רשימת מחלות ומוטציות בפאנל (2021-02-15) Extended
Hybrid Capture-Based Next Generation Sequencing

Mutation #	Gene: Disease Name {(Transcsript) "Mutation name"}
1	2p21:Hypotonia-cystinuria syndrome {"2p21"}
2 - 6	ABCA12:Ichthyosis, congenital, Autosomal Recessive 4A {(NM_173076.3) "c.1060C>T", "c.179G>C", "c.3456G>A", "c.4544G>A", "c.4553G>A"}
7 - 7	ABCA3:Congenital surfactant deficiency (Surfactant metabolism dysfunction, pulmonary, 3) {(NM_001089) "c.1474dupT"}
8 - 13	ABCA4:Cone-rod dystrophy 3 {(NM_000350.3) "c.1648G>A", "c.2791G>T", "c.3607G>A", "c.3608G>A", "c.5460+1G>A", "c.834delT"}
14 - 16	ABCB11:Cholestasis, progressive familial intrahepatic type 2 {(NM_003742) "c.1100_1101insTA" | (NM_003742.4) "c.1409G>A", "c.3268C>T"}
17 - 22	ABCC8:Hyperinsulinemic hypoglycemia, familial, 1 (PHHI/HHF1) {(NM_000352) "c.3339dupG" | (NM_000352.4) "c.1116dupT", "c.2506C>T", "c.3989-9G>A", "c.4160_4162delTCT", "c.560T>A"}
23 - 23	ABCD1:X-linked adrenoleukodystrophy {(NM_000033.4) "c.686T>C"}
24 - 25	ABHD5:Chanarin-Dorfman syndrome {(NM_016006.6) "c.412T>C", "c.934C>T"}
26 - 36	ACADM:Medium-chain Acyl-CoA dehydrogenase deficiency {(NM_000016.5) "c.1010A>C", "c.1045C>T", "c.1257C>A", "c.362C>T", "c.415_419delGATCA", "c.431_434delAGTA", "c.454G>T", "c.616C>T", "c.621_624delTGAT", "c.799G>A", "c.985A>G"}
37 - 45	ACADVL:Acyl-CoA dehydrogenase, very long-chain, VLCAD deficiency {(NM_000018.4) "c.1096C>T", "c.1748C>T", "c.260T>C", "c.367G>A", "c.637G>A", "c.65C>A", "c.779C>T", "c.799_802delGTTA", "c.894G>A"}
46 - 46	ACO2:Infantile cerebellar-retinal degeneration {(NM_001098.3) "c.336C>G"}
47 - 48	ACP5:Spondyloenchondrodysplasia with immune dysregulation {(NM_001111035) "c.772_790del" | (NM_001111035.2) "c.325G>A"}
49 - 49	ACSF3:Combined malonic and methylmalonic aciduria {(NM_174917) "c.1411C>T"}
50 - 51	ADA:Severe combined immunodeficiency due to ADA deficiency {(NM_000022.4) "c.703C>T", "c.792G>A"}
52 - 52	ADAM9:cone-rod dystrophy 9 {(NM_003816) "c.1087T>A"}
53 - 54	ADAMTS2:Ehlers Danlos syndrome, type VIIC {(NM_014244.5) "c.2384G>A", "c.673C>T"}
55 - 59	ADGRG1:Bilateral Frontoparietal Polymicrogyria (BFPP) {(NM_005682.7) "c.1036T>A", "c.1046G>C", "c.1167+3G>C", "c.1693C>T", "c.739_745delCAGGACC"}
60 - 61	ADGRV1:Usher syndrome, type 2C {(NM_032119.4) "c.14973-2A>G", "c.15494delA"}
62 - 62	AGA:Aspartylglucosaminuria {(NM_000027.4) "c.214T>C"}
63 - 67	AGL:Glycogen storage disease III {(NM_000642.3) "c.1078C>T", "c.1222C>T", "c.2812+2dupT", "c.3652C>T", "c.4456delT"}

68 - 81	AGXT:Hyperoxaluria, primary, type I {(NM_000030) "c.26C>A" | (NM_000030.3) "c.121G>A", "c.33dupC", "c.466G>A", "c.584T>G", "c.586G>A", "c.680+1G>A", "c.697C>T", "c.727G>C", "c.731T>C", "c.860_861delGCinsCG", "c.865C>T", "c.893T>C", "c.997A>T"}
82 - 84	AHI1:Joubert syndrome-3 {(NM_017651.4) "c.2212C>T", "c.3032C>G", "c.787dupC"}
85 - 85	AIMP1:Leukodystrophy, hypomyelinating, 3 {(NM_004757.3) "c.292_293delCA"}
86 - 88	AIPL1:Leber congenital amaurosis 4 {(NM_014336.5) "c.211G>T", "c.215G>A", "c.834G>A"}
89 - 94	AIRE:Autoimmune polyendocrinopathy syndrome , type I (APS-1) with or without reversible metaphyseal dysplasia {(NM_000383.4) "c.1163_1164insA", "c.247A>G", "c.254A>G", "c.44G>A", "c.47C>T", "c.769C>T"}
95 - 95	ALDH1A3:Microphthalmia, isolated 8 {(NM_000693.4) "c.211G>A"}
96 - 96	ALDH7A1:Epilepsy, pyridoxine-dependent {(NM_001182.5) "c.1489+5G>A"}
97 - 104	ALDOB:Fructose intolerance {(NM_000035) "c.178C>T", "c.360_363delCAAA", "c.612T>A", "c.612T>G", "c.865delC" | (NM_000035.4) "c.1005C>G", "c.448G>C", "c.524C>A"}
105 - 107	ALMS1:Alstrom syndrome {(NM_015120.4) "c.8008C>T", "c.808C>T", "c.8171_8181del"}
108 - 110	ALPL:Hypophosphatasia, infantile {(NM_000478) "c.1337delC" | (NM_000478.6) "c.1348C>T", "c.141C>A"}
111 - 111	AMT:Glycine encephalopathy, AMT-related {(NM_000481.3) "c.125A>G"}
112 - 112	ANO5:Limb-girdle muscular dystrophy {(NM_213599.2) "c.191dupA"}
113 - 113	AP4B1:Spastic paraplegia 47, Autosomal Recessive {(NM_006594.4) "c.664delC"}
114 - 114	APTX:Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia {(NM_175073.2) "c.837G>A"}
115 - 116	AQP2:Diabetes insipidus, nephrogenic {(NM_000486.5) "c.298G>T", "c.83T>C"}
117 - 117	ARFGEF2:Periventricular heterotopia with microcephaly {(NM_006420.3) "c.1958+1G>A"}
118 - 118	ARHGDIA:Nephrotic syndrome, type 8 {(NM_004309.6) "c.518G>T"}
119 - 119	ARL6:Bardet-Biedl syndrome 3 {(NM_032146.5) "c.364C>T"}
120 - 133	ARSA:Metachromatic leukodystrophy - MLD {(NM_000487) "c.1114C>T" | (NM_000487.6) "c.1136C>T", "c.1174C>T", "c.1283C>T", "c.211T>G", "c.263G>A", "c.292_293delTCinsCT", "c.465+1G>A", "c.47G>A", "c.542T>G", "c.576G>C", "c.827C>T", "c.937C>T" | (NM_001085425.3) "c.449C>T"}
134 - 134	ARSG:Usher syndrome, type IV {(NM_014960) "c.133G>T"}
135 - 135	ASL:Argininosuccinic aciduria {(NM_000048.4) "c.346C>T"}
136 - 136	ASNS:Asparagine synthetase deficiency {(NM_183356.3) "c.1084T>G"}
137 - 140	ASPA:Canavan Disease {(NM_001128085.1) "c.433-2A>G", "c.693C>A", "c.854A>C", "c.914C>A"}

141 - 141	ASS1:Citrullinemia,classic {(NM_000050) "c.1168G>A"}
142 - 155	ATM:Ataxia-tyelangiectasia {(NM_000051) "c.1514T>C" | (NM_000051.3) "c.103C>T", "c.1339C>T", "c.1547T>C", "c.2284_2285delCT", "c.2839-579_2839-576del4", "c.3245_3247delATCinsTGAT", "c.3576G>A", "c.368delA", "c.497del7514", "c.5763-1050A>G", "c.6672_6680delGGCTCTACGinsCTC", "c.7241_7244delAAGC", "deletion_exon_3-4"}
156 - 156	ATP6V0A2:Cutis laxa, Autosomal Recessive, type IIA {(NM_012463.4) "c.2375C>G"}
157 - 179	ATP7B:Wilson disease {(NM_000053) "c.3007G>A", "c.3784G>T" | (NM_000053.3) "c.122A>G", "c.1340_1343delAAAC", "c.1544G>A", "c.1639delC", "c.1703T>G", "c.1934T>G", "c.2293G>A", "c.2333G>T", "c.2337G>A", "c.2817G>T", "c.2906G>A", "c.3191A>C", "c.3207C>A", "c.3451C>T", "c.3551T>C", "c.3638G>T", "c.3649_3654delGTTCTG", "c.3659C>T", "c.3842G>A", "c.4152T>G", "c.845delT"}
180 - 180	ATP8B1:Cholestasis, progressive familial intrahepatic 1 {(NM_005603) "c.2854C>T"}
181 - 181	AVP:Familial neurohypophyseal diabetes insipidus {(NM_000490) "c.77C>T"}
182 - 182	B3GALNT2:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11) {(NM_152490.5) "c.236-1G>C"}
183 - 183	B4GALT1:Congenital disorder of glycosylation, type IId {(NM_001497) "c.61C>T"}
184 - 186	BBS10:Bardet-Biedl syndrome 10 {(NM_024685.4) "c.1091delA", "c.1399delA", "c.271dupT"}
187 - 188	BBS1:Bardet-Biedl syndrome 1 {(NM_024649.5) "c.1169T>G", "c.479G>A"}
189 - 193	BBS2:Bardet-Biedl syndrome 2 {(NM_031885.4) "c.1895G>C", "c.224T>G", "c.311A>C", "c.401C>G", "c.98C>A"}
194 - 195	BBS4:Bardet-Biedl syndrome 4 {(NM_033028.5) "c.77-1421_221-1229del5784 ", "c.884G>C"}
196 - 196	BBS7:Bardet-Biedl syndrome 7 {(NM_176824.3) "c.1786G>A"}
197 - 198	BBS9:Bardet-Biedl syndrome 9 {(NM_014451) "c.1063C>T", "c.1669+1G>A"}
199 - 205	BCKDHA:Maple syrup urine disease, type Ia {(NM_000709.4) "c.169delG", "c.718del", "c.792C>G", "c.859C>T", "c.890G>A", "c.909_910delGT", "c.935_937del"}
206 - 212	BCKDHB:Maple syrup urine disease, type Ib {(NM_000056.4) "c.1016C>T", "c.1114G>T", "c.356T>G", "c.548G>C", "c.670C>T", "c.800_803delAGGA", "c.832G>A"}
213 - 219	BLM:Bloom syndrome {(NM_000057) "c.1642C>T", "c.2512C>T" | (NM_000057.4) "c.1984_1985delAA", "c.2207_2212delATCTGAinsTAGATTC", "c.2407dupT", "c.3510T>A", "c.98+1G>T"}
220 - 220	BMPER:Diaphanospondylodysostosis {(NM_133468.5) "c.310C>T"}
221 - 221	BMPR1B:Brachydactyly type A2 {(NM_001256793.2) "c.377G>A"}
222 - 223	BSND:Bartter syndrome, type 4a infantile variant with

