypertension, primary, 2, 615342
SMARCA1	INTELLECTUAL DISABILITY	82.9	98.1	90.1	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/?term=29249292
SMARCA2	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	113.8	95.7	93.7	Nicolaides-Baraitser syndrome, 601358
SMARCA4	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	143.8	100	99.5	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCAD 1	SKIN DISORDERS MENDELIOME	85.5	99.6	96.1	Adermatoglyphia, 136000 Basan syndrome, 129200
SMARCAL1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	134.6	100	99.9	Schimke immunoosseous dysplasia, 242900
SMARCB1	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	214.3	100	100	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322

	HEREDITARY CANCER				{Schwannomatosis-1, susceptibility to}, 162091
SMARCC2	INTELLECTUAL DISABILITY	105.7	99.3	97.1	No OMIM phenotype ?Ivemark syndrome (Carss (2014) Hum Mol Genet 23,3269) ?Autism (Neale (2012) Nature 485,242)
SMARCD2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	92.9	87	85.7	Specific granule deficiency 2, 617475
SMARCE1	INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	73.6	96.5	86.8	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SMC1A	CRANIOFACIAL ANOMALIES EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	99.4	99.9	98.8	Cornelia de Lange syndrome 2, 300590
SMC3	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	81.4	93.8	87.6	Cornelia de Lange syndrome 3, 610759
SMCONGENITAL HEART DISEASE1	MENDELIOME MUSCLE DISORDERS	91.3	98.1	92.3	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMG9	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	101.8	100	99.9	Heart and brain malformation syndrome, 616920
SMN1	MENDELIOME PRECONCEPTION SCREENING	112.7	99.8	96.5	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SMO	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	149.2	96.5	93.4	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.8	99.5	97.3	Microphthalmia with limb anomalies, 206920
SMOC2	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	91.5	75.4	72.6	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY	123.5	99.6	97.9	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616

	MENDELIOME PRECONCEPTION SCREENING				
SMPX	HEARING IMPAIRMENT MENDELIOME	68.5	99.9	95.5	Deafness, X-linked 4, 300066
SMS	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	67.8	88.3	73.9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SMYD2		105.3	98.6	94.5	
SNAI2	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	129.8	100	99.8	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP25	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	133.7	100	99.9	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	153.5	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	MOVEMENT DISORDERS MENDELIOME PARK	129.8	100	100	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	MENDELIOME	79.3	100	99.9	Dementia, Lewy body, 127750
SNIP1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	139.3	99.2	96.7	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	MOVEMENT DISORDERS MENDELIOME	123456	123456	123456	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	VISION DISORDERS MENDELIOME	161.1	100	99.6	Retinitis pigmentosa 33, 610359
SNRPB	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	75.2	99.8	97.4	Cerebrocostomandibular syndrome, 117650
SNRPE	SKIN DISORDERS MENDELIOME	79	98.5	89.1	Hypotrichosis 11, 615059
SNRPN	INTELLECTUAL DISABILITY MENDELIOME	116.8	99.4	95	Prader-Willi syndrome, 176270
SNTA1	HEART PANEL MENDELIOME	97	82.3	77.3	Long QT syndrome 12, 612955

	RITME				
SNX10	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	118.9	96.2	96.1	Osteopetrosis, autosomal recessive 8, 615085
SNX14	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	70.1	95.2	82.9	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.7	92.9	85.4	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOCS4	PRIMARY IMMUNODEFICIENCIES	262.8	99.9	99.3	No OMIM phenotype Autoimmunity (Arts (2015) J Intern Med epub,epub)
SOD1	ALS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	161.9	100	100	Amyotrophic lateral sclerosis 1, 105400
SOHLH1	MENDELIOME	87	97.9	90	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	INTELLECTUAL DISABILITY MENDELIOME	159.7	98.4	94.4	ZTTK syndrome, 617140
SORT1		113.5	88.1	87.6	[Low density lipoprotein cholesterol level QTL6], 613589
SOS1	SKIN DISORDERS HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	97.1	98.5	92.8	Noonan syndrome 9, 616559
SOST	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	112.6	100	99.6	Craniodiaphyseal dysplasia, autosomal dominant, 122860 Sclerosteosis 1, 269500 Van Buchem disease, 239100
SOX10	MOVEMENT DISORDERS	65.8	98.2	91.3	PCWH syndrome, 609136

	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT HH NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME				Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX11	INTELLECTUAL DISABILITY MENDELIOME	118.2	99.7	96.8	Mental retardation, autosomal dominant 27, 615866
SOX17	 RENAL DISORDERS MENDELIOME	70.8	99.6	94.6	Vesicoureteral reflux 3, 613674
SOX18	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	21.2	62.6	48.8	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	VISION DISORDERS SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	128.8	98.3	93.1	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	DSD SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	37.7	86.4	71.5	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX5	INTELLECTUAL DISABILITY MENDELIOME	107.3	99.1	96.2	Lamb-Shaffer syndrome, 616803
SOX6	CRANIOFACIAL ANOMALIES	102.7	99.7	97.9	No OMIM phenotype Developmental delay and spinal syrinx (Scott (2014) J Child Neurol 29, NP164) Dystonia, dopa-responsive (Ebrahimi-Fakhari (2015) Pediatr Neurol 52,115) ?Craniosynostosis (Tagariello (2006) J Med Genet 43,534)
SOX9	CRANIOFACIAL ANOMALIES DSD SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	134	97.8	93.8	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SP110	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	121.6	100	99.5	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SP7	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	159.1	99.9	99.3	Osteogenesis imperfecta, type XII, 613849
SPAG1	CILIO	87.3	96.3	88.2	Ciliary dyskinesia, primary, 28, 615505

	MENDELIOME PRECONCEPTION SCREENING				
SPARC	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	161	100	100	Osteogenesis imperfecta, type XVII, 616507
SPART	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	132.4	99.8	98.2	Troyer syndrome, 275900
SPAST	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	63.8	93.1	81.9	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	MENDELIOME	142.2	99.7	96.6	?Spermatogenic failure 6, 102530
SPATA5	HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	132	99.9	99.2	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	VISION DISORDERS CILIO MENDELIOME PRECONCEPTION SCREENING	119.6	97.8	90.8	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
SPECC1L	CRANIOFACIAL ANOMALIES SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	157.9	100	100	?Facial clefting, oblique, 1, 600251 Hypertelorism, Teebi type, 145420 Opitz GBBB syndrome, type II, 145410
SPEG	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	100.9	93.3	86.5	Centronuclear myopathy 5, 615959
SPG11	ALS MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.2	99.2	96.9	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	121.4	98.6	94.8	Mast syndrome, 248900
SPG7	MOVEMENT DISORDERS MENDELIOME	119.2	93.3	92.4	Spastic paraplegia 7, autosomal recessive, 607259

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
SPINK1	MENDELIOME HEREDITARY CANCER	80.2	100	99.4	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK2	MENDELIOME	83.4	99.1	96.8	?Spermatogenic failure 29, 618091
SPINK5	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	145	99.4	96.5	Netherton syndrome, 256500
SPINT2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	71.5	97.8	84.2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPOCK1	INTELLECTUAL DISABILITY	118.7	100	99.8	No OMIM phenotype Developmental delay and microcephaly (Dhamija (2014) Eur J Med Genet 57,181)
SPP2	VISION DISORDERS	144.1	100	100	No OMIM phenotype Retinitis pigmentosa (Li (2015) Sci Rep 5,14867) ?Autism (Neale (2012) Nature 485,242)
SPR	MOVEMENT DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	166.5	98.9	90	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	164.3	98.7	96.7	Legius syndrome, 611431
SPRTN	MENDELIOME PRECONCEPTION SCREENING	167.8	100	100	Ruijs-Aalfs syndrome, 616200
SPRY4	SKIN DISORDERS HH MENDELIOME	138.7	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	MENDELIOME PRECONCEPTION SCREENING	119.4	99.8	98.9	Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140 Spherocytosis, type 3, 270970
SPTAN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	125.5	99.1	98.6	Epileptic encephalopathy, early infantile, 5, 613477

SPTB	MENDELIOME PRECONCEPTION SCREENING	148	100	99.9	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649
SPTBN2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	118	99.9	99.3	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	NEUROPATHIES MENDELIOME	83.9	96.6	89.1	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	NEUROPATHIES METABOLIC DISORDERS MENDELIOME	115.5	99	93.9	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	NEUROPATHIES METABOLIC DISORDERS MENDELIOME	160.2	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SQSTM1	ALS MENDELIOME MITOCHONDRIAL DISORDERS HEREDITARY CANCER	109.1	98.6	94.5	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
SRC	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	105.2	99.8	97.3	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
SRCAP	DSD SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME	153.9	99.8	99.1	Floating-Harbor syndrome, 136140
SRD5A2	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	77.6	100	96.4	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135.9	100	99.7	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRF		184.2	94.9	89.1	
SRI	HEART PANEL	114.3	97.9	88.9	No OMIM phenotype
SRP72	BONE MARROW FAILURE MENDELIOME	70	93.1	84	Bone marrow failure syndrome 1, 614675
SRPX2	INTELLECTUAL DISABILITY MENDELIOME	81.8	100	98.5	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
SRY	DSD	46.1	50	50	46XX sex reversal 1, 400045

	MENDELIOME				46XY sex reversal 1, 400044
SSR4	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	89.8	100	98.8	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	MENDELIOME	155.8	100	99.2	Somatostatin analog, resistance to, 0
SSX1	MENDELIOME	103.4	80.8	76.5	?Sarcoma, synovial, 300813
SSX2	MENDELIOME	69.8	63	57.8	?Sarcoma, synovial, 300813
ST14	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	154	98	97	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	144.5	100	99.9	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	SKIN DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	121.9	84.4	84.2	Salt and pepper developmental regression syndrome, 609056
STAC3	MENDELIOME PRECONCEPTION SCREENING	120	100	99.9	Myopathy, congenital, Baily-Bloch, 255995
STAG1	INTELLECTUAL DISABILITY MENDELIOME	104.4	98.8	95.5	Mental retardation, autosomal dominant 47, 617635
STAG3	MENDELIOME	112.3	93.5	92.8	Premature ovarian failure 8, 615723
STAMBP	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	112.3	99.3	96.5	Microcephaly-capillary malformation syndrome, 614261
STAR	DSD METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	124	100	100	Lipoid adrenal hyperplasia, 201710
STARD13		157.5	99.7	98.8	
STAT1	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	126.2	98	95.8	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	116	100	99.9	Immunodeficiency 44, 616636

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
STAT3	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	119.5	99.9	99	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT4	PRIMARY IMMUNODEFICIENCIES	144.2	98.7	97.1	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	130.6	99.7	97.2	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STAT6	PRIMARY IMMUNODEFICIENCIES	119.3	100	99.7	No OMIM phenotype {Schistosomiasis infection, association with} (He (2008) Genes Immun 9, 195) {Atopic asthma, association with} (Gao (2004) J Med Genet 41,535)
STEAP3	IRON DISORDERS MENDELIOME	199	100	99.8	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STIL	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	157.2	99.8	98.6	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	BONE MARROW FAILURE SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK11	BRSTKNK SKIN DISORDERS MENDELIOME HEREDITARY CANCER	111.9	99.7	95.8	Melanoma, malignant, somatic, 0 Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	138.9	100	99.3	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	BONE MARROW FAILURE MENDELIOME	94.4	99.9	99.5	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	MENDELIOME	177.7	89.6	89.5	Preeclampsia/eclampsia 4, 609404
STRA6	VISION DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS	116.5	100	99.9	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186

	MENDELIOME PRECONCEPTION SCREENING				
STRADA	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	134.1	100	98.9	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	119	99.9	98.4	Deafness, autosomal recessive 16, 603720
STS	SKIN DISORDERS METABOLIC DISORDERS MENDELIOME	91.6	99.7	97.8	Ichthyosis, X-linked, 308100
STT3A	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	156.2	100	100	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	125.1	99.2	96	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	176.1	100	98.9	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	311.4	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	RENAL DISORDERS MENDELIOME	140.6	99.8	97.8	Pseudohypoparathyroidism, type IB, 603233
STX1B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	152.2	100	98.4	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	124.5	96.8	96.8	Epileptic encephalopathy, early infantile, 4, 612164
STXBP2	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	102.3	88.9	83.8	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME	64.9	93.3	82.8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING				
SUCLG1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	101.3	99.6	95.4	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	METABOLIC DISORDERS MITOCHONDRIAL DISORDERS	57.8	91.1	78.7	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SUFU	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	122.6	99.9	99	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUGCT	METABOLIC DISORDERS MENDELIOME	129.1	94.1	87.6	Glutaric aciduria III, 231690
SULF1	SHORT STATURE/SKELETAL DYSPLASIA	164.2	100	99.9	No OMIM phenotype Mesomelia-synostoses syndrome (Isidor (2010) Am J Hum Genet 87,95) ?Hyperinsulinism (Proverbio (2013) PLoS One 8,e68740)
SULT2B1	SKIN DISORDERS MENDELIOME	111.4	100	100	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	MOVEMENT DISORDERS SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.3	98.6	91.1	Multiple sulfatase deficiency, 272200
SUMO1	CRANIOFACIAL ANOMALIES MENDELIOME	17.6	58.8	37	?Orofacial cleft 10, 613705
SUN5	MENDELIOME	115	99.9	99.1	Spermatogenic failure 16, 617187
SUOX	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	212.6	100	100	Sulfite oxidase deficiency, 272300
SURF1	NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	96.2	88.3	88.3	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000

SUZ12	INTELLECTUAL DISABILITY	100.2	87.6	81.2	No OMIM phenotype
SYCE1	MENDELIOME	92.9	98.7	93.6	?Premature ovarian failure 12, 616947 ?Spermatogenic failure 15, 616950
SYCP3	MENDELIOME	79	97.5	86.8	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYN1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	64.2	74	63.2	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	INTELLECTUAL DISABILITY	58.8	92.9	85.2	No OMIM phenotype ?Intellectual disability, nonsyndromic (Rauch (2012) Lancet epub)
SYNE1	MOVEMENT DISORDERS HEART PANEL MENDELIOME PRECONCEPTION SCREENING	136.6	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	HEART PANEL MENDELIOME	123.1	98.6	96	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	73.9	98.3	91.6	Deafness, autosomal recessive 76, 615540
SYNGAP1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	141.3	98.4	98	Mental retardation, autosomal dominant 5, 612621
SYNJ1	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	127.2	99.3	96.1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	72	99.8	94	Mental retardation, X-linked 96, 300802
SYT1	INTELLECTUAL DISABILITY MENDELIOME	171.3	99.8	98.6	Baker-Gordon syndrome, 618218
SYT14	MENDELIOME PRECONCEPTION SCREENING	113.5	59.9	53.8	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	NEUROPATHIES MENDELIOME	101.3	100	99	Myasthenic syndrome, congenital, 7, presynaptic, 616040
SZT2	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	149.5	99.5	99.2	Epileptic encephalopathy, early infantile, 18, 615476
T	MENDELIOME	141.2	98.8	93.6	Sacral agenesis with vertebral anomalies, 615709

					{Neural tube defects, susceptibility to}, 182940
TAB2	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	210.5	99.7	97.6	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	HH MENDELIOME PRECONCEPTION SCREENING	80.5	99.1	91.1	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	91.7	97	92.6	Mitochondrial complex IV deficiency, 220110
TACR3	HH MENDELIOME PRECONCEPTION SCREENING	180.3	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	223.6	99.1	96.6	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARK	112.4	99.4	96.6	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TAF13	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	81.8	100	99.9	Mental retardation, autosomal recessive 60, 617432
TAF2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	112.8	98.8	94.7	Mental retardation, autosomal recessive 40, 615599
TAF4B	MENDELIOME	146.1	96.5	90.9	?Spermatogenic failure 13, 615841
TAF6	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130	99.9	98.6	Alazami-Yuan syndrome, 617126
TAL1	MENDELIOME	43.7	73.9	63.3	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	MENDELIOME	112.5	100	100	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	130.5	100	99.9	Transaldolase deficiency, 606003
TANC2	INTELLECTUAL DISABILITY	159.1	99.9	99.3	No OMIM phenotype
TANGO2	MOVEMENT DISORDERS EPILEPSY METABOLIC DISORDERS	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878

	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS				
TAP1	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	103.3	100	99.1	Bare lymphocyte syndrome, type I, 604571
TAP2	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	95.2	99.6	98.6	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	100.7	96.1	94.3	Bare lymphocyte syndrome, type I, 604571
TAPT1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	83.5	88.5	85.9	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	ALS MENDELIOME	175.2	100	100	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TARS2	MENDELIOME MITOCHONDRIAL DISORDERS	98.8	99.9	98.5	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	SKIN DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.1	100	100	Tyrosinemia, type II, 276600
TAZ	CARDIO HEART PANEL PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	94	99.9	98.8	Barth syndrome, 302060
TBC1D20	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	145.7	94.2	94.1	Warburg micro syndrome 4, 615663