	sensorineuronal deafness {(NM_057176.3) "c.167_168insTTTCCC", "c.28G>A"}
224 - 225	BTD:Biotinidase deficiency {(NM_000060) "c.393delC" | (NM_000060.4) "c.100G>A"}
226 - 228	C12ORF65:Spastic paraplegia 55, Autosomal Recessive {(NM_152269) "c.346delG" | (NM_152269.5) "c.282+2T>A", "c.413_417delAACAA"}
229 - 229	C21orf59:Ciliary dyskinesia, primary, 26 {(NM_021254.4) "c.735C>G"}
230 - 235	C2ORF71:Retinitis pigmentosa 54 {(NM_001029883) "c.2334T>A", "c.2756_2768delAGCCAGCCCTGGA", "c.3289C>T", "c.478_479insA", "c.556C>T", "c.776_777delAG"}
236 - 238	C8orf37:Retinitis pigmentosa 64 {(NM_177965.4) "c.497T>A", "c.529C>T", "c.545A>G"}
239 - 241	CAPN3:Muscular dystrophy, limb-girdle, type 2A {(NM_000070) "c.1076C>T", "c.1469G>A", "c.367C>A"}
242 - 242	CASQ2:Ventricular tachycardia, catecholaminergic polymorphic, 2 {(NM_001232.3) "c.919G>C"}
243 - 247	CBS:Homocystinuria, thrombosis, hyperhomocysteinemic {(NM_000071.2) "c.1006C>T", "c.1224-2A>C", "c.1261delG", "c.785C>G" | (NM_001178008.2) "c.919G>A"}
248 - 248	CC2D1A:Mental retardation, Autosomal Recessive 3 - MRT3 {(NM_017721.5) "c.1468+1_1824-1del"}
249 - 249	CC2D2A:Mental retardation, Autosomal Recessive 3 - MRT3 {(NM_001080522) "c.308delG"}
250 - 250	CCDC114:Ciliary dyskinesia, primary, 20 {(NM_144577) "c.939delT"}
251 - 251	CCDC174:Birk Volodarsky PMR Synderome Hypotonia and psychomotor developmental delay {(NM_016474.5) "c.1404A>G"}
252 - 252	CCDC65:Ciliary dyskinesia, primary, 27 {(NM_033124.5) "c.877_878delAT"}
253 - 253	CCDC88C:Hydrocephalus, nonsyndromic, Autosomal Recessive {(NM_001080414.4) "c.934C>T"}
254 - 257	CCNO:Ciliary dyskinesia, primary, 29 {(NM_021147) "c.165delC", "c.258_262dupGGCCC", "c.481_482delCT", "c.638T>C"}
258 - 258	CD59:Hemolytic anemia & immune-mediated polyneuropathy, CD59-related {(NM_203330.2) "c.266G>A"}
259 - 259	CDAN1:Dyserythropoietic anemia, congenital, type Ia {(NM_138477.4) "c.3124C>T"}
260 - 260	CDH23:Usher Syndrome Type ID {(NM_022124.6) "c.7903G>T"}
261 - 261	CDK10:Al Kaissi syndrome {(NM_052988) "c.870_871del"}
262 - 262	CDK5:Lissencephaly 7 with cerebellar hypoplasia {(NM_004935.4) "c.580+1G>A"}
263 - 263	CEACAM16:Deafness, autosomal recessive {(NM_001039213) "c.703C>T"}
264 - 264	CECR1:Adenosine deaminase 2 deficency {(NM_001282225.2) "c.140_141insT"}
265 - 265	CECR1:Polyarteritis nodosa, childhood-onset {(NM_001282225.2) "c.139G>A"}
266 - 266	CENPJ:Microcephaly, primary, Autosomal Recessive {(NM_018451.5)

	"c.3243_3246delTCAG"}
267 - 267	CEP104:Joubert syndrome (JBTS) {(NM_014704.4) "c.1328_1329insT"}
268 - 268	CEP152:Microcephaly 9, primary, Autosomal Recessive {(NM_014985.3) "c.2281-2A>G"}
269 - 277	CEP290:Meckel syndrome 4 {(NM_025114) "c.5668G>T", "c.5824C>T", "c.6760A>T" | (NM_025114.3) "c.1225delA", "c.164_167delCTCA", "c.1666delA", "c.4393C>T", "c.4771C>T", "c.5788A>T"}
278 - 278	CERKL:Retinitis pigmentosa 26 {(NM_001030311.2) "c.238+1G>A"}
279 - 279	CFH:Hemolytic uremic syndrome, complement factor H deficiency {(NM_000186.3) "c.3677_*4del and c.3674A>T"}
280 - 709	CFTR:Cystic fibrosis {(NM_000492) "c.220C>T", "c.54-5811_164+2186del8108ins182" | (NM_000492.3) "c.1000C>T", "c.1001G>A", "c.1007T>A", "c.1013C>T", "c.1021T>C", "c.1021_1022dupTC", "c.1040G>A", "c.1040G>C", "c.1040G>T", "c.1055G>A", "c.1075C>A", "c.1081delT", "c.1116+1G>A", "c.1130dupA", "c.1155_1156dupTA", "c.115C>T", "c.11C>A", "c.1202G>A", "c.1203G>A", "c.1209+1G>A", "c.1240C>T", "c.1327_1330dupGATA", "c.1340delA", "c.1364C>A", "c.137C>A", "c.1393-1G>A", "c.1397C>A", "c.1397C>G", "c.1400T>C", "c.1418delG", "c.1438G>T", "c.1439G>A", "c.1466C>A", "c.1475C>T", "c.1477C>T", "c.1521_1523delCTT", "c.1545_1546delTA", "c.1558G>A", "c.1558G>T", "c.1572C>A", "c.1573C>T", "c.1585-1G>A", "c.1585-8G>A", "c.1624G>T", "c.1625G>A", "c.1645A>C", "c.1646G>A", "c.1646G>T", "c.1647T>G", "c.165-1G>A", "c.1650delA", "c.1651G>A", "c.1652G>A", "c.1654C>T", "c.1657C>T", "c.166G>A", "c.1673T>C", "c.1675G>A", "c.1679+1634A>G", "c.1679+1G>C", "c.1679G>A", "c.1679G>C", "c.1680-1G>A", "c.1682C>A", "c.1692delA", "c.1736A>G", "c.1753G>T", "c.175dupA", "c.1766+1G>A", "c.1766+1G>C", "c.1766+3A>G", "c.1766+5G>T", "c.178G>T", "c.1817_1900del84", "c.1841A>G", "c.1911delG", "c.1923_1931delCTCAAAACTinsA", "c.1973_1985delGAAATTCAATCCTinsAGAAA", "c.1976delA", "c.1986_1989delAACT", "c.1A>G", "c.200C>T", "c.2012delT", "c.2051_2052delAAinsG", "c.2052_2053insA", "c.2052delA", "c.2089dupA", "c.2125C>T", "c.2128A>T", "c.2175dupA", "c.2195T>G", "c.2215delG", "c.223C>T", "c.2290C>T", "c.233dupT", "c.2353C>T", "c.2374C>T", "c.2423_2424dupAT", "c.2453delT", "c.2463_2464delTG", "c.2464G>T", "c.2490+1G>A", "c.2491G>T", "c.2537G>A", "c.2547C>A", "c.254G>A", "c.2551C>T", "c.2583delT", "c.2619+1G>A", "c.2619+2dupT", "c.262_263delTT", "c.2657+2_2657+3insA", "c.2657+5G>A", "c.2658-1G>C ", "c.2668C>T", "c.273+1G>A", "c.273+3A>C", "c.2737_2738insG", "c.2739T>A", "c.274-1G>A", "c.274G>A", "c.274G>T", "c.2763_2764dupAG", "c.2780T>C", "c.2834C>T", "c.2856G>C", "c.2875delG", "c.2908G>C", "c.292C>T", "c.2930C>T", "c.2988+1173_c.3468+2111del8898", "c.2988+1G>A", "c.2988G>A", "c.2989-1G>A", "c.2989-977_3367+248del", "c.3002_3003delTG",

	"c.3039delC", "c.3041A>G", "c.3067_3072delATAGTG", "c.3139+10T>C", "c.313delA", "c.3140-26A>G", "c.3154T>G", "c.3160C>G", "c.3181G>C", "c.3194T>C", "c.3196C>T", "c.3197G>A", "c.3205G>A", "c.3208C>T", "c.3209G>A", "c.3222T>A", "c.3230T>C", "c.325_327delTATinsG", "c.3266G>A", "c.3276C>G", "c.328G>C", "c.3292T>C", "c.3299A>C", "c.3302T>A", "c.3310G>T", "c.343G>T", "c.3454G>C", "c.3469-2A>G", "c.3472C>T", "c.3484C>T", "c.349C>T", "c.350G>A", "c.3528delC", "c.3529A>T", "c.3532_3535dupTCAA", "c.3535_3538delACCA", "c.3587C>G", "c.3600delA", "c.3605delA", "c.3611G>A", "c.3612G>A", "c.3659delC", "c.366T>A", "c.3691delT ", "c.3700A>G", "c.3712C>T", "c.3718-1G>A", "c.3718-2477C>T", "c.3731G>A", "c.3744delA", "c.3752G>A", "c.3763T>C", "c.3764C>A", "c.3764C>T", "c.3773dupT", "c.3793G>A", "c.3808delG", "c.3846G>A", "c.3873+1G>A", "c.3883_3884insG", "c.3883_3886delATTT", "c.3883delA", "c.3889dupT", "c.3909C>G", "c.3937C>T", "c.3964-78_4242+577del", "c.4046G>A", "c.4077_4080delTGTTinsAA", "c.4111G>T", "c.413_415dupTAC", "c.416A>C", "c.416A>T", "c.4197_4198delCT", "c.422C>A", "c.4234C>T", "c.4242+1G>T", "c.4251delA", "c.4297G>A", "c.4300_4301dupAG", "c.4364C>G", "c.442delA", "c.487A>G", "c.489+1G>T", "c.523A>G", "c.531delT", "c.532G>A", "c.54-5940_273+10250del21Kb", "c.543_546delTAGT", "c.575A>G", "c.579+1G>T", "c.579+3A>G", "c.579+5G>A", "c.580-1G>T", "c.595C>T", "c.613C>T", "c.617T>G", "c.658C>T", "c.675T>A", "c.761delA", "c.803delA", "c.805_806delAT", "c.828C>A", "c.870-2A>G", "c.892delA", "c.933_935delCTT", "c.948delT", "c.988G>T", "c.[1075C>A;1079C>A]" | (NM_000492.4) "c.-9_14del23", "c.1001G>T", "c.1006_1007insG", "c.1029delC", "c.1037T>C", "c.1046C>T", "c.1054C>T", "c.1079C>A", "c.1117-1G>A", "c.1211delG", "c.1301_1307delCACTTCT", "c.1327G>T", "c.1358T>C", "c.1365_1366delGG", "c.1367T>C", "c.1373delG", "c.1393-2A>G", "c.1408G>C", "c.1408G>T", "c.1420G>A", "c.1477_1478delCA", "c.1487G>A", "c.14C>T", "c.1505T>C", "c.1519_1521delATC", "c.1538A>G", "c.1584+1G>A", "c.164+1G>A", "c.164+1G>T", "c.164+2T>C", "c.164+3_164+4insT", "c.1648G>T", "c.165-3C>T", "c.1670delC", "c.1679+1G>A", "c.1680-877G>T", "c.1680A>C", "c.1687T>A", "c.1687T>G", "c.169T>G", "c.1703delT", "c.1705T>G", "c.170G>A or c.171G>A", "c.1721C>A", "c.1724T>A", "c.174_177delTAGA", "c.1763A>T", "c.1766+1G>T", "c.178G>A", "c.1792_1798delAAAACTA", "c.1801A>T", "c.1826A>G", "c.1837G>A", "c.1853T>C", "c.1865G>A", "c.1882G>C or c.1882G>A", "c.1923_1931del9insA", "c.1943delA", "c.1973_1985del13insAGAAA", "c.2017G>T", "c.2053C>T", "c.2053dupC", "c.2143C>T", "c.2158C>T", "c.2241_2248delGATACTGC", "c.2249C>T", "c.2537G>A or c.2538G>A", "c.2589_2599delAATTTGGTGCT", "c.2601dupA", "c.263T>A", "c.263T>G", "c.2645G>A", "c.271G>A", "c.274-2A>G", "c.2770G>A", "c.2810dupT", "c.2825delT", "c.2855T>C", "c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC", "c.2896delA", "c.2900T>C", "c.2909G>A", "c.2936A>T", "c.296C>T", "c.2989-2A>G", "c.3011_3019delCTATAGCAG or c.3009_3017delAGCTATAGC", "c.3017C>A", "c.3039dupC",