	PRECONCEPTION SCREENING				
TBC1D23	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	86	95.7	91.5	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	SKIN DISORDERS HEARING IMPAIRMENT EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	179.2	100	100	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBC1D32	CILIO	81.2	96.6	91.2	OFD type IX
TBC1D7	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	105.5	99.6	96.6	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	152.9	95.5	92.3	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	EPILEPSY SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128	99.9	98.2	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TBCK	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	86.5	95.7	89.3	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	ALS MENDELIOME	102.5	97.8	90.7	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900
TBL1XR1	INTELLECTUAL DISABILITY MENDELIOME	79.4	91.5	73	Mental retardation, autosomal dominant 41, 616944 Pierpont syndrome, 602342
TBP	INTELLECTUAL DISABILITY MENDELIOME	129.5	100	98.1	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600
TBR1	INTELLECTUAL DISABILITY MENDELIOME	120.8	100	99.1	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	CRANIOFACIAL ANOMALIES CONGENITAL HEART DISEASE	75.3	77.1	67.4	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400

	HEART PANEL HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME				Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX15	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	131.7	100	99.4	Cousin syndrome, 260660
TBX18	RENAL DISORDERS MENDELIOME	94.4	98	95.2	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	MENDELIOME PRECONCEPTION SCREENING	174.4	100	100	Adrenocorticotropic hormone deficiency, 201400
TBX2	MENDELIOME	107.6	99.1	92.7	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	142.8	99.9	99.3	Atrial septal defect 4, 611363
TBX21	MENDELIOME	81.7	90	83.1	Asthma and nasal polyps, 208550 {Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	CRANIOFACIAL ANOMALIES MENDELIOME	121.8	99.2	96.3	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TBX3	SKIN DISORDERS MENDELIOME	80.5	99.6	95.3	Ulnar-mammary syndrome, 181450
TBX4	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	170.7	94.9	92.8	Ischiocoxopodopatellar syndrome, 147891
TBX5	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	141.3	100	100	Holt-Oram syndrome, 142900
TBX6	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	122.1	91.5	79.7	Spondylocostal dysostosis 5, 122600
TBXA2R	HEMOSTATIC/THROMBOTIC DISORDERS	83.7	97.6	92.9	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCAP	CARDIO HEART PANEL MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	89	100	99.2	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954

TCF12	CRANIOFACIAL ANOMALIES HH MENDELIOME	150.3	100	99.8	Craniosynostosis 3, 615314
TCF20	INTELLECTUAL DISABILITY	144.3	100	100	No OMIM phenotype Autism spectrum disorder (Babbs (2014) J Med Genet 51,737)
TCF3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	67.8	98.9	92.3	Agammaglobulinemia 8, autosomal dominant, 616941
TCF4	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	128	99.9	99.5	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TCF7L2	INTELLECTUAL DISABILITY	150.4	99.6	96.5	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCHH	SKIN DISORDERS MENDELIOME	148.1	100	100	?Uncombable hair syndrome 3, 617252
TCIRG1	BONE MARROW FAILURE SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TCN2	PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	175.6	100	100	Transcobalamin II deficiency, 275350
TCOF1	CRANIOFACIAL ANOMALIES MENDELIOME	98.6	99.5	97.3	Treacher Collins syndrome 1, 154500
TCTEX1D2	CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	126.1	100	99.3	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	98.8	95.7	92.8	Joubert syndrome 13, 614173
TCTN2	CILIO SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	144.2	99.5	97	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	VISION DISORDERS	127.6	100	99.8	Joubert syndrome 18, 614815

	CILIO DSD SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				Orofaciodigital syndrome IV, 258860
TDGF1	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	151.4	99.8	96.4	Forebrain defects, 0
TDP1	MOVEMENT DISORDERS NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	122.9	98.7	95.3	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	165.1	99.9	98.8	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	168.3	99	97.7	Cataract 36, 613887
TDRD9	MENDELIOME	121.2	98.5	96.5	?Spermatogenic failure 30, 618110
TEAD1	VISION DISORDERS MENDELIOME	158.6	99.8	98.2	Sveinsson chorioretinal atrophy, 108985
TECPR2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	161.1	100	99.9	Spastic paraplegia 49, autosomal recessive, 615031
TECR	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	94.6	99.9	97.9	Mental retardation, autosomal recessive 14, 614020
TECRL	HEART PANEL MENDELIOME	59.3	89.9	77.1	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	208	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	SKIN DISORDERS MENDELIOME	184.1	100	100	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195

TELO2	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	98.1	97.4	93.7	You-Hoover-Fong syndrome, 616954
TENM3	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	185.7	99.5	98.7	Microphthalmia, isolated, with coloboma 9, 615145
TENM4	MOVEMENT DISORDERS MENDELIOME	160.8	99.9	99.2	Essential tremor, hereditary, 5, 616736
TERC	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER	123456	123456	123456	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	SKIN DISORDERS HEREDITARY CANCER	116.7	100	97.6	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107) Chronic lymphocytic leukaemia (Speedy (2016) Blood 128,2319)
TERT	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES HEREDITARY CANCER	138.3	95.3	92	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742
TET2	MENDELIOME	212.4	100	99.9	Myelodysplastic syndrome, somatic, 614286
TEX11	MENDELIOME	77.3	91.6	83.4	Spermatogenic failure, X-linked, 2, 309120
TEX14	MENDELIOME	111.6	99.7	98	?Spermatogenic failure 23, 617707
TEX15	MENDELIOME	114.6	99.8	99	Spermatogenic failure 25, 617960
TF	IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	125.9	100	100	Atransferrinemia, 209300
TFAM	MENDELIOME	65	88.7	66.2	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	CRANIOFACIAL ANOMALIES SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	109.3	100	99.3	Branchiooculofacial syndrome, 113620
TFAP2B	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	153.5	98.8	96.3	Char syndrome, 169100 Patent ductus arteriosus 2, 617035
TFB2M	MITOCHONDRIAL DISORDERS	64.4	99.2	94.1	
TFE3	MENDELIOME	73.5	99.5	93.1	Renal cell carcinoma, papillary, 1, 300854
TFG	NEUROPATHIES MENDELIOME	121.5	93.9	90.7	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484

TFR2	IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	104.4	98.2	93.5	Hemochromatosis, type 3, 604250
TFRC	IRON DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	157.2	99.9	99.1	Immunodeficiency 46, 616740
TG	MENDELIOME PRECONCEPTION SCREENING	133.4	100	99.7	Thyroid dyshormonogenesis 3, 274700 {Autoimmune thyroid disease, susceptibility to, 3}, 608175
TGDS	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	82.4	98.1	88.8	Catel-Manzke syndrome, 616145
TGFB1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	86.9	99.7	95.1	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 {Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	SKIN DISORDERS HEART PANEL MENDELIOME	176.9	100	99.9	Loeys-Dietz syndrome 4, 614816
TGFB3	HEART PANEL MENDELIOME	171.5	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBI	VISION DISORDERS MENDELIOME	130.9	99	94.5	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082
TGFBR1	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEART PANEL INTELLECTUAL DISABILITY MENDELIOME	173.4	93.7	93.6	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEART PANEL MENDELIOME	193.5	100	99.9	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	CRANIOFACIAL ANOMALIES	138.3	100	100	Holoprosencephaly 4, 142946

	INTELLECTUAL DISABILITY MENDELIOME				
TGM1	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	158.8	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	SKIN DISORDERS MENDELIOME	187.1	100	99.8	?Uncombable hair syndrome 2, 617251
TGM5	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	173.9	100	100	Peeling skin syndrome 2, 609796
TGM6	MOVEMENT DISORDERS MENDELIOME	149.7	99.7	98	Spinocerebellar ataxia 35, 613908
TH	MOVEMENT DISORDERS METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PARK PRECONCEPTION SCREENING	68.2	97.6	88.7	Segawa syndrome, recessive, 605407
THAP1	MOVEMENT DISORDERS MENDELIOME	122	100	100	Dystonia 6, torsion, 602629
THBD	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES RENAL DISORDERS MENDELIOME	108.2	99.8	97.8	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THG1L	MITOCHONDRIAL DISORDERS	142.9	100	99.9	
THOC2	INTELLECTUAL DISABILITY MENDELIOME	77.9	96.2	86.6	Mental retardation, X-linked 12/35, 300957
THOC6	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	248.6	100	99.9	Beaulieu-Boycott-Innes syndrome, 613680
THPO	BONE MARROW FAILURE HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	88.2	100	100	Thrombocythemia 1, 187950
THRA	MENDELIOME	172.8	100	99.8	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	167.4	100	99.5	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650
TIA1	MENDELIOME	128.5	98.6	89.6	Welander distal myopathy, 604454
TICAM1	PRIMARY IMMUNODEFICIENCIES	111.1	100	99.1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TIMM22	MITOCHONDRIAL DISORDERS	95.4	100	99.9	

TIMM44	MITOCHONDRIAL DISORDERS	123.3	100	98.5	
TIMM50	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	108.2	98.8	95.3	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	MOVEMENT DISORDERS VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	46	94.5	78.8	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	MENDELIOME MITOCHONDRIAL DISORDERS	152.2	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP1		145	100	100	
TIMP2		127.2	90.7	82.5	
TIMP3	VISION DISORDERS MENDELIOME	147	100	100	Sorsby fundus dystrophy, 136900
TINF2	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME HEREDITARY CANCER	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	PRIMARY IMMUNODEFICIENCIES	136.4	100	100	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Pneumococcal disease, invasive, protection against}, 610799 {Tuberculosis, protection against}, 607948
TJP2	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	111.1	93.8	92.2	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748
TK2	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	105.7	93.4	89.4	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TKT	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	114.1	98.7	97.7	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	MENDELIOME	109.9	99.9	98.9	Preimplantation embryonic lethality, 616814

	PRECONCEPTION SCREENING				
TLK2	INTELLECTUAL DISABILITY MENDELIOME	113	98.8	94.7	Mental retardation, autosomal dominant 57, 618050
TLL1	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	140.1	100	99.9	Atrial septal defect 6, 613087
TLR3	PRIMARY IMMUNODEFICIENCIES	185.4	99.8	98.6	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 {HIV1 infection, resistance to}, 609423
TLR4	PRIMARY IMMUNODEFICIENCIES	132.7	100	99.9	Endotoxin hyporesponsiveness {Colorectal cancer, susceptibility to}, 114500 {Macular degeneration, age-related, 10}, 611488
TMC1	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	122.8	98.2	93.8	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	83.7	99.9	99	Epidermodysplasia verruciformis, 226400
TMC8	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	108.1	97.6	91.6	Epidermodysplasia verruciformis 2, 618231
TMCO1	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	78.7	88	86.5	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMCO3	VISION DISORDERS	131.4	99.9	98.4	No OMIM phenotype Cornea Guttata and Anterior Polar Cataract
TMEM106B	MOVEMENT DISORDERS MENDELIOME	120.2	99.8	96.4	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	CILIO RENAL DISORDERS MENDELIOME	163.8	100	100	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM126A	VISION DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	120.3	98.4	86.2	Optic atrophy 7, 612989
TMEM126B	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	79.2	99.8	97.7	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM127	HEREDITARY CANCER	107.4	98.2	94.7	{Pheochromocytoma, susceptibility to}, 171300

TMEM132E	HEARING IMPAIRMENT	115.9	94.8	91.8	No OMIM phenotype Deafness,autosomal dominant 99 (Li et al. Hum Mutat 2015 36(1) 98-105)
TMEM138	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.2	100	99.5	Joubert syndrome 16, 614465
TMEM14C	IRON DISORDERS	114.2	100	99.7	No OMIM phenotype ?combined porphyria and anemia, severe pathogenic effects are lethal but mild defects might modulate existing anemia and porphyria (Paw et al. (2013), Yien et al. (2014)).
TMEM165	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	113.9	99.8	98.1	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	100.8	98.7	93.4	STING-associated vasculopathy, infantile-onset, 615934
TMEM186	MITOCHONDRIAL DISORDERS	152.4	100	100	
TMEM199	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	105.1	100	99.9	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	111.9	100	98.7	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	VISION DISORDERS CILIO SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	VISION DISORDERS CILIO INTELLECTUAL DISABILITY	100.7	99.8	98.3	Joubert syndrome 14, 614424

	RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
TMEM240	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	112.2	99.8	97.4	Spinocerebellar ataxia 21, 607454
TMEM260	CILIO RENAL DISORDERS MENDELIOME	116.7	96.8	90	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	114.3	100	99.4	Osteogenesis imperfecta, type XIV, 615066
TMEM43	CARDIO HEART PANEL MENDELIOME	124.9	100	99.5	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM5	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	120.5	96.8	92.9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
TMEM65	MITOCHONDRIAL DISORDERS	50.2	79.2	65.9	
TMEM67	MOVEMENT DISORDERS VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	72.9	93.3	83.4	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TMEM70	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	138.7	94.6	90.3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM98	MENDELIOME	136.3	99.4	98.7	Nanophthalmos 4, 615972
TMIE	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	109.6	98.8	92.1	Deafness, autosomal recessive 6, 600971
TMLHE	METABOLIC DISORDERS INTELLECTUAL DISABILITY	100.9	99.9	97.6	{Autism, susceptibility to, X-linked 6}, 300872
TMPO	HEART PANEL	117.8	98.7	94.5	?Cardiomyopathy, dilated, 1T, 613740
TMPRSS15	MENDELIOME	116.9	95.5	89.1	Enterokinase deficiency, 226200

	PRECONCEPTION SCREENING				
TMPRSS3	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	125.5	100	99.9	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	IRON DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.7	100	99.1	Iron-refractory iron deficiency anemia, 206200
TMTC2	HEARING IMPAIRMENT	146.9	97.5	97.4	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/27311106
TMTC3	INTELLECTUAL DISABILITY MENDELIOME	83.2	98.9	93	Lissencephaly 8, 617255
TMX2	MITOCHONDRIAL DISORDERS	141	100	99.5	
TNC	HEARING IMPAIRMENT MENDELIOME	187.5	100	99.7	Deafness, autosomal dominant 56, 615629
TNFAIP3	PRIMARY IMMUNODEFICIENCIES MENDELIOME	135.9	100	99.9	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	MENDELIOME	114.1	100	100	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME HEREDITARY CANCER	146.3	93.3	91.4	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	224.8	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	102.1	100	99.7	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	55.8	76.5	66.8	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	93.2	90.8	87.9	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	PRIMARY IMMUNODEFICIENCIES MENDELIOME	51.6	97.3	85.4	?Immunodeficiency 16, 615593
TNFSF11	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	150.4	99.3	93.2	Osteopetrosis, autosomal recessive 2, 259710

	PRECONCEPTION SCREENING				
TNFSF12	PRIMARY IMMUNODEFICIENCIES	77.5	94.7	90.6	No OMIM phenotype Antibody deficiency (Wang (2013) Proc Natl Acad Sci USA 110, 5127)
TNIK	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	111.1	99.9	99.3	Mental retardation, autosomal recessive 54, 617028
TNNC1	CARDIO HEART PANEL MENDELIOME	174.5	100	100	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	 MENDELIOME MUSCLE DISORDERS	121.2	100	99.6	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNI3	CARDIO HEART PANEL MENDELIOME PRECONCEPTION SCREENING	86.7	98.1	86.5	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, hypertrophic, 7, 613690
TNNI3K	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	118.8	98.8	96	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	86.9	96.3	94	Nemaline myopathy 5, Amish type, 605355
TNNT2	CARDIO HEART PANEL MENDELIOME RITME	106.3	100	99.9	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, familial restrictive, 3, 612422 Cardiomyopathy, hypertrophic, 2, 115195 Left ventricular noncompaction 6, 601494
TNNT3	 MENDELIOME	121	99.9	97.8	Arthrogryposis, distal, type 2B, 601680
TNPO3	MENDELIOME MUSCLE DISORDERS	139.6	100	99.7	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNRC6A	MENDELIOME	162.9	99.9	99.4	?Epilepsy, familial adult myoclonic, 6, 618074
TNXB	 SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	96.4	98.4	91.4	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	MOVEMENT DISORDERS DSD EPILEPSY INTELLECTUAL DISABILITY	165.1	100	100	Pontocerebellar hypoplasia, type 7, 614969