	"c.3047T>C", "c.305T>G", "c.3095A>G", "c.3107C>A", "c.310delA", "c.3124C>T", "c.3139_3139+1delGG", "c.3217dupT", "c.3220T>C", "c.3222T>G", "c.3276C>A", "c.3293G>A or c.3294G>A", "c.3294G>C or c.3294G>T", "c.3297C>A", "c.3302T>G", "c.3304A>T", "c.330C>A", "c.3353C>T", "c.3368-2A>G", "c.3435G>A", "c.3458T>A", "c.3468+2dupT", "c.3468+5G>A", "c.3468G>A", "c.3475T>C", "c.3476C>T", "c.3485G>T", "c.349C>G", "c.350G>C", "c.350G>T", "c.358G>A", "c.3717+40A>G", "c.3717+4A>G", "c.3717+5G>A", "c.3717G>A", "c.3718-3T>G", "c.3719T>G", "c.3737C>T", "c.3745G>A", "c.3747delG", "c.3761T>G", "c.377G>A", "c.3806T>A", "c.3848G>T", "c.3872A>G", "c.3873+2T>C", "c.3873G>C", "c.3891dupT", "c.38C>T", "c.3908delA", "c.3971T>C", "c.3988C>T", "c.4004T>C", "c.4036_4042del", "c.4086dupT", "c.4097T>A", "c.409delC", "c.4124A>C", "c.4127_4131delTGGAT", "c.4144C>T", "c.4147dupA", "c.416A>G", "c.4231C>T", "c.4242+1G>A", "c.4426C>T", "c.44T>C", "c.470_483delTTAGTTTGATTTAT", "c.481T>G", "c.489+3A>G", "c.494T>C", "c.4C>T", "c.50delT", "c.53+1G>T", "c.571T>G", "c.577G>T", "c.57G>A", "c.580G>A", "c.581G>T", "c.601G>A", "c.647G>A", "c.680T>G", "c.695T>A", "c.709C>G", "c.717delG", "c.772A>G", "c.794T>G", "c.79G>A", "c.79G>T", "c.825C>G", "c.850dupA", "c.861_865delCTTAA", "c.88C>T", "c.92G>T", "c.933C>G", "c.941G>A", "c.987delA"}
710 - 713	CHRNE:Myasthenic syndrome, congenital, 4B, fast-channel {(NM_000080) "c.1161_1162insT", "c.1353dupG", "c.187_188insC", "c.637dupG"}
714 - 719	CLCN1:Myotonia congenita, Autosomal Recessive {(NM_000083) "c.1012C>T", "c.1437_1450del", "c.1444G>A", "c.1586C>T", "c.568_569delGGinsTC", "c.803C>T"}
720 - 725	CLCN5:Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis {(NM_000084.4) "c.1245delG", "c.1399C>T", "c.161dup", "c.1909C>T", "c.258delA", "c.82C>T"}
726 - 727	CLCNKB:Bartter syndrome, type 3 and Gitelman syndrome {(NM_000085) "c.1830G>A" | (NM_000085.4) "c.1313G>A"}
728 - 728	CLN5:Ceroid lipofuscinosis, neuronal, 5 {(NM_006493.3) "c.672delG"}
729 - 730	CLN6:Ceroid lipofuscinosis, neuronal, 6 {(NM_017882) "c.843G>A" | (NM_017882.3) "c.214G>T"}
731 - 731	CLN8:Neuronal ceroid lipofuscinosis type 8, including northern epilepsy {(NM_018941.3) "c.766C>G"}
732 - 735	CLRN1:Usher syndrome, type 3A {(NM_174878.2) "c.144T>G", "c.349_358del", "c.433+1G>A", "c.528T>G"}
736 - 737	CNGA1:Retinitis pigmentosa 49 {(NM_000087) "c.1540C>T", "c.94C>T"}
738 - 746	CNGA3:Achromatopsia-2 - total color blindness {(NM_001298) "c.1294delG", "c.1306C>T", "c.829C>T" | (NM_001298.2) "c.1114C>T", "c.1585G>A", "c.1640T>G", "c.67C>T", "c.940_942delATC", "c.985G>T"}
747 - 748	CNGB1:Retinitis pigmentosa 45 {(NM_001297) "c.2760G>A" | (NM_001297.5) "c.2284C>T"}
749 - 756	CNGB3:Achromatopsia-3,macular degeneration, juvenile

	{(NM_019098) "c.1207C>T", "c.2328delC", "c.41_42dupTA", "c.467C>T", "c.819delC" | (NM_019098.4) "c.1006G>T", "c.1148delC", "c.644-1G>C"}
757 - 758	CNNM4:Jalili syndrome {(NM_020184.4) "c.1813C>T", "c.599C>A"}
759 - 759	CNTNAP1:Lethal congenital contracture syndrome 7 {(NM_003632) "c.2015G>A"}
760 - 760	COL11A2:Otospondylomegaepiphyseal dysplasia (ZW) {(NM_080680.2) "c.3991C>T"}
761 - 765	COL17A1:Epidermolysis bullosa, junctional, {(NM_000494) "c.2226insTGGA", "c.3676C>T", "c.3766+1G>A", "c.4145_4148delAGAG", "c.737_738insA"}
766 - 769	COL4A3:Alport Syndrome, COL4A3-Related {(NM_000091.4) "c.1791_1793dupTCC", "c.227delG", "c.3518-2A>G", "c.4649T>G"}
770 - 771	COL4A4:Alport syndrome, COL4A4-Related {(NM_000092.4) "c.3933C>G", "c.785_792dupCACCTGAC"}
772 - 777	COL4A5:Alport syndrome, COL4A5-Related {(NM_000495) "c.1571delG" | (NM_000495.4) "c.2641G>T", "c.367delG", "c.4691G>C", "c.4946T>G", "c.5030G>A"}
778 - 781	COL7A1:Dystrophic epidermolysis bullosa, Autosomal Recessive,COL7A1-Related {(NM_000094) "c.2387G>A", "c.4888C>T", "c.6341delG", "c.682+1G>A"}
782 - 782	COLEC11:3MC syndrome 2 {(NM_199235.2) "c.627_628delCG"}
783 - 788	COLQ:Myasthenic syndrome, congenital, 5 {(NM_005677) "c.377delG", "c.788dupC", "c.893delA" | (NM_005677.4) "c.1228C>T", "c.718G>T", "c.792dupG"}
789 - 789	COQ4:Coenzyme Q10 deficiency, primary, 7 {(NM_016035.5) "c.718C>T"}
790 - 795	CPS1:Carbamoylphosphate synthetase I deficiency {(NM_001875.5) "c.1760G>A", "c.3265C>T", "c.3374C>T", "c.3558+1G>C", "c.4101+2T>C", "c.794C>T"}
796 - 796	CPT1A:Carnitine palmitoyltransferase 1 deficiency {(NM_001031847) "c.1361A>G"}
797 - 798	CPT2:CPT deficiency, hepatic, type II {(NM_000098) "c.110_111dupGC", "c.1239_1240delGA"}
799 - 813	CRB1:Leber congenital amaurosis 8 {(NM_201253.3) "c.1148G>A", "c.1576C>T", "c.1733T>A", "c.1842delT", "c.1844G>T", "c.2230C>T", "c.2234C>T", "c.2498G>A", "c.2555T>C", "c.2680_2684delAACCC", "c.3307G>A", "c.4005+1G>A", "c.4121_4130delCAACTCAGGG", "c.424G>T", "c.455G>A"}
814 - 817	CRB2:Ventriculomegaly with cystic kidney disease {(NM_173689.7) "c.1882C>T", "c.1928A>C", "c.2277G>A", "c.2400C>G"}
818 - 819	CRTAP:Osteogenesis imperfecta, type VII {(NM_006371) "c.976C>T" | (NM_006371.4) "c.793+1G>T"}
820 - 820	CSTA:Exfoliative ichthyosis, Autosomal Recessive, ichthyosis bullosa of Siemens-like {(NM_005213.4) "c.67-2A>T"}
821 - 825	CTNS:Cystinosis,CTNS-related {(NM_004937) "c.587dupA", "c.691C>T", "c.890G>A" | (NM_004937.2) "c.1015G>A", "c.530A>C"}
826 - 826	CTSC:Haim-Munk syndrome {(NM_001814.6) "c.857A>G"}

827 - 827	CTSK:Pycnodysostosis {(NM_000396.4) "c.990A>G"}
828 - 832	CYBA:Chronic granulomatous disease,autosomal, due to deficiency of CYBA {(NM_000101) "c.160_161insC" | (NM_000101.4) "c.164C>G", "c.171dupG", "c.70G>A", "c.71G>A"}
833 - 837	CYBB:Chronic granulomatous disease, X-linked {(NM_000397) "c.1016dupC", "c.1081T>C", "c.271C>T", "c.676C>T", "c.90_92delCCGinsGGT"}
838 - 839	CYP11A1:Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete {(NM_000781.3) "c.644T>C", "c.694C>T"}
840 - 840	CYP11B2:Hypoaldosteronism, congenital, due to CMO II deficiency {(NM_000498.3) "c.541C>T"}
841 - 843	CYP1B1:Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset {(NM_000104) "c.1405C>T" | (NM_000104.3) "c.1568G>A", "c.182G>A"}
844 - 848	CYP27A1:Cerebrotendinous xanthomatosis {(NM_000784.4) "c.1016C>T", "c.1184G>A", "c.355delC", "c.819delT", "c.845-1G>A"}
849 - 849	CYP4F22:Congenital recessive ichthyoses (CRI) {(NM_173483) "c.429dupG"}
850 - 850	CYP4V2:Bietti crystalline corneoretinal dystrophy {(NM_207352) "c.1123delC"}
851 - 851	CYP7B1:Spastic paraplegia 5A, Autosomal Recessive {(NM_004820.5) "c.1081C>T"}
852 - 852	DAG1:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 {(NM_001165928.3) "c.743delC"}
853 - 853	DARS2:Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation {(NM_018122.5) "c.492+2T>C"}
854 - 855	DBT:Maple syrup urine disease, type II {(NM_001918) "c.581C>G", "c.939G>C"}
856 - 857	DCAF17:Woodhouse-Sakati syndome {(NM_025000) "c.580C>T" | (NM_025000.4) "c.436delC"}
858 - 858	DCLRE1C:Severe combined immunodeficiency, Athabascan type {(NM_001033858.2) "c.1307_1308insAGGATGCT"}
859 - 859	DDR2:Spondylometaepiphyseal dysplasia, short limb-hand type {(NM_006182.4) "c.2254C>T"}
860 - 860	DDRGK1:Spondyloepimetaphyseal dysplasia (Shohat-type) {(NM_023935) "c.408+1G>A"}
861 - 861	DDX11:Warsaw breakage syndrome {(NM_030653.3) "c.1763-1G>C"}
862 - 862	DGAT1:Diarrhea 7, congenital {(NM_012079.6) "c.751+2T>C"}
863 - 864	DGUOK:Mitochondrial DNA depletion syndrome (hepatocerebral type) {(NM_080916.3) "c.255delA", "c.271delA"}
865 - 865	DHCR24:Desmosterolosis {(NM_014762.4) "c.307C>T"}
866 - 882	DHCR7:Smith Lemli Opitz syndrome {(NM_001360) "c.1055G>A", "c.1342G>A" | (NM_001360.2) "c.1054C>T", "c.1210C>T", "c.1228G>A", "c.1A>G", "c.278C>T", "c.452G>A", "c.453G>A", "c.506C>T", "c.724C>T", "c.725G>A", "c.755A>G", "c.906C>G", "c.964-1G>C", "c.964-1G>T", "c.976G>T"}
883 - 883	DHDDS:Retinitis pigmentosa 59 {(NM_024887.3) "c.124A>G"}
884 - 887	DLD:Dihydrolipoamide Dehydrogenase Deficiency {(NM_000108.5)