	MENDELIOME PRECONCEPTION SCREENING				
TOP1	MENDELIOME	97.9	99.5	97.3	DNA topoisomerase I, camptothecin-resistant, 0
TOP2A	MENDELIOME	123.8	99.6	97.2	DNA topoisomerase II, resistance to inhibition of, by amsacrine, 0
TOP3A	MENDELIOME MITOCHONDRIAL DISORDERS	129.8	98.9	96.5	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TOPORS	VISION DISORDERS MENDELIOME	210.6	100	100	Retinitis pigmentosa 31, 609923
TOR1A	MOVEMENT DISORDERS MENDELIOME	185	100	99.8	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}, 0
TOR1AIP1	HEART PANEL MENDELIOME	143.8	97.6	95.9	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53	BRSTKNK MENDELIOME HEREDITARY CANCER	92	99.9	98.1	Adrenocortical carcinoma, pediatric, 202300 Bone marrow failure syndrome 5, 618165 Breast cancer, somatic, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, somatic, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, somatic, 607107 Osteosarcoma, 259500 Pancreatic cancer, somatic, 260350 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800
TP53RK	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	37.4	91.3	76.5	Galloway-Mowat syndrome 4, 617730
TP63	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME	206.3	100	100	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPCN2	SKIN DISORDERS	144.2	94.1	89.6	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TPI1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103	99.2	96.7	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512

TPK1	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	112.7	99.8	97.3	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	CARDIO HEART PANEL MENDELIOME	132.9	99.7	97.9	Cardiomyopathy, dilated, 1Y, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Left ventricular noncompaction 9, 611878
TPM2	MENDELIOME MUSCLE DISORDERS	109.1	100	99.6	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	MENDELIOME MUSCLE DISORDERS	98.9	89.4	89.1	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPM4	HEMOSTATIC/THROMBOTIC DISORDERS	51.9	92.1	79.5	No OMIM phenotype Pleines et al.,2017. Mutations in tropomyosin 4 underlie a rare form of human macrothrombocytopenia. J Clin Invest. 2017 127:814-829.
TPMT	METABOLIC DISORDERS	45.5	94.7	78.6	{Thiopurines, poor metabolism of, 1}, 610460
TPO	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	134.8	99.9	98.5	Thyroid dyshormonogenesis 2A, 274500
TPP1	MOVEMENT DISORDERS VISION DISORDERS EPILEPSY METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	146.3	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	PRIMARY IMMUNODEFICIENCIES	119.2	98.9	94.3	No OMIM phenotype Evans syndrome, immunodeficiency and premature immunosenescence (Stepensky (2015) Blood 125, 753)
TPRKB	INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME	56.7	79.3	67.1	Galloway-Mowat syndrome 5, 617731
TPRN	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	62.7	74.7	65.4	Deafness, autosomal recessive 79, 613307
TRAC	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	170.9	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	PRIMARY IMMUNODEFICIENCIES	130.6	99.9	98.4	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5},

					614849
TRAF3IP1	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	90.3	96.3	92.8	Senior-Loken syndrome 9, 616629
TRAF3IP2	PRIMARY IMMUNODEFICIENCIES MENDELIOME	116.6	99.9	97.7	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TRAF6	CRANIOFACIAL ANOMALIES	106.9	96.1	87.1	No OMIM phenotype Ectodermal dysplasia, hypohidrotic (Wisniewski (2012) Br J Dermatol 166,1353)
TRAF7	INTELLECTUAL DISABILITY MENDELIOME	147.2	98.3	95	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	141.6	100	100	Seckel syndrome 9, 616777
TRAK1	METABOLIC DISORDERS MENDELIOME	152.3	99	98.1	Epileptic encephalopathy, early infantile, 68, 618201
TRAP1		134.9	97.6	94.1	
TRAPPC11	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	126.2	99.4	96.4	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	MENDELIOME	141.6	99.9	98.5	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	85.3	87	66.7	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	MITOCHONDRIAL DISORDERS	230.2	100	100	
TRAPPC6B	INTELLECTUAL DISABILITY MENDELIOME	61.8	99.4	94.4	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135	100	99.9	Mental retardation, autosomal recessive 13, 613192
TRDN	HEART PANEL MENDELIOME PRECONCEPTION SCREENING RITME	71.9	83.6	70.8	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREH	METABOLIC DISORDERS MENDELIOME	143.1	98	93.5	Trehalase deficiency, 612119
TREM2	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	149	99.9	99.6	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193

TREX1	MOVEMENT DISORDERS VISION DISORDERS SKIN DISORDERS EPILEPSY HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	242.4	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRH	PRECONCEPTION SCREENING	75.8	95.9	85.1	Thyrotropin-releasing hormone deficiency, 275120
TRHR	MENDELIOME	232	100	99.7	Thyrotropin-releasing hormone resistance, generalized, 0
TRIB1		161.6	92.9	75	
TRIM2	NEUROPATHIES MENDELIOME PRECONCEPTION SCREENING	157.7	93.6	91.4	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	VISION DISORDERS CILIO SKIN DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	MENDELIOME	156.8	99.1	96.8	?Anencephaly, 206500
TRIM37	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.2	98.2	97.2	Mulibrey nanism, 253250
TRIM44	MENDELIOME	141.7	100	100	?Aniridia 3, 617142
TRIM63	HEART PANEL	118.6	100	99.6	No OMIM phenotype Hypertrophic cardiomyopathy (Chen (2012) Circ Res 111,907)
TRIO	INTELLECTUAL DISABILITY MENDELIOME	134.3	97.9	95.4	Mental retardation, autosomal dominant 44, 617061
TRIOBP	HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	135.6	97	94.9	Deafness, autosomal recessive 28, 609823
TRIP11	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	84.3	95.2	87.4	Achondrogenesis, type IA, 200600
TRIP12	INTELLECTUAL DISABILITY MENDELIOME	139.8	99.5	98.8	Mental retardation, autosomal dominant 49, 617752
TRIP13	MENDELIOME	141.2	100	100	Mosaic variegated aneuploidy syndrome 3, 617598

	HEREDITARY CANCER				
TRIP4	MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	113.5	100	98.8	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	119.4	100	99.8	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	INTELLECTUAL DISABILITY	108	99.8	97.4	No OMIM phenotype Intellectual disability (Davarniya (2015) PLoS One 10,e0129631)
TRMT10A	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	135.2	100	99.4	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	131.4	99.8	98.8	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	208.7	99.2	93.9	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	99	100	99.6	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	VISION DISORDERS IRON DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	104.6	97.8	92.3	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TRPA1	MENDELIOME	92.4	91.9	85.3	?Episodic pain syndrome, familial, 1, 615040
TRPC3	MENDELIOME	180.8	98.7	95.6	?Spinocerebellar ataxia 41, 616410
TRPC6	RENAL DISORDERS MENDELIOME	103.8	99	96.1	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	161	100	99.6	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM4	HEART PANEL MENDELIOME RITME	109.2	99.8	98.5	Progressive familial heart block, type IB, 604559
TRPM6	EPILEPSY RENAL DISORDERS	151.1	99.8	98.7	Hypomagnesemia 1, intestinal, 602014

	MENDELIOME PRECONCEPTION SCREENING				
TRPS1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	175	100	99.8	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	SKIN DISORDERS MENDELIOME	144.9	100	99.4	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TRPV4	NEUROPATHIES SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME MUSCLE DISORDERS	172.4	99.5	98.7	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TRPV6	MENDELIOME	167.4	99.5	98.5	Hyperparathyroidism, transient neonatal, 618188
TSC1	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME HEREDITARY CANCER	128.8	99.8	98.8	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	SKIN DISORDERS EPILEPSY INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME HEREDITARY CANCER	131.2	100	99	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	74.2	99	93.6	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	MOVEMENT DISORDERS EPILEPSY MENDELIOME PRECONCEPTION SCREENING	123.8	100	99.8	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	MENDELIOME	53.5	90.5	85.7	?Pontocerebellar hypoplasia type 2C, 612390

	PRECONCEPTION SCREENING				
TSEN54	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	82.9	95.9	92.9	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSFM	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	127.2	100	100	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	MENDELIOME	102.5	98.5	93.1	?Spermatogenic failure 26, 617961
TSHB	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	271.7	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	MENDELIOME PRECONCEPTION SCREENING	216.5	99.2	96.8	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSHZ1	CRANIOFACIAL ANOMALIES MENDELIOME	166.6	98.8	98.5	Aural atresia, congenital, 607842
TSPAN12	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	129.4	100	99.5	Exudative vitreoretinopathy 5, 613310
TSPAN7	INTELLECTUAL DISABILITY MENDELIOME	120.7	99.9	98.6	Mental retardation, X-linked 58, 300210
TSPEAR	CRANIOFACIAL ANOMALIES SKIN DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	141.5	100	99	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	DSD MENDELIOME PRECONCEPTION SCREENING	141.5	100	99.4	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	BONE MARROW FAILURE MENDELIOME	81.6	100	99.1	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	MOVEMENT DISORDERS CILIO MENDELIOME	123.2	100	98.9	Spinocerebellar ataxia 11, 604432
TTC19	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME	92.1	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157

	MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS				
TTC21B	CILIO SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	100.7	99.7	97.6	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	CILIO MENDELIOME PRECONCEPTION SCREENING	103.4	100	99.5	Ciliary dyskinesia, primary, 35, 617092
TTC26	CILIO	134.3	99.8	98.3	Joubert BBS
TTC37	SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	124	99.6	98.1	Trichohepatoenteric syndrome 1, 222470
TTC7A	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	123	99.9	98.3	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	VISION DISORDERS CILIO INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	106.9	97.9	92	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TTI2	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	104.5	100	99.7	Mental retardation, autosomal recessive 39, 615541
TTLL5	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	152.7	99.9	98.7	Cone-rod dystrophy 19, 615860
TTN	 HEART PANEL MENDELIOME MUSCLE DISORDERS TTN	187.8	98.2	97.2	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, proximal, with early respiratory muscle involvement, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334

TTPA	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	101.5	83.6	76.6	Ataxia with isolated vitamin E deficiency, 277460
TTR	CARDIO HEART PANEL NEUROPATHIES MENDELIOME	152.3	94.6	94.6	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUB	VISION DISORDERS MENDELIOME	103	97.3	95.2	?Retinal dystrophy and obesity, 616188
TUBA1A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	113.2	99.9	97.8	Lissencephaly 3, 611603
TUBA3D	VISION DISORDERS MENDELIOME	144.5	100	99.6	Keratoconus 9, 617928
TUBA4A	ALS MENDELIOME	220.6	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	177.1	99.9	99.7	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBB	INTELLECTUAL DISABILITY MENDELIOME	158.6	99.3	97.4	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610
TUBB1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	186.5	100	100	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	109.9	96.7	95.6	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	100	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	VISION DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME	136.1	98.1	96.9	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TUBB4A	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	121.2	96	95.3	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	VISION DISORDERS MENDELIOME	109.8	98.4	96	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	MENDELIOME	111.2	90.9	90.5	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732

TUBB8	MENDELIOME	31.3	95.5	71	Oocyte maturation defect 2, 616780
TUBG1	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	164.2	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP4	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.8	99.1	96.2	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	152.2	99.9	98.9	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	135.4	100	99.7	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	VISION DISORDERS CILIO MENDELIOME PRECONCEPTION SCREENING	97.8	96.8	91.7	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	136.4	100	98.3	Mental retardation, autosomal recessive 7, 611093
TWIST1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	134.4	96.6	87.2	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
TWIST2	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	131.3	100	99.3	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	MOVEMENT DISORDERS NEUROPATHIES INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	178.8	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
TXN2	MENDELIOME MITOCHONDRIAL DISORDERS	81.2	100	100	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	MENDELIOME PRECONCEPTION SCREENING	109	100	99.5	Burn-McKeown syndrome, 608572

TXNRD2	HEART PANEL MENDELIOME	119.3	93.3	91.2	?Glucocorticoid deficiency 5, 617825
TYK2	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	119.2	99.9	98.8	Immunodeficiency 35, 611521
TYMP	METABOLIC DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	95.2	98.3	85	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	VISION DISORDERS SKIN DISORDERS HEARING IMPAIRMENT METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	185.3	100	100	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYROBP	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	95.2	100	99.9	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	VISION DISORDERS SKIN DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	176.9	100	99.9	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBA1	 MENDELIOME MUSCLE DISORDERS	162	99.8	98.9	Spinal muscular atrophy, X-linked 2, infantile, 301830
UBA5	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	75.4	94.1	77.1	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBB	CRANIOFACIAL ANOMALIES	61.9	100	99.7	Cleft palate, isolated, 119540
UBE2A	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	100.5	99.9	96.9	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE2T	BONE MARROW FAILURE MENDELIOME PRECONCEPTION SCREENING	107	100	99.3	Fanconi anemia, complementation group T, 616435
UBE3A	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	89.8	97.8	91.4	Angelman syndrome, 105830
UBE3B	INTELLECTUAL DISABILITY	127.8	100	99.9	Kaufman oculocerebrofacial syndrome, 244450

	MENDELIOME PRECONCEPTION SCREENING				
UBIAD1	VISION DISORDERS MENDELIOME	248.8	98.9	95.2	Corneal dystrophy, Schnyder type, 121800
UBQLN2	ALS MENDELIOME	136.3	99.7	98	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	128.2	99.2	96	Johanson-Blizzard syndrome, 243800
UBTF	INTELLECTUAL DISABILITY MENDELIOME	123.6	99.9	99	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	MENDELIOME	109.1	97.9	89.1	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UFC1	INTELLECTUAL DISABILITY MENDELIOME	150.3	100	100	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	INTELLECTUAL DISABILITY MENDELIOME	105.8	71	70.1	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	128.4	99.7	96.5	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
UGT1A1	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	240.6	100	100	Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 Hyperbilirubinemia, familial transient neonatal, 237900 [Bilirubin, serum level of, QTL1], 601816 [Gilbert syndrome], 143500
UMOD	RENAL DISORDERS MENDELIOME	127	97.8	97.2	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UMPS	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	173.6	99.3	97.2	Orotic aciduria, 258900
UNC119	VISION DISORDERS MENDELIOME	92.9	97.8	90.4	?Cone-rod dystrophy, 0 ?Immunodeficiency 13, 615518
UNC13A	INTELLECTUAL DISABILITY	140.8	99.1	97.8	No OMIM phenotype PMID 28192369, in house recurrency
UNC13D	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	97	99.6	97.7	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45B	VISION DISORDERS MENDELIOME	143	100	99.5	?Cataract 43, 616279
UNC80	INTELLECTUAL DISABILITY	133.4	99.9	99.1	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801

	MENDELIOME PRECONCEPTION SCREENING				
UNC93B1	PRIMARY IMMUNODEFICIENCIES	60	56.8	54.5	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	78.4	99.5	94.2	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	157.4	100	100	Beta-ureidopropionase deficiency, 613161
UPF3B	INTELLECTUAL DISABILITY MENDELIOME	47.4	91.2	76.6	Mental retardation, X-linked, syndromic 14, 300676
UPK3A	RENAL DISORDERS	110.5	99.2	96.7	No OMIM phenotype Renal hypodysplasia (Schonfelder (2006) Am J Kidney Dis 47, 1004) Renal aysplasia (Jenkins (2005) J Am Soc Nephrol 16, 2141)
UQCC1	MITOCHONDRIAL DISORDERS	96.7	100	100	
UQCC2	RENAL DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	96.6	100	99.1	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	MENDELIOME MITOCHONDRIAL DISORDERS	95	100	99.2	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	MITOCHONDRIAL DISORDERS	189.2	100	100	
UQCR11	MITOCHONDRIAL DISORDERS	158.7	100	100	
UQCRB	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	107.6	99.6	96.8	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	MITOCHONDRIAL DISORDERS	151.4	100	99.8	
UQCRC2	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	122.6	99.9	99.1	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	MITOCHONDRIAL DISORDERS	151.9	87	82.6	
UQCRH	MITOCHONDRIAL DISORDERS	131.1	99.9	98.3	
UQCRQ	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	131.3	100	99.9	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	132	99.9	99	?Urocanase deficiency, 276880
UROD	SKIN DISORDERS	163.1	99.8	97.9	Porphyria cutanea tarda, 176100

	METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING				Porphyria, hepatoerythropoietic, 176100
UROS	SKIN DISORDERS IRON DISORDERS METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	108.3	100	99.9	Porphyria, congenital erythropoietic, 263700
USB1	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	125	99.9	98.2	Poikiloderma with neutropenia, 604173
USH1C	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	97.5	100	99.4	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904
USH1G	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	195.3	98.4	96.3	Usher syndrome, type 1G, 606943
USH2A	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	148.5	100	99.7	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USMG5	MITOCHONDRIAL DISORDERS	16.2	76.5	27.7	
USP18	PRIMARY IMMUNODEFICIENCIES MENDELIOME	201.4	95.9	95.9	Pseudo-TORCH syndrome 2, 617397
USP27X	INTELLECTUAL DISABILITY MENDELIOME	248.7	100	100	Mental retardation, X-linked 105, 300984
USP7	INTELLECTUAL DISABILITY	99.4	93.2	88	No OMIM phenotype ?Autism spectrum disorder (Levy (2011) Neuron 70,886)
USP8	MENDELIOME	71.5	97.3	87.8	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	INTELLECTUAL DISABILITY MENDELIOME	108.2	97.2	91.1	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
USP9Y	MENDELIOME	30.8	47.2	39.9	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	149.4	99.1	98.4	UV-sensitive syndrome 3, 614640

VAC14	MENDELIOME PRECONCEPTION SCREENING	108.4	99.8	98.5	Striatonigral degeneration, childhood-onset, 617054
VAMP1	MOVEMENT DISORDERS MENDELIOME	131.5	100	100	Spastic ataxia 1, autosomal dominant, 108600
VANGL1	MENDELIOME	165.3	100	100	Caudal regression syndrome, 600145 {Neural tube defects, susceptibility to}, 182940
VANGL2	MENDELIOME	184.6	100	99.6	Neural tube defects, 182940
VAPB	ALS MENDELIOME	107.8	99	95.9	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VARS	MENDELIOME	126.1	99.9	98	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	110.9	99.9	98.9	Combined oxidative phosphorylation deficiency 20, 615917
VAV1	PRIMARY IMMUNODEFICIENCIES	105.7	98.3	94.7	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
VAX1	VISION DISORDERS CRANIOFACIAL ANOMALIES MENDELIOME	52.2	88.4	78	?Microphthalmia, syndromic 11, 614402
VCAN	VISION DISORDERS MENDELIOME	186.5	100	100	Wagner syndrome 1, 143200
VCL	CARDIO HEART PANEL MENDELIOME	115.8	100	99.8	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255
VCP	ALS MOVEMENT DISORDERS NEUROPATHIES MENDELIOME MUSCLE DISORDERS	144.8	99.9	99.5	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VDR	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	123.3	98	95.2	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	SKIN DISORDERS MENDELIOME	164.5	100	99.5	Lymphatic malformation 4, 615907
VHL	CILIO SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	119.7	92.6	85.3	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300

VIM	VISION DISORDERS MENDELIOME	126.5	99.1	97.2	Cataract 30, pulverulent, 116300
VIPAS39	HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	144.6	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	162.1	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	200.9	99.9	99.4	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	MENDELIOME MUSCLE DISORDERS	42.3	95.2	81.3	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	144.9	95.3	93.2	Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	MOVEMENT DISORDERS MENDELIOME PRECONCEPTION SCREENING	69.5	95.3	85.3	Choreoacanthocytosis, 200150
VPS13B	VISION DISORDERS SKIN DISORDERS PRIMARY IMMUNODEFICIENCIES METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	143.8	98.6	96.8	Cohen syndrome, 216550
VPS13C	MENDELIOME PARK PRECONCEPTION SCREENING	106.6	96.6	90	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	MOVEMENT DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS	158.6	99.9	99.4	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	MENDELIOME	106.3	96.2	95.1	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS RENAL DISORDERS	138.3	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085

	MENDELIOME PRECONCEPTION SCREENING				
VPS35	PARK	92.5	95.1	88.4	{Parkinson disease 17}, 614203
VPS37A	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	73.6	86.6	66.4	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	BONE MARROW FAILURE PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING	131.5	96.2	94.9	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS53	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	129.2	91.4	90.4	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING MUSCLE DISORDERS	124.8	97.5	94.2	Pontocerebellar hypoplasia type 1A, 607596
VSX1	VISION DISORDERS MENDELIOME	52.2	85.5	76.1	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	77.7	99.8	97.3	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	141.5	99.8	98.4	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	HEMOSTATIC/THROMBOTIC DISORDERS MENDELIOME	120.9	100	99.6	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAC	INTELLECTUAL DISABILITY MENDELIOME	165.5	99.7	96.7	Desanto-Shinawi syndrome, 616708
WARS	NEUROPATHIES MENDELIOME	123.8	99.7	98	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS	140.7	100	99.5	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	BONE MARROW FAILURE SKIN DISORDERS	66.1	88.2	78.7	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900

	HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME HEREDITARY CANCER				Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WASF1	INTELLECTUAL DISABILITY	85.6	100	99.2	No OMIM phenotype
WASHC4	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	91.8	95.3	89.6	?Mental retardation, autosomal recessive 43, 615817
WASHC5	MOVEMENT DISORDERS MENDELIOME	146.6	99.6	98.1	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	HEARING IMPAIRMENT MENDELIOME	93.7	100	100	Deafness, autosomal recessive 107, 617639
WDFY3	MENDELIOME	133.6	99.7	98.1	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	VISION DISORDERS CILIO INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	107.3	93.9	88.9	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	PRIMARY IMMUNODEFICIENCIES	114.1	100	99.2	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
WDR11	HH MENDELIOME	130.6	96.9	96.4	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	INTELLECTUAL DISABILITY	122.6	99.9	98.9	No OMIM phenotype Intellectual disability,X-linked (Whibley (2010) Am J Hum Genet 87,173)
WDR19	VISION DISORDERS CRANIOFACIAL ANOMALIES CILIO SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR26	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	97.5	98.3	94.6	Skraban-Deardorff syndrome, 617616
WDR34	CILIO SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	106.6	99.5	96.2	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	CRANIOFACIAL ANOMALIES CILIO	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091

	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING				
WDR36	MENDELIOME	112.3	97.7	90.1	Glaucoma 1, open angle, G, 609887
WDR4	INTELLECTUAL DISABILITY	133.4	100	99.8	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/30079490
WDR45	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PARK	75	97.4	90.1	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	INTELLECTUAL DISABILITY MENDELIOME	85.5	95.8	85.6	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	CILIO DSD SHORT STATURE/SKELETAL DYSPLASIA RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR62	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	161.5	100	99.7	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR66	MENDELIOME	141	100	99.9	Spermatogenic failure 33, 618152
WDR72	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	132.2	96.5	95.4	Amelogenesis imperfecta, type IIA3, 613211
WDR73	MOVEMENT DISORDERS INTELLECTUAL DISABILITY RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	138.9	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	163.3	99.9	99.4	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	MENDELIOME	102.2	99.6	95.7	Oocyte maturation defect 5, 617996

	PRECONCEPTION SCREENING				
WFS1	VISION DISORDERS HEARING IMPAIRMENT INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	251.4	100	99.7	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296 {Diabetes mellitus, noninsulin-dependent, association with}, 125853
WHRN	VISION DISORDERS HEARING IMPAIRMENT MENDELIOME PRECONCEPTION SCREENING	114	99.8	98.8	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	SKIN DISORDERS HEMOSTATIC/THROMBOTIC DISORDERS PRIMARY IMMUNODEFICIENCIES MENDELIOME	77.5	100	99.2	?Wiskott-Aldrich syndrome 2, 614493
WISP3	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	118.4	100	100	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230
WNK1	NEUROPATHIES RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	167.7	99.9	99.5	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	RENAL DISORDERS MENDELIOME	123.1	99.7	98.7	Pseudohypoaldosteronism, type IIB, 614491
WNT1	SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	188.8	100	99.9	Osteogenesis imperfecta, type XV, 615220 {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221
WNT10A	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	114	100	99.1	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	CRANIOFACIAL ANOMALIES SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	144.7	100	99.9	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT2B	MENDELIOME	170.5	95.9	90	Diarrhea 9, 618168
WNT3	MENDELIOME PRECONCEPTION SCREENING	166.8	100	99.6	?Tetra-amelia syndrome 1, 273395
WNT4	DSD RENAL DISORDERS MENDELIOME	263.1	93.4	92.7	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330

	PRECONCEPTION SCREENING				
WNT5A	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME	155.7	100	100	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	216.8	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	BONE MARROW FAILURE SKIN DISORDERS DYSKERATOSIS CONGENITA PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	123.6	98.3	94.6	Werner syndrome, 277700
WT1	DSD RENAL DISORDERS MENDELIOME HEREDITARY CANCER	76.5	91.8	81.4	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
WWOX	MOVEMENT DISORDERS EPILEPSY INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	130.9	100	99.7	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	METABOLIC DISORDERS RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	109.1	100	99.9	Xanthinuria, type I, 278300
XIAP	PRIMARY IMMUNODEFICIENCIES MENDELIOME	107.1	91.9	86.6	Lymphoproliferative syndrome, X-linked, 2, 300635
XIRP2	HEART PANEL	138.5	100	99.7	No OMIM phenotype ?Schizphrenia (Fromer (2014) Nature 506,179)
XIST	MENDELIOME	123456	123456	123456	X-inactivation, familial skewed, 300087
XK	MOVEMENT DISORDERS EPILEPSY	96.8	99.9	99.1	McLeod syndrome with or without chronic granulomatous disease, 300842