	"c.104dupA", "c.1123G>A", "c.1436A>T", "c.685G>T"}
888 - 888	DLL3:Spondylocostal dysostosis 1, Autosomal Recessive {(NM_016941.3) "c.395delG"}
889 - 890	DNAH11:Ciliary dyskinesia, primary, 7, with or without situs inversus {(NM_001277115.2) "c.11929G>T", "c.13242_13245delAAAG"}
891 - 892	DNAH5:Ciliary dyskinesia, primary, 3, with or without situs inversus (CILD3/PCD) {(NM_001369.2) "c.7502G>C", "c.8011-2A>G"}
893 - 893	DNAI1:Ciliary dyskinesia, primary, 1, with or without situs inversus {(NM_012144.4) "c.1490G>A"}
894 - 895	DNAI2:Ciliary dyskinesia, primary, 9, with or without situs inversus {(NM_023036.6) "c.1304G>A", "c.1494+1G>A"}
896 - 896	DNAL1:Ciliary dyskinesia, primary, 16 {(NM_031427.4) "c.449A>G"}
897 - 897	DOCK8:Hyper-IgE recurrent infection syndrome, autosomal recessive {(NM_203447) "c.5132C>A"}
898 - 899	DOLK:Congenital disorder of glycosylation, type Im {(NM_014908.3) "c.1222C>G", "c.912G>T"}
900 - 901	DSG1:Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE {(NM_001942.4) "c.1861delG", "c.395C>A"}
902 - 902	DST:Epidermolysis bullosa simplex, Autosomal Recessive 2 {(NM_183380.3) "c.14865delA"}
903 - 903	DSTYK:Spastic paraplegia, complicated {(NM_015375) "4-kbdeletion/20-bpinsertion"}
904 - 909	DYSF:Muscular dystrophy, limb-girdle, type 2B {(NM_003494.4) "c.2372C>G", "c.2779delG", "c.4741C>T", "c.4872_4876delGCCCGinsCCCC", "c.5057+5G>A", "c.5429G>A"}
910 - 910	ECHS1:Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency {(NM_004092) "c.476A>G"}
911 - 911	ECM1:Urbach-Wiethe disease {(NM_004425) "c.70+1G>C"}
912 - 912	EDAR:Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, Autosomal Recessive {(NM_022336) "c.259T>C"}
913 - 915	ELP1:Dysautonomia, familial {(NM_003640.5) "c.2087G>C", "c.2204+6T>C", "c.2741C>T"}
916 - 916	EOGT:Adams-Oliver syndrome 4 {(NM_001278689.2) "c.1074delA"}
917 - 920	EPG5:Vici syndrome {(NM_020964) "c.1007A>G", "c.3446G>A", "c.5993C>G" | (NM_020964.3) "c.5704dupT"}
921 - 921	EPM2A:Epilepsy, progressive myoclonic 2A (Lafora) {(NM_005670) "56_kb_incl._ex._2"}
922 - 922	ERBB3:Lethal congenital contractural syndrome 2 {(NM_001982.3) "c.1184-9A>G"}
923 - 923	ERCC2:Xeroderma pigmentosum, group D {(NM_000400.3) "c.2048G>A"}
924 - 924	ERCC5:Xeroderma pigmentosum/Cockayne {(NM_000123.3) "c.205C>T"}
925 - 925	ERCC6:Cockayne syndrome, type B {(NM_000124.4) "c.1034_1035insT"}
926 - 928	ERCC8:Cockayne syndrome, type A {(NM_000082.3) "c.37G>T", "c.843+1G>C", "c.966C>A"}

929 - 929	ESCO2:Roberts-SC phocomelia syndrome {(NM_001017420.3) "c.1674-2A>G"}
930 - 933	ETFDH:Glutaric acidemia IIC {(NM_004453.4) "c.1074G>C", "c.1084G>A", "c.1425C>A", "c.299T>A"}
934 - 934	EXOSC3:Pontocerebellar hypoplasia, type 1B {(NM_016042.4) "c.571G>T"}
935 - 935	EXOSC8:Pontocerebellar hypoplasia, type 1C {(NM_181503.3) "c.5C>T"}
936 - 950	EYS:Retinitis pigmentosa 25 {(NM_001142800.2) "400kb deletion in 6q12", "c.1211dupA", "c.3699delG", "c.3715G>T", "c.403delA", "c.410_424del15", "c.4361_4362delinsAG", "c.5450G>A", "c.5928-37922_6078+38716del", "c.6976C>T", "c.8155_8156delCA", "c.8168delA", "c.8216_8217delAC", "c.8231del", "c.9286_9295del10"}
951 - 952	F7:Factor VII deficiency {(NM_000131) "c.1109G>T" | (NM_000131.4) "c.1256C>T"}
953 - 953	FA2H:Spastic paraplegia 35, Autosomal Recessive {(NM_024306.5) "c.786+1G>A"}
954 - 960	FAH:Tyrosinemia, type I {(NM_000137.2) "c.1062+5G>A", "c.1069G>T", "c.192G>T", "c.554-1G>T", "c.707-1G>C", "c.782C>T", "c.786G>A"}
961 - 966	FAM161A:Retinitis pigmentosa 28 {(NM_001201543.2) "c.1003C>T", "c.1309A>T", "c.1321dupC", "c.1355_1356delCA", "c.1567C>T", "c.1786C>T"}
967 - 967	FAM20A:Amelogenesis imperfecta, type IG (enamel-renal syndrome) {(NM_017565.4) "c.1523delC"}
968 - 976	FANCA:Fanconi anemia, complementation group A {(NM_000135) "c.3382C>T" | (NM_000135.4) "c.189+1G>A", "c.2172dupG", "c.3788_3790delTCT", "c.4168-2A>C", "c.4261-2A>C", "c.4275delT", "c.891_893+1delCTGG", "c.Del_exon_31-37"}
977 - 983	FANCC:Fanconi anemia, complementation group C {(NM_000136) "c.8_9delAA" | (NM_000136.3) "c.1642C>T", "c.1661T>C", "c.37C>T", "c.456+4A>T", "c.553C>T", "c.67delG"}
984 - 985	FANCG:Fanconi Anemia - complementation group G {(NM_004629.1) "c.212T>C", "c.510+3A>G"}
986 - 986	FDX1L:Mitochondrial muscle myopathy {(NM_001031734.4) "c.10A>T"}
987 - 989	FERMT1:Kindler syndrome {(NM_017671.4) "c.0_-19+470del ", "c.137_140delTAGT", "c.749G>A"}
990 - 990	FGB:Afibrinogenemia congenital {(NM_005141.4) "c.1400G>A"}
991 - 991	FH:Fumarase deficiency, leiomyomatosis and renal cell cancer {(NM_000143.3) "c.905-1G>A"}
992 - 994	FKBP10:Osteogenesis imperfecta, type XI {(NM_021939) "c.1271_1272delCCinsA", "c.391+4A>T" | (NM_021939.3) "c.310C>T"}
995 - 995	FKRP:Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 {(NM_024301.5) "c.160C>T"}
996 - 996	FKTN:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 - Walker Warburg syndrome {(NM_001079802.1) "c.1167dupA"}

997 - 997	FLT4:Autosomal Recessive Hereditary Lymphedema {(NM_182925.5) "c.3704C>G"}
998 - 998	FOXRED1:Mitochondrial encephalomyopathy complex I deficiency {(NM_017547.4) "c.1054C>T"}
999 - 999	FRMD4A:Microcephaly intellectual disability and dysmorphism {(NM_018027) "c.2134_2146dup13"}
1000 - 1000	FTO:Growth retardation, developmental delay, coarse facies, and early death {(NM_001080432.3) "c.947G>A"}
1001 - 1002	G6PC3:Neutropenia, severe congenital 4, Autosomal Recessive {(NM_138387.3) "c.765_766delAG", "c.785G>A"}
1003 - 1014	G6PC:Glycogen storage disease Ia - GDS1a {(NM_000151.4) "c.1039C>T", "c.247C>T", "c.248G>A", "c.379_380dupTA", "c.497T>G", "c.508C>T", "c.562G>C", "c.648G>T", "c.724C>T", "c.79delC", "c.809G>T", "c.979_981delTTC"}
1015 - 1027	GAA:Pompe (Glycogen storage disease type II) {(NM_000152) "c.1001G>A", "c.2456G>T" | (NM_000152.5) "c.1064T>C", "c.1082C>T", "c.1210G>A", "c.1564C>A", "c.1935C>A", "c.1942G>A", "c.2560C>T", "c.340_341insT", "c.670C>T", "c.896T>C", "c.896T>G"}
1028 - 1030	GALC:Krabbe disease {(NM_000153.4) "c.1630G>A", "c.1748A>C", "c.1796T>G"}
1031 - 1032	GALNT3:Tumoral calcinosis, hyperphosphatemic, familial {(NM_004482.4) "c.1524+1G>A", "c.1524+5G>A"}
1033 - 1042	GALT:Galactosemia {(NM_000155.3) "5.5-KB_DEL", "c.152G>A", "c.253-2A>G", "c.404C>T", "c.413C>T", "c.512T>C", "c.563A>G", "c.584T>C", "c.626A>G", "c.855G>T"}
1043 - 1044	GAN:Giant axonal neuropathy 1 {(NM_022041) "c.103G>T" | (NM_022041.3) "c.973G>A"}
1045 - 1045	GATC:Hypertophic Cardiomyopathy {(NM_176818) "c.233T>G"}
1046 - 1046	GATM:Cerebral creatine deficiency syndrome 3 {(NM_001482.3) "c.1111dupA"}
1047 - 1058	GBA:Gaucher disease, , type I {(NM_001005741.3) "c.115+1G>A", "c.1226A>G", "c.1294T>A", "c.1297G>T", "c.1342G>C", "c.1448T>C", "c.1504C>T", "c.1505G>A", "c.1604G>A", "c.259C>T", "c.703T>C", "c.84dupG"}
1059 - 1070	GCDH:Glutaricaciduria type I {(NM_000159.4) "c.1168G>C", "c.1173delG", "c.1204C>T", "c.1205G>A", "c.1247C>T", "c.1262C>T", "c.1306G>T", "c.301G>A", "c.505+1G>A", "c.848T>C", "c.877G>A", "c.914C>T"}
1071 - 1072	GH1:Growth hormone deficiency, isolated, type IA {(NM_000515.5) "c.456+5G>C", "c.67G>T"}
1073 - 1078	GHR:Laron dwarfism {(NM_000163.5) "c.11G>A", "c.594A>G", "c.62G>A", "c.703C>T", "c.744delT", "del5,6ex"}
1079 - 1079	GHRHR:Growth hormone deficiency, isolated, type IB {(NM_000823.4) "c.1069C>T"}
1080 - 1080	GIPC3:Deafness, autosomal recessive 15 {(NM_133261) "c.937T>C"}
1081 - 1095	GJB2:Deafness, autosomal recessive 1A {(NM_004004.6) "c.109G>A", "c.167delT", "c.229T>C", "c.230G>A", "c.235delC", "c.250G>A", "c.269T>C", "c.358_360delGAG", "c.35delG", "c.370C>T",

	"c.51_62delCACCAGCATTGGinsA", "c.551G>C", "c.614T>C", "c.71G>A", "c.94C>T"}
1096 - 1096	GJB6:Deafness, Autosomal Recessive 1B {(NM_006783.4) "309_kb"}
1097 - 1102	GLB1:GM1-gangliosidosis, type I {(NM_000404.4) "c.1038G>C", "c.485delT", "c.602G>A", "c.824A>G", "c.827A>C", "c.914+4A>G"}
1103 - 1106	GLDC:Glycine encephalopathy and non-ketoic hyperglycinemia, GLDC-related {(NM_000170.2) "c.2405C>T", "c.2607C>A", "c.2T>C", "c.985C>A"}
1107 - 1107	GLRA1:Hyperekplexia, hereditary 1, autosomal dominant or recessive {(NM_001146040.1) "c.298C>T"}
1108 - 1108	GMPPA:Alacrima, achalasia, and mental retardation syndrome {(NM_013335.3) "c.1000A>C"}
1109 - 1110	GMPPB:Muscular dystrophy-dystroglycanopathy {(NM_013334.3) "c.656T>C", "c.860G>A"}
1111 - 1111	GNE:Hereditary inclusion body myopathy (HIBM) {(NM_005476.6) "c.2135T>C"}
1112 - 1118	GNPTAB:Mucolipidosis III alpha/beta {(NM_024312.5) "c.118-2A>G", "c.2314_2315insA", "c.2918dupT", "c.3434+1G>A", "c.3434+715G>A", "c.3503_3504delTC", "c.3613C>T"}
1119 - 1119	GNPTG:Mucolipidosis III gamma {(NM_032520.5) "c.499dupC"}
1120 - 1121	GPC6:Omodysplasia 1 {(NM_005708) "g.93997007_94063501del66495insATAAATCACTTAGAGATGT", "g.94252984_94352299del99316insCTA"}
1122 - 1122	GPSM2:Chudley-McCullough syndrome {(NM_013296.5) "c.379C>T"}
1123 - 1123	GRHPR:Hyperoxaluria, primary, type II {(NM_012203.2) "c.975A>G"}
1124 - 1131	GUCY2D:Leber congenital amaurosis 1 , Cone-rod dystrophy 6 {(NM_000180.3) "c.1992T>G", "c.2129C>T", "c.2513G>A", "c.2618C>G", "c.389delC", "c.529C>T", "c.620delC", "c.693delG"}
1132 - 1132	HACD1:Congenital myopathy {(NM_014241.4) "c.744C>A"}
1133 - 1133	HADHA:Long-Chain hydroxylacyl-CoA dehydrogenase deficiency (LCHAD) {(NM_000182.5) "c.1528G>C"}
1134 - 1134	HAX1:Severe congenital neutropenia type 3 (SCN3), a.k.a. Kostmann disease {(NM_006118) "c.125dupG"}
1135 - 1161	HBB:Hemoglobipathies (Including sickle-cell anemia and beta thalassemia, Hb C, D, E, O) {(NM_000518) "c.-138C>A", "c.-50-101C>T", "c.-78A>C", "c.-80T>A", "c.112delT", "c.114G>A", "c.118C>T", "c.135delC", "c.19G>A", "c.315+1G>A", "c.364G>A", "c.364G>C", "c.79G>A", "c.82G>T", "c.92+5G>C", "c.92+6T>C", "c.92G>C", "c.93-22_95del25" | (NM_000518.5) "1.78_Mb", "c.17_18delCT", "c.20A>T", "c.25_26delAA", "c.27dupG", "c.316-106C>G", "c.47G>A", "c.92+1G>A", "c.93-21G>A"}
1162 - 1184	HEXA:Tay-Sachs disease {(NM_000520) "c.1176G>A", "c.1528C>T" | (NM_000520.5) "c.1073+1G>A", "c.1274_1277dupTATC", "c.1351C>G", "c.1421+1G>C", "c.1444G>A", "c.316C>T", "c.459+2dupT", "c.496delC", "c.509G>A", "c.532C>T", "c.533G>A", "c.533G>T", "c.540C>G", "c.571-2A>G", "c.749G>A", "c.749G>T", "c.78G>A", "c.805+1G>A", "c.805G>A", "c.835T>C", "c.910_912delTTC"}

1185 - 1185	HEXB:Sandhoff disease, infantile, juvenile, and adult forms {(NM_000521) "c.1082+5G>A"}
1186 - 1186	HGD:Alkaptonuria {(NM_000187) "c.16-272_87+305del"}
1187 - 1187	HGSNAT:Retinitis pigmentosa 73 {(NM_152419.3) "c.370A>T"}
1188 - 1188	HIKESHI:Leukodystrophy, early onset spastic paraparesis,acquired microcephaly, optic atrophy and risk of early death {(NM_016401.4) "c.160G>C"}
1189 - 1191	HMGCL:HMG-CoA lyase deficiency {(NM_000191.3) "c.122G>A", "c.125A>G", "c.521G>A"}
1192 - 1192	HOGA1:Hyperoxaluria, primary, type III {(NM_138413) **"c.944_946delAGG"}
1193 - 1195	HPD:Thyrosinemia type III {(NM_002150.3) "c.325-1G>A", "c.415-1G>A", "c.481G>C"}
1196 - 1196	HPS1:Hermansky-Pudlak syndrome 1 {(NM_000195.5) "c.972delC"}
1197 - 1200	HPS3:Hermansky-Pudlak syndrome 3 {(NM_032383.5) "c.-2993_217+690del3900", "c.1163+1G>A", "c.1691+2T>G", "c.2482-2A>G"}
1201 - 1201	HPS6:Hermansky-Pudlak syndrome 6 {(NM_024747.5) "c.1065dupG"}
1202 - 1202	HSPD1:Leukodystrophy, hypomyelinating, 4, HLD4 (HSP60) {(NM_199440.1) "c.86A>G"}
1203 - 1203	IBA57:Spastic paraplegia 74, Autosomal Recessive {(NM_001010867.4) "c.678A>G"}
1204 - 1207	IDUA:Mucopolysaccharidosis Type IH - Hurler syndrome {(NM_000203.5) "c.1096A>C", "c.192C>A", "c.208C>T", "c.928C>T"}
1208 - 1209	IGHMBP2:Neuronopathy, distal hereditary motor, type VI {(NM_002180.2) "c.114delA", "c.707T>G"}
1210 - 1210	IL10RA:Inflammatory bowel disease 28, early onset, autosomal recessive {(NM_001558) "c.537G>A"}
1211 - 1214	INSR:Leprechaunism, Donohue syndrome {(NM_000208) "c.2683-542_2842+544del" | (NM_000208.4) "c.167T>C", "c.3079C>T", "c.857G>A"}
1215 - 1215	INVS:Nephronophthisis 2, infantile {(NM_014425.5) "c.2719C>T"}
1216 - 1216	ISPD:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), {(NM_001101426.4) "c.165dupG"}
1217 - 1220	ITGA2B:Glanzmann thrombasthenia, ITGA2B-related {(NM_000419) "c.2374delG" | (NM_000419.4) "c.1947-1G>A", "c.818G>A", "c.97A>G"}
1221 - 1223	ITGB3:Glanzmann thrombasthenia, ITGB3-related {(NM_000212) "c.1616_1617delTT" | (NM_000212.2) "11.2kbincl.ex.10-partex.13", "c.428T>G"}
1224 - 1224	ITGB4:Epidermolysis bullosa, junctional, with pyloric atresia - Carmi syndrome {(NM_000213.5) "c.3280_3793+176del2279 "}
1225 - 1225	ITK:Lymphoproliferative syndrome {(NM_005546) "c.1764C>G"}
1226 - 1229	IVD:Isovaleric academia {(NM_002225.4) "c.148C>T", "c.286+2T>C", "c.456+2T>C", "c.932C>T"}
1230 - 1230	JAK3:SCID, autosomal recessive, T-negative/B-positive type {(NM_000215) "c.2680+89G>A"}

1231 - 1231	KCNJ10:SESAME syndrome {(NM_002241.5) "c.524G>A"}
1232 - 1232	KIAA1279:Goldberg-Shprintzen megacolon syndrome {(NM_015634) "c.1516dupA"}
1233 - 1233	KIF1C:Spastic ataxia 2, Autosomal Recessive {(NM_006612) "c.2191C>T"}
1234 - 1234	KIZ:Retinitis pigmentosa 69 {(NM_018474) "c.226C>T"}
1235 - 1235	KLHL40:Nemaline myopathy 8, Autosomal Recessive {(NM_152393.4) "c.581T>A"}
1236 - 1236	KREMEN1:Ectodermal dysplasia {(NM_032045) "c.626T>C"}
1237 - 1238	KRT14:Epidermolysis bullosa simplex {(NM_000526) "c.400C>T", "c.915G>A"}
1239 - 1240	KY:Myopathy, myofibrillar, 7 {(NM_178554) "c.405C>A", "c.51_52insTATCGACATGTGCTGTATCTATCGACAT"}
1241 - 1246	LAMA2:Muscular dystrophy, congenital, due to partial LAMA2 deficiency {(NM_000426) "c.4609_4631del" | (NM_000426.3) "c.3718C>T", "c.5260delG", "c.828C>G", "c.8665G>A", "c.8689C>T"}
1247 - 1250	LAMA3:Laryngoonychocutaneous Syndrome {(NM_000227.4) "c.1981C>T", "c.2975delA", "c.4815G>T", "c.893_894insT"}
1251 - 1261	LAMB3:Epidermolysis bullosa, junctional, non-Herlitz type {(NM_000228) "c.129insA" | (NM_000228.3) "c.124C>T", "c.1295dupA", "c.1903C>T", "c.1978C>T", "c.2528delA", "c.2914C>T", "c.3024delT", "c.3247C>T", "c.430C>T", "c.727C>T"}
1262 - 1263	LAMC2:Epidermolysis bullosa, junctional, Herlitz type {(NM_018891.2) "c.1756C>T", "c.368_373delinsACCAC"}
1264 - 1268	LCA5:Leber congenital amaurosis 5 {(NM_181714.3) "c.1062_1068delCGAAAAC", "c.1714C>T", "c.238C>T", "c.835C>T", "c.94delT"}
1269 - 1270	LIFR:Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome-LIFR related {(NM_002310.5) "c.1601-1G>A", "c.2472_2476delTATGT"}
1271 - 1272	LIPA:Wolman disease {(NM_001127605.2) "c.260G>T", "c.398delC"}
1273 - 1273	LONP1:CODAS syndrome {(NM_004793) "c.2009C>T"}
1274 - 1275	LOXHD1:Deafness, Autosomal Recessive 77 {(NM_144612) "c.5894dupG" | (NM_144612.6) "c.4714C>T"}
1276 - 1277	LRBA:Immunodeficiency, common variable, 8, with autoimmunity {(NM_001199282) "c.8139_8142dupCATG" | (NM_001199282.2) "c.7937T>G"}
1278 - 1279	MAK:Retinitis pigmentosa 62 {(NM_001242957.2) "c.497G>A" | (NM_005906) "c.394_395insCTTC"}
1280 - 1280	MAN1B1:Mental retardation, Autosomal Recessive 15 {(NM_016219.5) "c.1863G>A"}
1281 - 1281	MATN3:Spondyloepimetaphyseal dysplasia {(NM_002381.5) "c.910T>A"}
1282 - 1282	MCIDAS:Mucociliary clearance disorder {(NM_001190787.2) "c.1142G>A"}
1283 - 1287	MCOLN1:Mucolipidosis type IV - ML4 {(NM_020533) "c.1135-1G>C" | (NM_020533.3) "c.-1015_788del6433", "c.1207C>T", "c.406-2A>G", "c.964C>T"}

1288 - 1289	MECR:Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities {(NM_016011) "c.695G>A", "c.830+2dupT"}
1290 - 1290	MED17:Microcephaly, postnatal progressive, with seizures and brain atrophy ((ICCA) {(NM_004268.5) "c.1112T>C"}
1291 - 1291	MED25:Basel-Vanagaite-Smirin-Yosef syndrome {(NM_030973.3) "c.116A>G"}
1292 - 1292	MEGF10:Myopathy, areflexia, respiratory distress, and dysphagia, early-onset {(NM_001256545.2) "c.1325delC"}
1293 - 1293	MERTK:Retinitis pigmentosa 38 {(NM_006343) "c.2164C>T"}
1294 - 1295	MFSD8:Ceroid lipofuscinosis, neuronal, 7 {(NM_152778) "c.103C>T" | (NM_152778.2) "c.472G>A"}
1296 - 1297	MKS1:Meckel syndrome 1 {(NM_017777.3) "c.1048C>T", "c.472C>T"}
1298 - 1300	MLC1:Megalencephalic leukoencephalopathy with subcortical cysts {(NM_015166.3) "c.176G>A", "c.274C>T", "c.278C>T"}
1301 - 1301	MLPH:Griscelli syndrome, type 3 {(NM_024101.7) "c.103C>T"}
1302 - 1302	MMACHC:Methylmalonic aciduria and homocystinuria, cblC type {(NM_015506.3) "c.271dupA"}
1303 - 1305	MOCS1:Molybdenum cofactor deficiency A {(NM_001075098.3) "c.1510C>T", "c.722delT", "c.971G>A"}
1306 - 1307	MOCS2:Molybdenum cofactor deficiency Type B {(NM_004531.5) "c.226G>A", "c.377+1G>A"}
1308 - 1311	MPDU1:Congenital disorder of glycosylation, type If {(NM_004870) "c.511delC" | (NM_004870.4) "c.218G>A", "c.2T>C", "c.356T>C"}
1312 - 1317	MPL:Thrombocytopenia, congenital amegakaryocytic {(NM_005373) "c.212+5G>A", "c.76C>T" | (NM_005373.2) "c.1031T>A", "c.127C>T", "c.460T>C", "c.79+2T>A"}
1318 - 1318	MPV17:Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) {(NM_002437.5) "c.278A>C"}
1319 - 1319	MRE11A:Ataxia Telangiectasia like disorder {(NM_005591.3) "c.290A>G"}
1320 - 1322	MTHFR:Homocystinuria due to MTHFR deficiency {(NM_005957) "c.1072C>T" | (NM_005957.4) "c.16delA", "c.474A>T"}
1323 - 1326	MTTP:Abetalipoproteinemia ABL {(NM_000253.3) "c.2212delT", "c.2593G>T", "c.307A>T", "c.62-2A>G"}
1327 - 1328	MUT:Methylmalonic acidemia, mut(0) type {(NM_000255) "c.1240G>T" | (NM_000255.4) "c.655A>T"}
1329 - 1329	MVK:Hyper-IgD syndrome {(NM_000431.4) "c.1129G>A"}
1330 - 1332	MYBPC1:Lethal congenital contracture syndrome 4 {(NM_002465.4) "c.556G>A", "c.688G>A", "c.952C>T"}
1333 - 1334	MYH2:Proximal myopathy and ophthalmoplegia {(NM_017534.6) "c.2400delG", "c.706G>A"}
1335 - 1341	MYO15A:Deafness, Autosomal Recessive 3 {(NM_016239) "c.1223C>T", "c.9861C>T" | (NM_016239.4) "c.373_374delCG", "c.4240G>A", "c.7207G>T", "c.8183G>A", "c.8467G>A"}
1342 - 1361	MYO7A:Usher syndrome, type 1B {(NM_000260) "c.3262C>T" | (NM_000260.4) "c.1190C>A", "c.1996C>T", "c.2065delC", "c.2187+1G>A", "c.2307delC", "c.2476G>A", "c.2777T>A", "c.29T>C", "c.3892G>A", "c.4153-2A>G", "c.470+1G>A", "c.5068dupC",

	"c.5581C>T", "c.5660C>T", "c.6196delC", "c.620A>G", "c.640G>A", "c.6487G>A", "c.700C>T"}
1362 - 1362	NAGLU:Mucopolysaccharidosis type IIIB (Sanfilippo B) {(NM_000263.4) "c.2021G>A"}
1363 - 1363	NARS2:Combined oxidative phosphorylation deficiency 24 (COXPD24) {(NM_024678) "c.500A>G"}
1364 - 1365	NBEAL2:Gray platelet syndrome {(NM_015175.2) "c.2701C>T", "c.5413dupG"}
1366 - 1368	NCF1:Chronic granulomatous disease due to deficiency of NCF-1 {(NM_000265) "c.75_76delGT" | (NM_000265.6) "c.153+1G>A", *"c.579G>A"}
1369 - 1372	NCF2:Chronic granulomatous disease due to deficiency of NCF-2 {(NM_000433) "exon9+10insertionafterexon10" | (NM_000433.3) "c.1171_1175delAAGCT", "c.196C>T", "c.304C>T"}
1373 - 1373	NDUFA11:Mitochondrial complex I deficiency - NDUFA11 gene {(NM_001193375.1) "c.97+5G>A"}
1374 - 1374	NDUFAF5:Mitochondrial complex I deficiency - NDUFAF5 gene {(NM_024120.5) "c.749G>T"}
1375 - 1375	NDUFS2:Mitochondrial complex I deficiency-NDUFS2 gene {(NM_004550.4) "c.1237T>C"}
1376 - 1376	NDUFS4:Leigh syndrome {(NM_002495.4) "c.462delA"}
1377 - 1377	NDUFS6:Mitochondrial complex I deficiency - NDUFS6 gene {(NM_004553.4) "c.344G>A"}
1378 - 1381	NEB:Nemaline myopathy 2 {(NM_001271208.2) "c.17118+1G>A", "c.18808C>T", "c.9619-2A>G" | (NM_004543.4) "c.7431+1917_7536+372del"}
1382 - 1382	NECTIN1:Cleft lip/palate ectodermal dysplasia, CLPED1 (Zlotogora-Ogur syndrome) {(NM_203285) "c.556delG"}
1383 - 1383	NGLY1:Congenital disorder of deglycosylation {(NM_018297.4) "c.1294G>T"}
1384 - 1384	NNT:Glucocorticoid deficiency 4 {(NM_182977.3) "c.598G>A"}
1385 - 1403	NPC1:Niemann-Pick disease type C1 {(NM_000271) "c.3742_3753del" | (NM_000271.5) "c.1211G>A", "c.1241_1242delTC", "c.1437_1442delCACCAT", "c.1552C>T", "c.1761delT", "c.2279_2281delTCT", "c.2780C>T", "c.2972_2973delAG", "c.2974G>A", "c.2974G>C", "c.2974G>T", "c.3007C>T", "c.3347_3348delTC", "c.3467A>G", "c.3557G>A", "c.3614C>A", "c.3637T>G", "c.3673T>G"}
1404 - 1404	NPHP1:Joubert syndrome {"del exons 2-7"}
1405 - 1413	NPHS1:Nephrotic syndrome type 1 {(NM_004646.3) "c.1138C>T", "c.121_122delCT", "c.1707C>G", "c.2104G>A", "c.2160dupC", "c.3325C>T", "c.3478C>T", "c.514_516delACC", "c.532C>T"}
1414 - 1415	NPHS2:Nephrotic syndrome {(NM_014625) "c.388G>A" | (NM_014625.3) "c.412C>T"}
1416 - 1417	NRL:Retinitis pigmentosa 27 {(NM_006177) "c.444_445insGCTGCGGG", "c.91C>T"}
1418 - 1421	NTRK1:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_002529.3) "c.1250C>T", "c.1860_1861insT", "c.207_208delTG",

	"c.2084C>T"}
1422 - 1422	NUP62:Striatonigral degeneration, Infantile Bilateral Striatal Necrosis (IBSN) {(NM_016553.4) "c.1172A>C"}
1423 - 1423	OAT:Gyrate atrophy of choroid and retina with or without ornithinemia {(NM_000274) "c.159delC"}
1424 - 1426	OCA2:Albinism, oculocutaneous, type II {(NM_000275) "c.79G>A" | (NM_000275.3) "c.1320G>C", "c.1327G>A"}
1427 - 1427	OPA3:3-methylglutaconic aciduria, type III - Costeff {(NM_025136.3) "c.143-1G>C"}
1428 - 1430	OTC:Ornithine transcarbamylase deficiency {(NM_000531.6) "c.717+1G>T", "c.829C>T", "c.958C>T"}
1431 - 1432	OTOA:Deafness, Autosomal Recessive 22 {(NM_144672) "c.1025A>T", "c.2359G>T"}
1433 - 1434	OTOF:Deafness, Autosomal Recessive 9 {(NM_194248) "c.5332G>T" | (NM_194248.2) "c.2866+1G>A"}
1435 - 1435	P3H2:Myopia, high, with cataract and vitreoretinal degeneration {(NM_018192) "c.1523G>T"}
1436 - 1475	PAH:Phenylketonuria {(NM_000277) "c.1089delG", "c.1139C>T", "c.116T>C", "c.1184C>G", "c.1243G>A", "c.168+1G>A", "c.169-4951del6604ins8 6.7kb_del", "c.169_171delGAG", "c.283A>T", "c.311C>A", "c.350delC", "c.442-5C>G", "c.48dupT", "c.506G>A", "c.526C>T", "c.592_613del22", "c.632delC", "c.838G>A", "c.842+5G>A", "c.967_969delACA", "c.969+1G>A" | (NM_000277.3) "c.1045T>C", "c.1066-11G>A", "c.1208C>T", "c.1222C>T", "c.1315+1G>A", "c.143T>C", "c.165T>G", "c.165delT", "c.441+5G>T", "c.473G>A", "c.689T>C", "c.722G>A", "c.727C>T", "c.754C>T", "c.782G>A", "c.782G>C", "c.842C>T", "c.889C>T", "c.898G>T"}
1476 - 1476	PARK2:Parkinson disease, early onset {(NM_004562) "c.101delA"}
1477 - 1477	PAX7:Myopathy, congenital, progressive, with scoliosis {(NM_001135254) "c.1403-2A>G"}
1478 - 1478	PCCA:Propionic acidemia, PCCA-related {(NM_000282.4) "c.923dupT"}
1479 - 1479	PCCB:Propionic acidemia, PCCB-related {(NM_000532.5) "c.1173dupT"}
1480 - 1481	PCDH12:Microcephaly, seizures, spasticity, and brain calcification (MISSBC) {(NM_016580) "c.2515C>T", "c.995delT"}
1482 - 1482	PCDH15:Usher syndrome, type 1F {(NM_033056.3) "c.733C>T"}
1483 - 1483	PCK1:Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency {(NM_002591.4) "c.134T>C"}
1484 - 1485	PCNT:Microcephalic osteodysplastic primordial dwarfism, type II {(NM_006031) "c.2984_2994delCAGACTTTGAG" | (NM_006031.5) "c.3465-1G>A"}
1486 - 1490	PDE6A:Retinitis pigmentosa 43 {(NM_000440) "c.1957C>T", "c.1960C>T", "c.2081_2085delAACAG", "c.409delGinsCT", "c.769C>T"}
1491 - 1491	PDE6B:Retinitis pigmentosa-40 {(NM_001145291) "c.1417delC"}
1492 - 1492	PDE6G:Retinitis pigmentosa 57 {(NM_002602.4) "c.187+1G>T"}
1493 - 1495	PEPD:Prolidase deficiency {(NM_000285.4) "c.1103T>G", "c.605C>T", "c.634G>C"}

1496 - 1498	PEX1:Peroxisome biogenesis disorder 1A (Zellweger) {(NM_000466) "c.2916delA" | (NM_000466.3) "c.2097dupT", "c.2528G>A"}
1499 - 1501	PEX2:Peroxisome biogenesis disorder 5A (Zellweger) {(NM_001079867.1) "c.355C>T", "c.550delT", "c.669G>A"}
1502 - 1506	PEX6:Peroxisome biogenesis disorder 4B (Zellweger syndrome) {(NM_000287.4) "c.1715C>T", "c.1944delC", "c.1947delG", "c.2094+2T>C", "c.2534T>C"}
1507 - 1507	PEX7:Rhizomelic chondrodysplasia punctata type 1 {(NM_000288.4) "c.283T>G"}
1508 - 1508	PGAP3:Hyperphosphatasia with mental retardation syndrome 4 {(NM_033419.5) "c.845A>G"}
1509 - 1509	PGM1:Congenital disorder of glycosylation, type It {(NM_002633) "c.112A>T"}
1510 - 1510	PHGDH:Phosphoglycerate dehydrogenase deficiency {(NM_006623.3) "c.1468G>A"}
1511 - 1511	PHKG2:Glycogen storage disease IXc {(NM_000294.3) "c.71A>G"}
1512 - 1512	PHYH:Refsum disease {(NM_001037537.1) "c.523C>T"}
1513 - 1514	PIGN:Multiple congenital anomalies-hypotonia-seizures syndrome 1 {(NM_012327.5) "c.2126G>A", "c.755A>T"}
1515 - 1516	PIGT:Multiple congenital anomalies-hypotonia-seizures syndrome 3 {(NM_015937.6) "c.1564T>G", "c.761delG"}
1517 - 1517	PIP5K1C:Lethal congenital contractural syndrome 3 {(NM_012398.2) "c.757G>A"}
1518 - 1518	PJVK:Deafness, Autosomal Recessive 59 {(NM_001042702.4) "c.406C>T"}
1519 - 1529	PKHD1:Polycystic kidney & hepatic disease, PKHD1-related {(NM_138694) "c.10444C>T", "c.1486C>T", "c.5895dupA", "c.8870T>C", "c.9689delA" | (NM_138694.4) "c.107C>T", "c.1350delC", "c.2279G>A", "c.3761_3762delCCinsG", "c.6122-12G>A", "c.824C>T"}
1530 - 1534	PLA2G6:Infantile neuroaxonal dystrophy 1 (INAD) {(NM_003560) "c.1594A>T" | (NM_003560.4) "c.1040G>C", "c.2070_2072delTGT", "c.2251G>A", "c.668C>A"}
1535 - 1535	PLAA:Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies {(NM_001031689.3) "c.2254C>T"}
1536 - 1536	PLEKHG2:Leukodystrophy and acquired microcephaly with or without dystonia {(NM_022835.3) "c.610C>T"}
1537 - 1540	PMM2:Congenital disorder of glycosylation Ia {(NM_000303) "c.338C>T", "c.357C>A", "c.422G>A", "c.691G>A"}
1541 - 1541	POC1A:Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis {(NM_015426.5) "c.512T>C"}
1542 - 1542	POMGNT2:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8) {(NM_032806.6) "c.1232_1233delAG"}
1543 - 1544	POMT1:Walker-Warburg Syndrome, type A, 1 {(NM_007171) "c.2167dupG", "c.428-1G>C"}
1545 - 1545	POMT2:Walker-Warburg Syndrome, type A, 2 {(NM_013382) "c.924-2A>C"}

1546 - 1546	POR:Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis {(NM_000941.3) "c.1615G>A"}
1547 - 1547	PPIB:Osteogenesis imperfecta, type IX {(NM_000942.4) "c.563_566delACAG"}
1548 - 1548	PPP1R13L:Cardio-Cutaneous Syndrome DCM {(NM_006663.4) "c.2241C>G"}
1549 - 1549	PPT1:Ceroid lipofuscinosis, neuronal, 1 {(NM_000310.3) "c.169dupA"}
1550 - 1550	PRCD:Retinitis pigmentosa 36 {(NM_001077620) "c.64C>T"}
1551 - 1551	PRICKLE1:Epilepsy, progressive myoclonic 1B {(NM_153026.3) "c.311G>A"}
1552 - 1552	PSMB8:Autoinflammation, lipodystrophy, and dermatosis syndrome {(NM_148919.4) "c.405C>A"}
1553 - 1553	PTPN23:Developmental delay, cognitive impairment, and atopic atrophy {(NM_015466) "c.3886_3888del"}
1554 - 1554	PUS1:Mitochondrial myopathy and sideroblastic anemia 1 {(NM_001002020.3) "c.346C>T"}
1555 - 1555	RAB27A:Griscelli syndrome, type 2 {(NM_004580) "c.148_149delinsC"}
1556 - 1556	RAB28:Cone-rod dystrophy 18 {(NM_001017979) "c.409C>T"}
1557 - 1558	RAG1:Severe combined immudeficiency, B cell-negative, RAG1-related {(NM_000448.2) "c.1361T>A", "c.1410_1413delCTTG"}
1559 - 1563	RAG2:Severe combined immudeficiency, B cell-negative, RAG2-related {(NM_000536.3) "c.1438G>T", "c.193G>T", "c.379A>T", "c.470G>T", "c.685C>T"}
1564 - 1567	RAPSN:Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency {(NM_005055.5) "c.-210A>G", "c.-27C>G", "c.264C>A", "c.672_673insACT"}
1568 - 1568	RAPSN:Severe combined immudeficiency, B cell-negative, RAG2-related {(NM_005055) "c.648T>A"}
1569 - 1569	RARS2:Pontocerebellar hypoplasia, type 6 {(NM_020320.5) "c.110+5A>G"}
1570 - 1577	RDH12:Leber congenital amaurosis 13 {(NM_152443.3) "c.146C>T", "c.164C>T", "c.295C>A", "c.377C>T", "c.481C>T", "c.658+1G>A", "c.716G>A", "c.740T>C"}
1578 - 1579	RECQL2:Werner syndrome {(NM_000553.5) "c.1105C>T", "c.2665C>T"}
1580 - 1580	RFX5:Bare lymphocyte syndrome, type II (SCID) {(NM_000449) "c.715C>T"}
1581 - 1581	RIN2:Macrocephaly, alopecia, cutis laxa, and scoliosis {(NM_018993.3) "c.1731delC"}
1582 - 1582	RNASEH2B:Aicardi-Goutieres syndrome 2 {(NM_024570.3) "c.529G>A"}
1583 - 1583	ROGDI:Kohlschutter-Tonz syndrome {(NM_024589.2) "c.469C>T"}
1584 - 1585	RP1:Retinitis pigmentosa 1 {(NM_006269) "c.688G>T" | (NM_006269.2) "c.4941dupT"}
1586 - 1591	RPE65:Leber congenital amaurosis 2 {(NM_000329.3) "c.1301C>G", "c.227A>C", "c.361dupT", "c.722A>T", "c.886dupA", "c.95-2A>T"}

1592 - 1595	RPGRIP1:Cone-rod dystrophy 13 {(NM_020366) "c.1615_1624delGAACTGGAGG", "c.2935C>T", "c.2974delA", "c.3663_3666delAGAA"}
1596 - 1596	RPGRIP1L:Meckel syndrome 5 {(NM_015272.5) "c.118C>T"}
1597 - 1597	RRM2B:Mitochondrial DNA depletion syndrome 8 {(NM_015713.5) "c.215C>G"}
1598 - 1598	RSPH9:Ciliary dyskinesia, primary, 12 {(NM_152732.5) "c.804_806delGAA"}
1599 - 1603	RTEL1:Dyskeratosis congenita {(NM_001283009.1) "c.1476G>T", "c.2848C>T", "c.2869C>T", "c.2920C>T", "c.3791G>A"}
1604 - 1606	RYR1:Minicore myopathy with external ophthalmoplegia {(NM_000540) "c.1366G>A", "c.9047A>G" | (NM_000540.2) "c.9623C>T"}
1607 - 1608	SAMD9:Tumoral calcinosis, familial, normophosphatemic {(NM_017654.4) "c.1030C>T", "c.4483A>G"}
1609 - 1613	SAMHD1:Aicardi Goutieres syndrome {(NM_015474) "c.359_370delATCCTATCCATG" | (NM_015474.3) "9.1-KB_DEL", "c.1106T>C", "c.649_650insG", "c.676C>G"}
1614 - 1614	SARS2:Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis {(NM_017827.3) "c.1169A>G"}
1615 - 1615	SCAPER:Retinitis pigmentosa with intellectual disability {(NM_020843) "c.2806delC"}
1616 - 1617	SCN9A:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_002977.3) "c.1124delG", "c.2687G>A"}
1618 - 1618	SCNN1A:Pseudohypoaldosteronism type I - SCNN1A gene {(NM_001038) "c.1522C>T"}
1619 - 1620	SCNN1B:Pseudohypoaldosteronism type I - SCNN1B gene {(NM_000336) "c.648dupA", "c.915delC"}
1621 - 1622	SDHA:Cardiomyopathy, dilated , 1GG neonatal isolated {(NM_004168) "c.1A>G" | (NM_004168.4) "c.1664G>A"}
1623 - 1624	SEC23B:Dyserythropoietic anemia, congenital, type II {(NM_006363.6) "c.2129C>T", "c.325G>A"}
1625 - 1629	SERAC1:3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome {(NM_032861) "c.1102C>T", "c.1339C>T" | (NM_032861.4) "c.1018delT", "c.128+4A>G", "c.698_699delinsAGTATA"}
1630 - 1630	SGCG:Muscular dystrophy, limb-girdle, type 2C {(NM_000231.2) "c.525delT"}
1631 - 1639	SGSH:Mucopolysaccharidisis type IIIA (Sanfilippo A) {(NM_000199) "c.1231C>T", "c.267C>A", "c.697C>T" | (NM_000199.5) "c.1093C>T", "c.1298G>A", "c.332T>C", "c.416C>T", "c.544C>T", "c.812C>T"}
1640 - 1640	SLC12A3:Bartter Syndrome, Gitelman Variant {(NM_000339.3) "c.1313G>A"}
1641 - 1641	SLC17A5:Sialic acid storage disorder, infantile (ISSD) {(NM_012434.5) "c.983G>A"}
1642 - 1642	SLC18A3:Myasthenia gravis, congenital {(NM_003055) "c.1078G>C"}
1643 - 1644	SLC19A2:Thiamine-responsive megaloblastic anemia syndrome {(NM_006996.3) "c.1223+1G>A", "c.725delC"}

1645 - 1647	SLC1A4:Spastic tetraplegia, thin corpus callosum, and progressive microcephaly {(NM_003038.5) "c.1369C>T", "c.766G>A", "c.944_945del"}
1648 - 1648	SLC22A5:Carnitine deficiency, systemic primary {(NM_003060.3) "c.1196G>A"}
1649 - 1650	SLC25A15:Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome {(NM_014252) "c.562_564delTTC" | (NM_014252.3) "c.706A>G"}
1651 - 1652	SLC25A1:Combined D-2- and L-2-hydroxyglutaric aciduria {(NM_005984.5) "c.389G>A", "c.845G>A"}
1653 - 1654	SLC25A20:Carnitine-acylcarnitine translocase deficiency - CACT {(NM_000387.6) "c.609-3C>G", "c.713A>G"}
1655 - 1655	SLC26A3:Congenital chloride diarhhea (CLD) {(NM_000111.2) "c.559G>T"}
1656 - 1666	SLC26A4:Pendred syndrome {(NM_000441.2) "c.1001G>T", "c.1151A>G", "c.1198delT", "c.1246A>C", "c.1341+1delG", "c.1458dupT", "c.2000T>G", "c.2168A>G", "c.349C>T", "c.707T>C", "c.716T>A"}
1667 - 1670	SLC29A3:Histiocytosis-lymphadenopathy plus syndrome {(NM_018344.5) "c.1157G>A" | (NM_018344.6) "c.1045delC", "c.1279G>A", "c.1309G>A"}
1671 - 1673	SLC2A2:Fanconi-Bickel syndrome {(NM_000340.2) "c.372A>C", "c.734A>C", "c.901C>T"}
1674 - 1674	SLC30A9:Birk-Landau-Perez cerebro-renal syndrome {(NM_016474.5) "c.1047_1049delCAG"}
1675 - 1676	SLC35A3:Arthrogryposis, mental retardation, and seizures {(NM_012243.3) "c.514C>T", "c.886A>G"}
1677 - 1677	SLC35C1:Congenital disorder of glycosylation, type IIc {(NM_018389.4) "c.923C>G"}
1678 - 1681	SLC37A4:Glycogen storage disease Ib {(NM_001164277.1) "c.1042_1043delCT", "c.1179G>A", "c.446G>A", "c.83G>A"}
1682 - 1682	SLC39A4:Acrodermatitis enteropathica {(NM_130849.3) "c.1224delC"}
1683 - 1683	SLC45A2:Albinism, oculocutaneous, type IV {(NM_001012509) "c.1076_1077delAG"}
1684 - 1684	SLC46A1:Folate malabsorption, hereditary {(NM_080669) "c.337C>T"}
1685 - 1685	SLC4A4:Renal tubular acidosis (RTA), proximal, with ocular abnormalities and mental retardation {(NM_003759.3) "c.2321G>A"}
1686 - 1686	SLCO2A1:Hypertrophic osteoarthropathy, primary, Autosomal Recessive 2 {(NM_005630.2) "c.1292delC"}
1687 - 1688	SMARCAL1:Schimke immunoosseous dysplasia {(NM_014140.3) "c.2542G>T", "c.863-2A>G"}
1689 - 1689	SMN1:Spinal muscular atrophy-1 {(NM_000344) "c.835_*3del"}
1690 - 1702	SMPD1:Niemann-Pick disease type B, SMPD1-related {(NM_000543) "c.1092-1G>C", "c.880C>A" | (NM_000543.5) "c.1493G>T", "c.1705T>C", "c.1758T>G", "c.1805G>C", "c.1828_1830delCGC", "c.442T>A", "c.573delT", "c.730G>A", "c.911T>C", "c.96G>A",

	"c.996delC"}
1703 - 1703	SNAP29:Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome - CEDNIK Syndrome {(NM_004782) "c.223delG"}
1704 - 1704	SNX10:Osteopetrosis, Autosomal Recessive 8 {(NM_001199835.1) "c.152G>A"}
1705 - 1708	SPG11:Spastic paraplegia 11, Autosomal Recessive {(NM_025137) "c.5986dupT" | (NM_025137.4) "c.118C>T", "c.2471dupT", "c.4339C>T"}
1709 - 1713	SPINK5:Netherton syndrome {(NM_001127698.1) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC", "c.995delT"}
1714 - 1714	ST3GAL3:Early infantile epileptic encephalopathy 15 {(NM_006279.4) "c.958G>C"}
1715 - 1715	STRA6:Microphthalmia {(NM_001142617.1) "c.1678G>C"}
1716 - 1717	STRC:Deafness, Autosomal Recessive 16 {(NM_153700.2) "EX7_EX29DEL", "c.4171C>G"}
1718 - 1718	SUCLA2:Mitochondrial DNA depletion syndrome 5 {(NM_003850) "c.788_802+29del"}
1719 - 1720	SUMF1:Multiple sulfatase deficiency {(NM_182760.3) "c.1043C>T", "c.463T>C"}
1721 - 1722	SURF1:Leigh syndrome, due to COX deficiency {(NM_003172) "c.312_321delTCTGCCAGCCinsAT", "c.575_576insTGCG"}
1723 - 1723	SYNE4:Deafness, Autosomal Recessive 76 {(NM_001039876.3) "c.228_229delAT"}
1724 - 1724	SZT2:Epileptic encephalopathy, early infantile, 18 {(NM_015284.3) "c.73C>T"}
1725 - 1726	SepSecS:Pontocerebellar hypoplasia type 2D {(NM_016955.4) "c.1001A>G", "c.715G>A"}
1727 - 1727	TAF2:Mental retardation, Autosomal Recessive 40 {(NM_003184.4) "c.557C>G"}
1728 - 1728	TBCD:Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM_005993.4) "c.1423G>A"}
1729 - 1730	TBCE:Hypoparathyroidism retardation dysmorphism syndrome {(NM_003193.5) "c.155_166delGCCACGAAGGGA", "c.355_356del"}
1731 - 1731	TBX19:Adrenocorticotropic hormone deficiency {(NM_005149.3) "c.574_577delATAG"}
1732 - 1735	TCIRG1:Osteopetrosis, Autosomal Recessive 1 {(NM_006019.4) "c.117+4A>T", "c.1331G>T", "c.1384_1386delAAC", "c.674delG"}
1736 - 1736	TCTN2:Meckel syndrome 8 {(NM_024809.5) "c.1506-2A>G"}
1737 - 1739	TECPR2:Spastic paraplegia 49, Autosomal Recessive {(NM_001172631.2) "c.1319delT", "c.3416delT", "c.566C>T"}
1740 - 1740	TGM1:Ichthyosis, congenital, Autosomal Recessive 1 {(NM_000359) "c.2290C>T"}
1741 - 1741	THG1L:Cerebellar ataxia and developmental delay {(NM_017872.5) "c.164T>C"}
1742 - 1742	TIMM50:3-methylglutaconic aciduria, type IX {(ENST00000314349.4) "c.649C>T"}
1743 - 1745	TK2:Mitochondrial DNA depletion syndrome 2 (myopathic type)

	{(NM_004614.5) "c.360_361delGCinsAA", "c.361C>A", "c.635T>A"}
1746 - 1746	TKT:Short stature, developmental delay, and congenital heart defects {(NM_001135055.2) "c.769_770insCTACCTCCTTATCTTCTG"}
1747 - 1751	TMC1:Deafness, Autosomal Recessive 7 {(NM_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"}
1752 - 1752	TMEM165:Congenital disorder of glycosylation {(NM_018475.4) "c.792+182G>A"}
1753 - 1755	TMEM216:Joubert syndrome 2 (MKS2) {(NM_001173990.3) "c.218G>A", "c.218G>T", "c.230G>C"}
1756 - 1756	TMEM231:Meckel syndrome 11 {(NM_001077418.3) "c.664+4A>G"}
1757 - 1757	TMEM260:Neurodevelopmental, Cardiac, and Renal Syndrome {(NM_017799.3) "c.1393C>T"}
1758 - 1759	TMEM38B:Osteogenesis imperfecta, type XIV {(NM_018112) "c.455_542del", "c.507G>A"}
1760 - 1762	TMEM67:Joubert syndrome type 6 (MSK3) {(NM_153704) "c.1065+1delG" | (NM_153704.5) "c.1975C>T", "c.725A>G"}
1763 - 1765	TMEM70:ATPase deficiency, nuclear encoded {(NM_017866) "c.336T>A" | (NM_017866.6) "c.238C>T", "c.316+1G>T"}
1766 - 1767	TMPRSS3:Deafness, Autosomal Recessive 8/10 {(NM_024022) "c.1177_1184delins" | (NM_024022.2) "c.989delA"}
1768 - 1768	TNNT1:Nemaline myopathy 5, Amish type {(NM_003283) "c.574_577delinsTAGTGCTGT"}
1769 - 1769	TPP1:Ceroid lipofuscinosis, neuronal, 2 {(NM_000391) "c.775delC"}
1770 - 1770	TRAK1:Encephalopathy, fatal {(NM_001042646.2) "c.287-2A>C"}
1771 - 1771	TRAPPC9:Mental retardation, Autosomal Recessive 13 {(NM_031466.7) "c.1423C>T"}
1772 - 1772	TRIM32:Bardet-Biedl syndrome 11 {(NM_012210) "c.388C>T"}
1773 - 1773	TRIOBP:Deafness, Autosomal Recessive 28 {(NM_001039141) "c.1741C>T"}
1774 - 1775	TRMT10A:Microcephaly, short stature, and impaired glucose metabolism {(NM_152292.4) "c.616G>A", "c.727C>T"}
1776 - 1778	TRMU:LIFT, Liver failure infantile transient {(NM_018006) "c.500_509del10", "c.835G>A" | (NM_018006.5) "c.229T>C"}
1779 - 1782	TRPM1:Night blindness, congenital stationary (complete), 1C, Autosomal Recessive {(NM_002420.5) "36.4-KB_DEL,_EX2-7", "c.2567G>A", "c.2629C>T", "c.880A>T"}
1783 - 1784	TRPM6:Hypomagnesemia 1, intestinal {(NM_017662.5) "c.1010+5G>C", "c.2009+1G>A"}
1785 - 1786	TSHR:Hypothyroidism, congenital, nongoitrous, 1 {(NM_000369) "c.202C>T" | (NM_000369.2) "c.1825C>T"}
1787 - 1787	TSPAN12:Vitroretinal vascular malformations, congenital {(NM_012338) "c.542G>T"}
1788 - 1788	TSPEAR:Ectodermal dysplasia {(NM_144991.3) "c.1877T>C"}
1789 - 1790	TTN:Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis {(NM_003319.4) "c.58881dupA" | (NM_133432) "c.36122delC"}
1791 - 1791	TUBGCP4:Microcephaly, primary, Autosomal Recessive {(NM_014444.5) "c.579dupT"}

1792 - 1794	TULP1:Retinitis pigmentosa 14 {(NM_003322) "c.849_852dup" | (NM_003322.6) "c.1349G>A", "c.1495+2dupT"}
1795 - 1796	TYMP:Mitochondrial DNA depletion syndrome 1 (MNGIE type) {(NM_001113755.2) "c.433G>A", "c.866A>C"}
1797 - 1813	TYR:Albinism, oculocutaneous, type IA (OCA1A) {(NM_000372) "c.1A>G" | (NM_000372.5) "c.1037-1G>A", "c.1037-7T>A", "c.1118C>A", "c.1204C>T", "c.1217C>T", "c.1357C>T", "c.140G>A", "c.149C>G", "c.454C>T", "c.649C>T", "c.649delC", "c.74dupT", "c.757G>A", "c.832C>T", "c.880G>A", "c.896G>A"}
1814 - 1814	UNC13D:Hemophagocytic lymphohistiocytosis, familial, 3 {(NM_199242.2) "c.753+1G>T"}
1815 - 1816	UNC80:Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM_032504) "c.7183C>T" | (NM_032504.1) "c.151C>T"}
1817 - 1817	UPB1:Beta-ureidopropionase deficiency {(NM_016327.3) "c.917-1G>A"}
1818 - 1818	UQCRQ:Mitochondrial complex III deficiency, nuclear type 4 {(NM_014402.5) "c.134C>T"}
1819 - 1821	USH1C:Usher syndrome, type 1C {(NM_005709.3) "c.1220delG", "c.238dupC", "c.497-2delA"}
1822 - 1822	USH1G:Usher syndrome, type 1G {(NM_173477) "c.205dup"}
1823 - 1848	USH2A:Usher syndrome, type 2A {(NM_206933.3) "c.1000C>T", "c.10211delC", "c.12052G>A", "c.12067-2A>G", "c.12575G>A", "c.13274C>T", "c.14020A>G", "c.14023A>T", "c.14413G>A", "c.14424C>A", "c.2167+5G>A", "c.2209C>T", "c.236_239dupGTAC", "c.3368A>G", "c.377delG", "c.3959C>T", "c.4544C>T", "c.5078G>A", "c.5519G>T", "c.5776+1G>A", "c.6159delA", "c.6937G>T", "c.802G>A", "c.8558+1G>T", "c.8719A>C", "c.9685delG"}
1849 - 1849	USMG5:Leigh syndrome realted to USMG5 {(NM_032747) "c.87+1G>C"}
1850 - 1851	VDR:Rickets, vitamin D-resistant, type IIA {(NM_001017535.1) "c.277+1G>T", "c.885C>A"}
1852 - 1852	VIPAS39:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome 2 {(NM_022067) "c.808C>T"}
1853 - 1853	VPS11:Hypomyelination and developmental delay {(NM_021729.5) "c.2536T>G"}
1854 - 1857	VPS13A:Choreoacanthocytosis {(NM_033305.3) "c.2343delA", "c.6059delC", "c.9446_9449dup", "delexon70-73"}
1858 - 1859	VPS13B:Cohen syndrome {(NM_017890) "c.4894C>T" | (NM_017890.4) "c.6732+1G>A"}
1860 - 1861	VPS33B:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome {(NM_018668.4) "c.403+1G>A", "c.700G>C"}
1862 - 1862	VPS37A:Spastic paraplegia 53, Autosomal Recessive {(NM_152415.3) "c.1146A>T"}
1863 - 1863	VPS45:Neutropenia, severe congenital, 5, Autosomal Recessive {(NM_007259.5) "c.671C>A"}
1864 - 1865	VPS53:Pontocerebellar hypoplasia, type 2E (PCCA2) {(NM_001128159.3) "c.1556+5G>A", "c.2084A>G"}

1866 - 1866	VRK1:Pontocerebellar hypoplasia type 1A {(NM_003384.3) "c.1072C>T"}
1867 - 1869	WFS1:Wolfram-like syndrome, Autosomal Dominant {(NM_006005) "c.1230_1233delCTCT", "c.1770_1773delGTCT", "c.2590G>A"}
1870 - 1871	WISP3:Arthropathy, progressive pseudorheumatoid, of childhood {(NM_003880.3) "c.156C>A ", "c.536_537delGT"}
1872 - 1872	XPC:Xeroderma pigmentosum, group C {(NM_004628.4) "c.566_567delAT"}
1873 - 1873	XRCC2:Fanconi Anemia {(NM_005431.1) "c.643C>T"}
1874 - 1874	ZBTB24:Immunodeficiency-centromeric instability-facial anomalies syndrome-2 {(NM_014797.2) "c.501dupA"}
1875 - 1875	ZMPSTE24:Mandibuloacral dysplasia with type B lipodystrophy {(NM_005857) "c.1085dupT"}
1876 - 1877	ZNF469:Brittle cornea syndrome 1 {(NM_001127464.2) "c.5943delA", "c.9531delG"}

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לפי החלטת איגוד הגנטיקאים הישראלי, המוטציה מדווחת חיובית רק ליהודים ממוצא קווקזי, De boer M, Gavrieli R, Van leeuwen K, et al. A false-carrier state for the c.579G>A mutation in the NCF1 gene in Ashkenazi Jews. J Med Genet. 2018;55(3):166-172.

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