	HEART PANEL MENDELIOME MUSCLE DISORDERS				
XPA	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	52.9	98.5	88.9	Xeroderma pigmentosum, group A, 278700
XPC	SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING HEREDITARY CANCER	140.7	100	99.7	Xeroderma pigmentosum, group C, 278720
XPNPEP3	CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	134	100	99.2	Nephronophthisis-like nephropathy 1, 613159
XPR1	MOVEMENT DISORDERS MENDELIOME PARK	131.7	100	99.8	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	MOVEMENT DISORDERS MENDELIOME	106.6	99.7	97.5	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	BONE MARROW FAILURE BRSTKNK MENDELIOME	165.6	93	89.4	?Fanconi anemia, complementation group U, 617247
XRCC4	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	103.2	99.7	97.3	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	132.5	90.4	87.1	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA METABOLIC DISORDERS MENDELIOME PRECONCEPTION SCREENING	136.3	98.9	94.9	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YAP1	VISION DISORDERS HEARING IMPAIRMENT	95.6	87.8	81.6	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433

	MENDELIOME				
YARS	NEUROPATHIES MENDELIOME	122.4	100	100	Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	IRON DISORDERS MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING MUSCLE DISORDERS	173.2	99.8	98.9	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	VISION DISORDERS INTELLECTUAL DISABILITY MENDELIOME MITOCHONDRIAL DISORDERS PRECONCEPTION SCREENING	105.3	97.7	91.9	?Optic atrophy 11, 617302
YWHAE	INTELLECTUAL DISABILITY	114.6	99.7	96.1	No OMIM phenotype Develomental delay,facial dysmorphology and growth retardation (Enomoto (2012) Am J Med Genet A 158A) Developmental delay and mild brain structural abnormalities (Bi (2009) Nat Genet 41,168)
YWHAG	EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	226.6	100	100	Epileptic encephalopathy, early infantile, 56, 617665
YY1	INTELLECTUAL DISABILITY MENDELIOME	134.8	100	98.6	Gabriele-de Vries syndrome, 617557
YY1AP1	MENDELIOME PRECONCEPTION SCREENING	159.2	98.3	97	Grange syndrome, 602531
ZAP70	PRIMARY IMMUNODEFICIENCIES MENDELIOME PRECONCEPTION SCREENING SCID	185.6	99.9	99.5	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB16	SHORT STATURE/SKELETAL DYSPLASIA INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	151.4	100	100	Leukemia, acute promyelocytic, PL2F/RARA type, 0 Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB17	HEART PANEL	142	100	100	No OMIM phenotype
ZBTB18	INTELLECTUAL DISABILITY MENDELIOME	222.7	99.7	99	Mental retardation, autosomal dominant 22, 612337
ZBTB20	SKIN DISORDERS INTELLECTUAL DISABILITY MENDELIOME	216.9	100	100	Primrose syndrome, 259050
ZBTB24	PRIMARY IMMUNODEFICIENCIES INTELLECTUAL DISABILITY	178.1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069

	MENDELIOME PRECONCEPTION SCREENING				
ZBTB42	MENDELIOME	125.6	100	100	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	184.1	99.6	97.2	Mental retardation, autosomal recessive 56, 617125
ZC4H2	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME MUSCLE DISORDERS	78.6	99.8	98.1	Wieacker-Wolff syndrome, 314580
ZDHHC15	MENDELIOME	89.8	97.9	92.6	?Mental retardation, X-linked 91, 300577
ZDHHC9	INTELLECTUAL DISABILITY MENDELIOME	55.5	98.4	89.4	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	VISION DISORDERS MENDELIOME	192.7	100	99.3	Corneal dystrophy, Fuchs endothelial, 6, 613270 Corneal dystrophy, posterior polymorphous, 3, 609141
ZEB2	CRANIOFACIAL ANOMALIES EPILEPSY INTELLECTUAL DISABILITY MENDELIOME	157	99.8	98.8	Mowat-Wilson syndrome, 235730
ZFHX2	MENDELIOME	119	99.9	99.2	?Marsili syndrome, 147430
ZFP57	MENDELIOME	104.4	99.8	98.7	Diabetes mellitus, transient neonatal, 1, 601410
ZFPM2	CONGENITAL HEART DISEASE DSD HEART PANEL MENDELIOME	196.3	100	99.6	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500
ZFYVE26	MOVEMENT DISORDERS INTELLECTUAL DISABILITY MENDELIOME PRECONCEPTION SCREENING	120.3	99.9	99.4	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	MOVEMENT DISORDERS MENDELIOME	118.3	100	100	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	231.1	100	100	Craniosynostosis 6, 616602
ZIC2	CRANIOFACIAL ANOMALIES INTELLECTUAL DISABILITY MENDELIOME	122.5	90.5	78.9	Holoprosencephaly 5, 609637
ZIC3	CONGENITAL HEART DISEASE HEART PANEL MENDELIOME	113.7	100	99.8	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390

ZMPSTE24	SKIN DISORDERS SHORT STATURE/SKELETAL DYSPLASIA MENDELIOME PRECONCEPTION SCREENING	113.3	100	99.1	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYND10	CILIO MENDELIOME PRECONCEPTION SCREENING	136.5	100	100	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	INTELLECTUAL DISABILITY MENDELIOME	137	100	99.8	Mental retardation, autosomal dominant 30, 616083
ZMYND15	MENDELIOME	129.6	99.5	96	?Spermatogenic failure 14, 615842
ZNF141	MENDELIOME	160.6	100	99.9	?Polydactyly, postaxial, type A6, 615226
ZNF148	INTELLECTUAL DISABILITY MENDELIOME	190.8	99.8	98.8	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	INTELLECTUAL DISABILITY	134.7	98.8	97.2	No OMIM phenotype ?Autism (Neale (2012) Nature 485,242)
ZNF335	MENDELIOME	139.9	99.6	98.9	?Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	INTELLECTUAL DISABILITY	176.8	99.2	98.4	No OMIM phenotype Intellectual disability and autism (Ren (2013) Biochim Biophys Acta 1832,431) Cognitive impairment, failure to thrive, hypotonia and dysmorfic features (Kambouris (2014) Orphanet J Rare Dis 9)
ZNF408	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	135.7	100	100	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF41	INTELLECTUAL DISABILITY	103.3	100	99.7	Mental retardation, X-linked 89, 300848
ZNF423	VISION DISORDERS CILIO RENAL DISORDERS MENDELIOME PRECONCEPTION SCREENING	250.8	100	100	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844
ZNF462	INTELLECTUAL DISABILITY	205.2	100	99.9	No OMIM phenotype https://www.ncbi.nlm.nih.gov/pubmed/28513610
ZNF469	VISION DISORDERS SKIN DISORDERS MENDELIOME PRECONCEPTION SCREENING	93.1	98.7	96.3	Brittle cornea syndrome 1, 229200
ZNF513	VISION DISORDERS MENDELIOME PRECONCEPTION SCREENING	110.7	100	99.7	?Retinitis pigmentosa 58, 613617
ZNF592	MOVEMENT DISORDERS	150.1	100	99.9	No OMIM phenotype

	SKIN DISORDERS				
ZNF644	VISION DISORDERS MENDELIOME	156.3	100	99.8	Myopia 21, autosomal dominant, 614167
ZNF687	MENDELIOME	164.7	100	100	Paget disease of bone 6, 616833
ZNF711	INTELLECTUAL DISABILITY MENDELIOME	137.7	98.7	95.5	Mental retardation, X-linked 97, 300803
ZNF750	SKIN DISORDERS MENDELIOME	150.3	100	99.9	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNHIT3	MENDELIOME	121	74.4	74.4	PEHO syndrome, 260565
ZP1	MENDELIOME PRECONCEPTION SCREENING	196	100	100	Oocyte maturation defect 1, 615774
ZP3	MENDELIOME	181.7	100	100	Oocyte maturation defect 3, 617712
ZSWIM6	INTELLECTUAL DISABILITY MENDELIOME	150.9	93.1	89.1	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 31st, 2018.
This list is accurate for panel version DG 2.15

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors