(2020-11-26) Ver 2 - Extended רשימת מחלות ומוטציות בפאנל
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 - 80	AGXT:Hyperoxaluria, primary, type I {(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"}
81 - 83	AHI1:Joubert syndrome-3 {(NM_017651.4) "c.2212C>T", "c.3032C>G", "c.787dupC"}
84 - 84	AIMP1:Leukodystrophy, hypomyelinating, 3 {(NM_004757.3) "c.292_293delCA"}
85 - 87	AIPL1:Leber congenital amaurosis 4 {(NM_014336.5) "c.211G>T", "c.215G>A", "c.834G>A"}
88 - 93	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"}
94 - 94	ALDH1A3:Microphthalmia, isolated 8 {(NM_000693.4) "c.211G>A"}
95 - 95	ALDH7A1:Epilepsy, pyridoxine-dependent {(NM_001182.5) "c.1489+5G>A"}
96 - 103	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"}
104 - 106	ALMS1:Alstrom syndrome {(NM_015120.4) "c.8008C>T", "c.808C>T", "c.8171_8181del"}
107 - 109	ALPL:Hypophosphatasia, infantile {(NM_000478) "c.1337delC" | (NM_000478.6) "c.1348C>T", "c.141C>A"}
110 - 110	AMT:Glycine encephalopathy, AMT-related {(NM_000481.3) "c.125A>G"}
111 - 111	ANO5:Limb-girdle muscular dystrophy {(NM_213599.2) "c.191dupA"}
112 - 112	AP4B1:Spastic paraplegia 47, Autosomal Recessive {(NM_006594.4) "c.664delC"}
113 - 113	APTX:Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia {(NM_175073.2) "c.837G>A"}
114 - 115	AQP2:Diabetes insipidus, nephrogenic {(NM_000486.5) "c.298G>T", "c.83T>C"}
116 - 116	ARFGEF2:Periventricular heterotopia with microcephaly {(NM_006420.3) "c.1958+1G>A"}
117 - 117	ARHGDIA:Nephrotic syndrome, type 8 {(NM_004309.6) "c.518G>T"}
118 - 118	ARL6:Bardet-Biedl syndrome 3 {(NM_032146.5) "c.364C>T"}
119 - 132	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"}
133 - 133	ARSG:Usher syndrome, type IV {(NM_014960) "c.133G>T"}
134 - 134	ASL:Argininosuccinic aciduria {(NM_000048.4) "c.346C>T"}
135 - 135	ASNS:Asparagine synthetase deficiency {(NM_183356.3) "c.1084T>G"}
136 - 139	ASPA:Canavan Disease {(NM_001128085.1) "c.433-2A>G", "c.693C>A", "c.854A>C", "c.914C>A"}
140 - 140	ASS1:Citrullinemia,classic {(NM_000050) "c.1168G>A"}
141 - 154	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"}
155 - 155	ATP6V0A2:Cutis laxa, Autosomal Recessive, type IIA {(NM_012463.4) "c.2375C>G"}
156 - 178	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"}
179 - 179	ATP8B1:Cholestasis, progressive familial intrahepatic 1 {(NM_005603) "c.2854C>T"}
180 - 180	AVP:Familial neurohypophyseal diabetes insipidus {(NM_000490) "c.77C>T"}
181 - 181	B3GALNT2:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11) {(NM_152490.5) "c.236-1G>C"}
182 - 182	B4GALT1:Congenital disorder of glycosylation, type IId {(NM_001497) "c.61C>T"}
183 - 185	BBS10:Bardet-Biedl syndrome 10 {(NM_024685.4) "c.1091delA", "c.1399delA", "c.271dupT"}
186 - 187	BBS1:Bardet-Biedl syndrome 1 {(NM_024649.5) "c.1169T>G", "c.479G>A"}
188 - 192	BBS2:Bardet-Biedl syndrome 2 {(NM_031885.4) "c.1895G>C", "c.224T>G", "c.311A>C", "c.401C>G", "c.98C>A"}
193 - 194	BBS4:Bardet-Biedl syndrome 4 {(NM_033028.5) "c.77-1422_221-753del ", "c.884G>C"}
195 - 195	BBS7:Bardet-Biedl syndrome 7 {(NM_176824.3) "c.1786G>A"}
196 - 197	BBS9:Bardet-Biedl syndrome 9 {(NM_014451) "c.1063C>T", "c.1669+1G>A"}
198 - 204	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"}
205 - 211	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"}
212 - 218	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"}
219 - 219	BMPER:Diaphanospondylodysostosis {(NM_133468.5) "c.310C>T"}
220 - 220	BMPR1B:Brachydactyly type A2 {(NM_001256793.2) "c.377G>A"}
221 - 222	BSND:Bartter syndrome, type 4a infantile variant with sensorineuronal deafness {(NM_057176.3) "c.167_168insTTTCCC", "c.28G>A"}
223 - 224	BTD:Biotinidase deficiency {(NM_000060) "c.393delC" | (NM_000060.4) "c.100G>A"}
225 - 227	C12ORF65:Spastic paraplegia 55, Autosomal Recessive {(NM_152269) "c.346delG" | (NM_152269.5) "c.282+2T>A", "c.413_417delAACAA"}
228 - 228	C21orf59:Ciliary dyskinesia, primary, 26 {(NM_021254.4) "c.735C>G"}
229 - 234	C2ORF71:Retinitis pigmentosa 54 {(NM_001029883) "c.2334T>A", "c.2756_2768delAGCCAGCCCTGGA", "c.3289C>T", "c.478_479insA", "c.556C>T", "c.776_777delAG"}
235 - 237	C8orf37:Retinitis pigmentosa 64 {(NM_177965.4) "c.497T>A", "c.529C>T", "c.545A>G"}

238 - 240	CAPN3:Muscular dystrophy, limb-girdle, type 2A {(NM_000070) "c.1076C>T", "c.1469G>A", "c.367C>A"}
241 - 241	CASQ2:Ventricular tachycardia, catecholaminergic polymorphic, 2 {(NM_001232.3) "c.919G>C"}
242 - 246	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"}
247 - 247	CC2D1A:Mental retardation, Autosomal Recessive 3 - MRT3 {(NM_017721.5) "c.1468+1_1824-1del"}
248 - 248	CC2D2A:Mental retardation, Autosomal Recessive 3 - MRT3 {(NM_001080522) "c.308delG"}
249 - 249	CCDC114:Ciliary dyskinesia, primary, 20 {(NM_144577) "c.939delT"}
250 - 250	CCDC174:Birk Volodarsky PMR Synderome Hypotonia and psychomotor developmental delay {(NM_016474.5) "c.1404A>G"}
251 - 251	CCDC65:Ciliary dyskinesia, primary, 27 {(NM_033124.5) "c.877_878delAT"}
252 - 252	CCDC88C:Hydrocephalus, nonsyndromic, Autosomal Recessive {(NM_001080414.4) "c.934C>T"}
253 - 256	CCNO:Ciliary dyskinesia, primary, 29 {(NM_021147) "c.165delC", "c.258_262dupGGCCC", "c.481_482delCT", "c.638T>C"}
257 - 257	CD59:Hemolytic anemia & immune-mediated polyneuropathy, CD59-related {(NM_203330.2) "c.266G>A"}
258 - 258	CDAN1:Dyserythropoietic anemia, congenital, type Ia {(NM_138477.4) "c.3124C>T"}
259 - 259	CDH23:Usher Syndrome Type ID {(NM_022124.6) "c.7903G>T"}
260 - 260	CDK10:Al Kaissi syndrome {(NM_052988) "c.870_871del"}
261 - 261	CDK5:Lissencephaly 7 with cerebellar hypoplasia {(NM_004935.4) "c.580+1G>A"}
262 - 262	CEACAM16:Deafness, autosomal recessive {(NM_001039213) "c.703C>T"}
263 - 263	CECR1:Adenosine deaminase 2 deficency {(NM_001282225.2) "c.140_141insT"}
264 - 264	CECR1:Polyarteritis nodosa, childhood-onset {(NM_001282225.2) "c.139G>A"}
265 - 265	CENPJ:Microcephaly, primary, Autosomal Recessive {(NM_018451.5) "c.3243_3246delTCAG"}
266 - 266	CEP104:Joubert syndrome (JBTS) {(NM_014704.4) "c.1328_1329insT"}
267 - 267	CEP152:Microcephaly 9, primary, Autosomal Recessive {(NM_014985.3) "c.2281-2A>G"}
268 - 276	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"}
277 - 277	CERKL:Retinitis pigmentosa 26 {(NM_001030311.2) "c.238+1G>A"}
278 - 278	CFH:Hemolytic uremic syndrome, complement factor H deficiency {(NM_000186.3) "c.3677_*4del"}
279 - 708	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"}
709 - 712	CHRNE:Myasthenic syndrome, congenital, 4B, fast-channel {(NM_000080) "c.1161_1162insT", "c.1353dupG", "c.187_188insC", "c.637dupG"}
713 - 716	CLCN1:Myotonia congenita, Autosomal Recessive {(NM_000083) "c.1444G>A", "c.1586C>T", "c.568_569delGGinsTC", "c.803C>T"}
717 - 718	CLCN1:Myotonia congenita,Autosomal Recessive {(NM_000083) "c.1012C>T", "c.1437_1450del"}

719 - 724	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"}
725 - 726	CLCNKB:Bartter syndrome, type 3 and Gitelman syndrome {(NM_000085) "c.1830G>A" | (NM_000085.4) "c.1313G>A"}
727 - 727	CLN5:Ceroid lipofuscinosis, neuronal, 5 {(NM_006493.3) "c.672delG"}
728 - 729	CLN6:Ceroid lipofuscinosis, neuronal, 6 {(NM_017882) "c.843G>A" | (NM_017882.3) "c.214G>T"}
730 - 730	CLN8:Neuronal ceroid lipofuscinosis type 8, including northern epilepsy {(NM_018941.3) "c.766C>G"}
731 - 734	CLRN1:Usher syndrome, type 3A {(NM_174878.2) "c.144T>G", "c.349_358del", "c.433+1G>A", "c.528T>G"}
735 - 736	CNGA1:Retinitis pigmentosa 49 {(NM_000087) "c.1540C>T", "c.94C>T"}
737 - 745	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"}
746 - 747	CNGB1:Retinitis pigmentosa 45 {(NM_001297) "c.2760G>A" | (NM_001297.5) "c.2284C>T"}
748 - 755	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"}
756 - 757	CNNM4:Jalili syndrome {(NM_020184.4) "c.1813C>T", "c.599C>A"}
758 - 758	CNTNAP1:Lethal congenital contracture syndrome 7 {(NM_003632) "c.2015G>A"}
759 - 759	COL11A2:Otospondylomegaepiphyseal dysplasia (ZW) {(NM_080680.2) "c.3991C>T"}
760 - 764	COL17A1:Epidermolysis bullosa, junctional, {(NM_000494) "c.2226insTGGA", "c.3676C>T", "c.3766+1G>A", "c.4145_4148delAGAG", "c.737_738insA"}
765 - 768	COL4A3:Alport Syndrome, COL4A3-Related {(NM_000091.4) "c.1791_1793dupTCC", "c.227delG", "c.3518-2A>G", "c.4649T>G"}
769 - 770	COL4A4:Alport syndrome, COL4A4-Related {(NM_000092.4) "c.3933C>G", "c.785_792dupCACCTGAC"}
771 - 776	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"}
777 - 780	COL7A1:Dystrophic epidermolysis bullosa, Autosomal Recessive,COL7A1-Related {(NM_000094) "c.2387G>A", "c.4888C>T", "c.6341delG", "c.682+1G>A"}
781 - 781	COLEC11:3MC syndrome 2 {(NM_199235.2) "c.627_628delCG"}
782 - 787	COLQ:Myasthenic syndrome, congenital, 5 {(NM_005677) "c.377delG", "c.788dupC", "c.893delA" | (NM_005677.4) "c.1228C>T", "c.718G>T", "c.792dupG"}
788 - 788	COQ4:Coenzyme Q10 deficiency, primary, 7 {(NM_016035.5) "c.718C>T"}
789 - 794	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"}
795 - 795	CPT1A:Carnitine palmitoyltransferase 1 deficiency {(NM_001031847) "c.1361A>G"}

796 - 797	CPT2:CPT deficiency, hepatic, type II {(NM_000098) "c.110_111dupGC", "c.1239_1240delGA"}
798 - 812	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"}
813 - 816	CRB2:Ventriculomegaly with cystic kidney disease {(NM_173689.7) "c.1882C>T", "c.1928A>C", "c.2277G>A", "c.2400C>G"}
817 - 818	CRTAP:Osteogenesis imperfecta, type VII {(NM_006371) "c.976C>T" | (NM_006371.4) "c.793+1G>T"}
819 - 819	CSTA:Exfoliative ichthyosis, Autosomal Recessive, ichthyosis bullosa of Siemens-like {(NM_005213.4) "c.67-2A>T"}
820 - 824	CTNS:Cystinosis,CTNS-related {(NM_004937) "c.587dupA", "c.691C>T", "c.890G>A" | (NM_004937.2) "c.1015G>A", "c.530A>C"}
825 - 825	CTSC:Haim-Munk syndrome {(NM_001814.6) "c.857A>G"}
826 - 826	CTSK:Pycnodysostosis {(NM_000396.4) "c.990A>G"}
827 - 831	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"}
832 - 836	CYBB:Chronic granulomatous disease, X-linked {(NM_000397) "c.1016dupC", "c.1081T>C", "c.271C>T", "c.676C>T", "c.90_92delCCGinsGGT"}
837 - 838	CYP11A1:Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete {(NM_000781.3) "c.644T>C", "c.694C>T"}
839 - 839	CYP11B2:Hypoaldosteronism, congenital, due to CMO II deficiency {(NM_000498.3) "c.541C>T"}
840 - 842	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"}
843 - 847	CYP27A1:Cerebrotendinous xanthomatosis {(NM_000784.4) "c.1016C>T", "c.1184G>A", "c.355delC", "c.819delT", "c.845-1G>A"}
848 - 848	CYP4F22:Congenital recessive ichthyoses (CRI) {(NM_173483) "c.429dupG"}
849 - 849	CYP4V2:Bietti crystalline corneoretinal dystrophy {(NM_207352) "c.1123delC"}
850 - 850	CYP7B1:Spastic paraplegia 5A, Autosomal Recessive {(NM_004820.5) "c.1081C>T"}
851 - 851	DAG1:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 {(NM_001165928.3) "c.743delC"}
852 - 852	DARS2:Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation {(NM_018122.5) "c.492+2T>C"}
853 - 854	DBT:Maple syrup urine disease, type II {(NM_001918) "c.581C>G", "c.939G>C"}
855 - 856	DCAF17:Woodhouse-Sakati syndome {(NM_025000) "c.580C>T" | (NM_025000.4) "c.436delC"}
857 - 857	DCLRE1C:Severe combined immunodeficiency, Athabascan type {(NM_001033858.2) "c.1307_1308insAGGATGCT"}
858 - 858	DDR2:Spondylometaepiphyseal dysplasia, short limb-hand type {(NM_006182.4) "c.2254C>T"}
859 - 859	DDRGK1:Spondyloepimetaphyseal dysplasia (Shohat-type) {(NM_023935)

	"c.408+1G>A"}
860 - 860	DDX11:Warsaw breakage syndrome {(NM_030653.3) "c.1763-1G>C"}
861 - 861	DGAT1:Diarrhea 7, congenital {(NM_012079.6) "c.751+2T>C"}
862 - 863	DGUOK:Mitochondrial DNA depletion syndrome (hepatocerebral type) {(NM_080916.3) "c.255delA", "c.271delA"}
864 - 864	DHCR24:Desmosterolosis {(NM_014762.4) "c.307C>T"}
865 - 881	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"}
882 - 882	DHDDS:Retinitis pigmentosa 59 {(NM_024887.3) "c.124A>G"}
883 - 886	DLD:Dihydrolipoamide Dehydrogenase Deficiency {(NM_000108.5) "c.104dupA", "c.1123G>A", "c.1436A>T", "c.685G>T"}
887 - 887	DLL3:Spondylocostal dysostosis 1, Autosomal Recessive {(NM_016941.3) "c.395delG"}
888 - 889	DNAH11:Ciliary dyskinesia, primary, 7, with or without situs inversus {(NM_001277115.2) "c.11929G>T", "c.13242_13245delAAAG"}
890 - 891	DNAH5:Ciliary dyskinesia, primary, 3, with or without situs inversus (CILD3/PCD) {(NM_001369.2) "c.7502G>C", "c.8011-2A>G"}
892 - 892	DNAI1:Ciliary dyskinesia, primary, 1, with or without situs inversus {(NM_012144.4) "c.1490G>A"}
893 - 894	DNAI2:Ciliary dyskinesia, primary, 9, with or without situs inversus {(NM_023036.6) "c.1304G>A", "c.1494+1G>A"}
895 - 895	DNAL1:Ciliary dyskinesia, primary, 16 {(NM_031427.4) "c.449A>G"}
896 - 896	DOCK8:Hyper-IgE recurrent infection syndrome, autosomal recessive {(NM_203447) "c.5132C>A"}
897 - 898	DOLK:Congenital disorder of glycosylation, type Im {(NM_014908.3) "c.1222C>G", "c.912G>T"}
899 - 900	DSG1:Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE {(NM_001942.4) "c.1861delG", "c.395C>A"}
901 - 901	DST:Epidermolysis bullosa simplex, Autosomal Recessive 2 {(NM_183380.3) "c.14865delA"}
902 - 902	DSTYK:Spastic paraplegia, complicated {(NM_015375) "4-kbdeletion/20-bpinsertion"}
903 - 908	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"}
909 - 909	ECHS1:Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency {(NM_004092) "c.476A>G"}
910 - 910	ECM1:Urbach-Wiethe disease {(NM_004425) "c.70+1G>C"}
911 - 911	EDAR:Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, Autosomal Recessive {(NM_022336) "c.259T>C"}
912 - 914	ELP1:Dysautonomia, familial {(NM_003640.5) "c.2087G>C", "c.2204+6T>C", "c.2741C>T"}
915 - 915	EOGT:Adams-Oliver syndrome 4 {(NM_001278689.2) "c.1074delA"}
916 - 919	EPG5:Vici syndrome {(NM_020964) "c.1007A>G", "c.3446G>A", "c.5993C>G" |

	(NM_020964.3) "c.5704dupT"}
920 - 920	EPM2A:Epilepsy, progressive myoclonic 2A (Lafora) {(NM_005670) "56_kb_incl._ex._2"}
921 - 921	ERBB3:Lethal congenital contractural syndrome 2 {(NM_001982.3) "c.1184-9A>G"}
922 - 922	ERCC2:Xeroderma pigmentosum, group D {(NM_000400.3) "c.2048G>A"}
923 - 923	ERCC5:Xeroderma pigmentosum/Cockayne {(NM_000123.3) "c.205C>T"}
924 - 924	ERCC6:Cockayne syndrome, type B {(NM_000124.4) "c.1034_1035insT"}
925 - 927	ERCC8:Cockayne syndrome, type A {(NM_000082.3) "c.37G>T", "c.843+1G>C", "c.966C>A"}
928 - 928	ESCO2:Roberts-SC phocomelia syndrome {(NM_001017420.3) "c.1674-2A>G"}
929 - 932	ETFDH:Glutaric acidemia IIC {(NM_004453.4) "c.1074G>C", "c.1084G>A", "c.1425C>A", "c.299T>A"}
933 - 933	EXOSC3:Pontocerebellar hypoplasia, type 1B {(NM_016042.4) "c.571G>T"}
934 - 934	EXOSC8:Pontocerebellar hypoplasia, type 1C {(NM_181503.3) "c.5C>T"}
935 - 949	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"}
950 - 951	F7:Factor VII deficiency {(NM_000131) "c.1109G>T" | (NM_000131.4) "c.1256C>T"}
952 - 952	FA2H:Spastic paraplegia 35, Autosomal Recessive {(NM_024306.5) "c.786+1G>A"}
953 - 959	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"}
960 - 965	FAM161A:Retinitis pigmentosa 28 {(NM_001201543.2) "c.1003C>T", "c.1309A>T", "c.1321dupC", "c.1355_1356delCA", "c.1567C>T", "c.1786C>T"}
966 - 966	FAM20A:Amelogenesis imperfecta, type IG (enamel-renal syndrome) {(NM_017565.4) "c.1523delC"}
967 - 975	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"}
976 - 982	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"}
983 - 984	FANCG:Fanconi Anemia - complementation group G {(NM_004629.1) "c.212T>C", "c.510+3A>G"}
985 - 985	FDX1L:Mitochondrial muscle myopathy {(NM_001031734.4) "c.10A>T"}
986 - 988	FERMT1:Kindler syndrome {(NM_017671.4) "c.0_-19+470del ", "c.137_140delTAGT", "c.749G>A"}
989 - 989	FGB:Afibrinogenemia congenital {(NM_005141.4) "c.1400G>A"}
990 - 990	FH:Fumarase deficiency, leiomyomatosis and renal cell cancer {(NM_000143.3) "c.905-1G>A"}
991 - 993	FKBP10:Osteogenesis imperfecta, type XI {(NM_021939)

	"c.1271_1272delCCinsA", "c.391+4A>T" | (NM_021939.3) "c.310C>T"}
994 - 994	FKRP:Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 {(NM_024301.5) "c.160C>T"}
995 - 995	FKTN:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 - Walker Warburg syndrome {(NM_001079802.1) "c.1167dupA"}
996 - 996	FLT4:Autosomal Recessive Hereditary Lymphedema {(NM_182925.5) "c.3704C>G"}
997 - 997	FOXRED1:Mitochondrial encephalomyopathy complex I deficiency {(NM_017547.4) "c.1054C>T"}
998 - 998	FRMD4A:Microcephaly intellectual disability and dysmorphism {(NM_018027) "c.2134_2146dup13"}
999 - 999	FTO:Growth retardation, developmental delay, coarse facies, and early death {(NM_001080432.3) "c.947G>A"}
1000 - 1001	G6PC3:Neutropenia, severe congenital 4, Autosomal Recessive {(NM_138387.3) "c.765_766delAG", "c.785G>A"}
1002 - 1013	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"}
1014 - 1026	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"}
1027 - 1029	GALC:Krabbe disease {(NM_000153.4) "c.1630G>A", "c.1748A>C", "c.1796T>G"}
1030 - 1031	GALNT3:Tumoral calcinosis, hyperphosphatemic, familial {(NM_004482.4) "c.1524+1G>A", "c.1524+5G>A"}
1032 - 1041	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"}
1042 - 1043	GAN:Giant axonal neuropathy 1 {(NM_022041) "c.103G>T" | (NM_022041.3) "c.973G>A"}
1044 - 1044	GATC:Hypertophic Cardiomyopathy {(NM_176818) "c.233T>G"}
1045 - 1045	GATM:Cerebral creatine deficiency syndrome 3 {(NM_001482.3) "c.1111dupA"}
1046 - 1057	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"}
1058 - 1069	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"}
1070 - 1071	GH1:Growth hormone deficiency, isolated, type IA {(NM_000515.5) "c.456+5G>C", "c.67G>T"}
1072 - 1077	GHR:Laron dwarfism {(NM_000163.5) "c.11G>A", "c.594A>G", "c.62G>A", "c.703C>T", "c.744delT", "del5,6ex"}
1078 - 1078	GHRHR:Growth hormone deficiency, isolated, type IB {(NM_000823.4)

	"c.1069C>T"}
1079 - 1079	GIPC3:Deafness, autosomal recessive 15 {(NM_133261) "c.937T>C"}
1080 - 1094	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"}
1095 - 1095	GJB6:Deafness, Autosomal Recessive 1B {(NM_006783.4) "309_kb"}
1096 - 1101	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"}
1102 - 1105	GLDC:Glycine encephalopathy and non-ketoic hyperglycinemia, GLDC-related {(NM_000170.2) "c.2405C>T", "c.2607C>A", "c.2T>C", "c.985C>A"}
1106 - 1106	GLRA1:Hyperekplexia, hereditary 1, autosomal dominant or recessive {(NM_001146040.1) "c.298C>T"}
1107 - 1107	GMPPA:Alacrima, achalasia, and mental retardation syndrome {(NM_013335.3) "c.1000A>C"}
1108 - 1109	GMPPB:Muscular dystrophy-dystroglycanopathy {(NM_013334.3) "c.656T>C", "c.860G>A"}
1110 - 1110	GNE:Hereditary inclusion body myopathy (HIBM) {(NM_005476.6) "c.2135T>C"}
1111 - 1117	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"}
1118 - 1118	GNPTG:Mucolipidosis III gamma {(NM_032520.5) "c.499dupC"}
1119 - 1120	GPC6:Omodysplasia 1 {(NM_005708) "g.93997007_94063501del66495insATAAATCACTTAGAGATGT", "g.94252984_94352299del99316insCTA"}
1121 - 1121	GPSM2:Chudley-McCullough syndrome {(NM_013296.5) "c.379C>T"}
1122 - 1122	GRHPR:Hyperoxaluria, primary, type II {(NM_012203.2) "c.975A>G"}
1123 - 1130	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"}
1131 - 1131	HACD1:Congenital myopathy {(NM_014241.4) "c.744C>A"}
1132 - 1132	HADHA:Long-Chain hydroxylacyl-CoA dehydrogenase deficiency (LCHAD) {(NM_000182.5) "c.1528G>C"}
1133 - 1133	HAX1:Severe congenital neutropenia type 3 (SCN3), a.k.a. Kostmann disease {(NM_006118) "c.125dupG"}
1134 - 1160	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"}
1161 - 1183	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"}
1184 - 1184	HEXB:Sandhoff disease, infantile, juvenile, and adult forms {(NM_000521) "c.1082+5G>A"}
1185 - 1185	HGD:Alkaptonuria {(NM_000187) "c.16-272_87+305del"}
1186 - 1186	HGSNAT:Retinitis pigmentosa 73 {(NM_152419.3) "c.370A>T"}
1187 - 1187	HIKESHI:Leukodystrophy, early onset spastic paraparesis,acquired microcephaly, optic atrophy and risk of early death {(NM_016401.4) "c.160G>C"}
1188 - 1190	HMGCL:HMG-CoA lyase deficiency {(NM_000191.3) "c.122G>A", "c.125A>G", "c.521G>A"}
1191 - 1191	HOGA1:Hyperoxaluria, primary, type III {(NM_138413) **"c.944_946delAGG"}
1192 - 1194	HPD:Thyrosinemia type III {(NM_002150.3) "c.325-1G>A", "c.415-1G>A", "c.481G>C"}
1195 - 1195	HPS1:Hermansky-Pudlak syndrome 1 {(NM_000195.5) "c.972delC"}
1196 - 1199	HPS3:Hermansky-Pudlak syndrome 3 {(NM_032383.5) "c.-2993_217+690del3900", "c.1163+1G>A", "c.1691+2T>G", "c.2482-2A>G"}
1200 - 1200	HPS6:Hermansky-Pudlak syndrome 6 {(NM_024747.5) "c.1065dupG"}
1201 - 1201	HSPD1:Leukodystrophy, hypomyelinating, 4, HLD4 (HSP60) {(NM_199440.1) "c.86A>G"}
1202 - 1202	IBA57:Spastic paraplegia 74, Autosomal Recessive {(NM_001010867.4) "c.678A>G"}
1203 - 1206	IDUA:Mucopolysaccharidosis Type IH - Hurler syndrome {(NM_000203.5) "c.1096A>C", "c.192C>A", "c.208C>T", "c.928C>T"}
1207 - 1208	IGHMBP2:Neuronopathy, distal hereditary motor, type VI {(NM_002180.2) "c.114delA", "c.707T>G"}
1209 - 1209	IL10RA:Inflammatory bowel disease 28, early onset, autosomal recessive {(NM_001558) "c.537G>A"}
1210 - 1213	INSR:Leprechaunism, Donohue syndrome {(NM_000208) "c.2683-542_2842+544del" | (NM_000208.4) "c.167T>C", "c.3079C>T", "c.857G>A"}
1214 - 1214	INVS:Nephronophthisis 2, infantile {(NM_014425.5) "c.2719C>T"}
1215 - 1215	ISPD:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), {(NM_001101426.4) "c.165dupG"}
1216 - 1219	ITGA2B:Glanzmann thrombasthenia, ITGA2B-related {(NM_000419) "c.2374delG" | (NM_000419.4) "c.1947-1G>A", "c.818G>A", "c.97A>G"}
1220 - 1222	ITGB3:Glanzmann thrombasthenia, ITGB3-related {(NM_000212) "c.1616_1617delTT" | (NM_000212.2) "11.2kbincl.ex.10-partex.13", "c.428T>G"}
1223 - 1223	ITGB4:Epidermolysis bullosa, junctional, with pyloric atresia - Carmi syndrome {(NM_000213.5) "c.3224_3793+120del"}
1224 - 1224	ITK:Lymphoproliferative syndrome {(NM_005546) "c.1764C>G"}
1225 - 1228	IVD:Isovaleric academia {(NM_002225.4) "c.148C>T", "c.286+2T>C", "c.456+2T>C", "c.932C>T"}
1229 - 1229	JAK3:SCID, autosomal recessive, T-negative/B-positive type {(NM_000215) "c.2680+89G>A"}
1230 - 1230	KCNJ10:SESAME syndrome {(NM_002241.5) "c.524G>A"}

1231 - 1231	KIAA1279:Goldberg-Shprintzen megacolon syndrome {(NM_015634) "c.1516dupA"}
1232 - 1232	KIF1C:Spastic ataxia 2, Autosomal Recessive {(NM_006612) "c.2191C>T"}
1233 - 1233	KIZ:Retinitis pigmentosa 69 {(NM_018474) "c.226C>T"}
1234 - 1234	KLHL40:Nemaline myopathy 8, Autosomal Recessive {(NM_152393.4) "c.581T>A"}
1235 - 1235	KREMEN1:Ectodermal dysplasia {(NM_032045) "c.626T>C"}
1236 - 1237	KRT14:Epidermolysis bullosa simplex {(NM_000526) "c.400C>T", "c.915G>A"}
1238 - 1239	KY:Myopathy, myofibrillar, 7 {(NM_178554) "c.405C>A", "c.51_52insTATCGACATGTGCTGTATCTATCGACAT"}
1240 - 1245	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"}
1246 - 1249	LAMA3:Laryngoonychocutaneous Syndrome {(NM_000227.4) "c.1981C>T", "c.2975delA", "c.4815G>T", "c.893_894insT"}
1250 - 1260	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"}
1261 - 1262	LAMC2:Epidermolysis bullosa, junctional, Herlitz type {(NM_018891.2) "c.1756C>T", "c.368_373delinsACCAC"}
1263 - 1267	LCA5:Leber congenital amaurosis 5 {(NM_181714.3) "c.1062_1068delCGAAAAC", "c.1714C>T", "c.238C>T", "c.835C>T", "c.94delT"}
1268 - 1269	LIFR:Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome-LIFR related {(NM_002310.5) "c.1601-1G>A", "c.2472_2476delTATGT"}
1270 - 1271	LIPA:Wolman disease {(NM_001127605.2) "c.260G>T", "c.398delC"}
1272 - 1273	LOXHD1:Deafness, Autosomal Recessive 77 {(NM_144612) "c.5894dupG" | (NM_144612.6) "c.4714C>T"}
1274 - 1275	LRBA:Immunodeficiency, common variable, 8, with autoimmunity {(NM_001199282) "c.8139_8142dupCATG" | (NM_001199282.2) "c.7937T>G"}
1276 - 1277	MAK:Retinitis pigmentosa 62 {(NM_001242957.2) "c.497G>A" | (NM_005906) "c.394_395insCTTC"}
1278 - 1278	MAN1B1:Mental retardation, Autosomal Recessive 15 {(NM_016219.5) "c.1863G>A"}
1279 - 1279	MATN3:Spondyloepimetaphyseal dysplasia {(NM_002381.5) "c.910T>A"}
1280 - 1280	MCIDAS:Mucociliary clearance disorder {(NM_001190787.2) "c.1142G>A"}
1281 - 1285	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"}
1286 - 1287	MECR:Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities {(NM_016011) "c.695G>A", "c.830+2dupT"}
1288 - 1288	MED17:Microcephaly, postnatal progressive, with seizures and brain atrophy ((ICCA) {(NM_004268.5) "c.1112T>C"}
1289 - 1289	MED25:Basel-Vanagaite-Smirin-Yosef syndrome {(NM_030973.3) "c.116A>G"}

1290 - 1290	MEGF10:Myopathy, areflexia, respiratory distress, and dysphagia, early-onset {(NM_001256545.2) "c.1325delC"}
1291 - 1291	MERTK:Retinitis pigmentosa 38 {(NM_006343) "c.2164C>T"}
1292 - 1293	MFSD8:Ceroid lipofuscinosis, neuronal, 7 {(NM_152778) "c.103C>T" | (NM_152778.2) "c.472G>A"}
1294 - 1295	MKS1:Meckel syndrome 1 {(NM_017777.3) "c.1048C>T", "c.472C>T"}
1296 - 1298	MLC1:Megalencephalic leukoencephalopathy with subcortical cysts {(NM_015166.3) "c.176G>A", "c.274C>T", "c.278C>T"}
1299 - 1299	MLPH:Griscelli syndrome, type 3 {(NM_024101.7) "c.103C>T"}
1300 - 1300	MMACHC:Methylmalonic aciduria and homocystinuria, cblC type {(NM_015506.3) "c.271dupA"}
1301 - 1303	MOCS1:Molybdenum cofactor deficiency A {(NM_001075098.3) "c.1510C>T", "c.722delT", "c.971G>A"}
1304 - 1305	MOCS2:Molybdenum cofactor deficiency Type B {(NM_004531.5) "c.226G>A", "c.377+1G>A"}
1306 - 1309	MPDU1:Congenital disorder of glycosylation, type If {(NM_004870) "c.511delC" | (NM_004870.4) "c.218G>A", "c.2T>C", "c.356T>C"}
1310 - 1315	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"}
1316 - 1316	MPV17:Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) {(NM_002437.5) "c.278A>C"}
1317 - 1317	MRE11A:Ataxia Telangiectasia like disorder {(NM_005591.3) "c.290A>G"}
1318 - 1320	MTHFR:Homocystinuria due to MTHFR deficiency {(NM_005957) "c.1072C>T" | (NM_005957.4) "c.16delA", "c.474A>T"}
1321 - 1324	MTTP:Abetalipoproteinemia ABL {(NM_000253.3) "c.2212delT", "c.2593G>T", "c.307A>T", "c.62-2A>G"}
1325 - 1326	MUT:Methylmalonic acidemia, mut(0) type {(NM_000255) "c.1240G>T" | (NM_000255.4) "c.655A>T"}
1327 - 1327	MVK:Hyper-IgD syndrome {(NM_000431.4) "c.1129G>A"}
1328 - 1330	MYBPC1:Lethal congenital contracture syndrome 4 {(NM_002465.4) "c.556G>A", "c.688G>A", "c.952C>T"}
1331 - 1332	MYH2:Proximal myopathy and ophthalmoplegia {(NM_017534.6) "c.2400delG", "c.706G>A"}
1333 - 1339	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"}
1340 - 1359	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"}
1360 - 1360	NAGLU:Mucopolysaccharidosis type IIIB (Sanfilippo B) {(NM_000263.4) "c.2021G>A"}
1361 - 1361	NARS2:Combined oxidative phosphorylation deficiency 24 (COXPD24) {(NM_024678) "c.500A>G"}
1362 - 1363	NBEAL2:Gray platelet syndrome {(NM_015175.2) "c.2701C>T", "c.5413dupG"}

1364 - 1366	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"}
1367 - 1370	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"}
1371 - 1371	NDUFA11:Mitochondrial complex I deficiency - NDUFA11 gene {(NM_001193375.1) "c.97+5G>A"}
1372 - 1372	NDUFAF5:Mitochondrial complex I deficiency - NDUFAF5 gene {(NM_024120.5) "c.749G>T"}
1373 - 1373	NDUFS2:Mitochondrial complex I deficiency-NDUFS2 gene {(NM_004550.4) "c.1237T>C"}
1374 - 1374	NDUFS4:Leigh syndrome {(NM_002495.4) "c.462delA"}
1375 - 1375	NDUFS6:Mitochondrial complex I deficiency - NDUFS6 gene {(NM_004553.4) "c.344G>A"}
1376 - 1379	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"}
1380 - 1380	NECTIN1:Cleft lip/palate ectodermal dysplasia, CLPED1 (Zlotogora-Ogur syndrome) {(NM_203285) "c.556delG"}
1381 - 1381	NGLY1:Congenital disorder of deglycosylation {(NM_018297.4) "c.1294G>T"}
1382 - 1382	NNT:Glucocorticoid deficiency 4 {(NM_182977.3) "c.598G>A"}
1383 - 1401	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"}
1402 - 1402	NPHP1:Joubert syndrome {"del exons 2-7"}
1403 - 1411	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"}
1412 - 1413	NPHS2:Nephrotic syndrome {(NM_014625) "c.388G>A" | (NM_014625.3) "c.412C>T"}
1414 - 1415	NRL:Retinitis pigmentosa 27 {(NM_006177) "c.444_445insGCTGCGGG", "c.91C>T"}
1416 - 1419	NTRK1:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_002529.3) "c.1250C>T", "c.1860_1861insT", "c.207_208delTG", "c.2084C>T"}
1420 - 1420	NUP62:Striatonigral degeneration, Infantile Bilateral Striatal Necrosis (IBSN) {(NM_016553.4) "c.1172A>C"}
1421 - 1421	OAT:Gyrate atrophy of choroid and retina with or without ornithinemia {(NM_000274) "c.159delC"}
1422 - 1423	OCA2:Albinism, oculocutaneous, type II {(NM_000275) "c.79G>A" | (NM_000275.3) "c.1327G>A"}
1424 - 1424	OPA3:3-methylglutaconic aciduria, type III - Costeff {(NM_025136.3) "c.143-1G>C"}
1425 - 1427	OTC:Ornithine transcarbamylase deficiency {(NM_000531.6) "c.717+1G>T", "c.829C>T", "c.958C>T"}

1428 - 1429	OTOA:Deafness, Autosomal Recessive 22 {(NM_144672) "c.1025A>T", "c.2359G>T"}
1430 - 1431	OTOF:Deafness, Autosomal Recessive 9 {(NM_194248) "c.5332G>T" | (NM_194248.2) "c.2866+1G>A"}
1432 - 1432	P3H2:Myopia, high, with cataract and vitreoretinal degeneration {(NM_018192) "c.1523G>T"}
1433 - 1472	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"}
1473 - 1473	PARK2:Parkinson disease, early onset {(NM_004562) "c.101delA"}
1474 - 1474	PAX7:Myopathy, congenital, progressive, with scoliosis {(NM_001135254) "c.1403-2A>G"}
1475 - 1475	PCCA:Propionic acidemia, PCCA-related {(NM_000282.4) "c.923dupT"}
1476 - 1476	PCCB:Propionic acidemia, PCCB-related {(NM_000532.5) "c.1173dupT"}
1477 - 1478	PCDH12:Microcephaly, seizures, spasticity, and brain calcification (MISSBC) {(NM_016580) "c.2515C>T", "c.995delT"}
1479 - 1479	PCDH15:Usher syndrome, type 1F {(NM_033056.3) "c.733C>T"}
1480 - 1480	PCK1:Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency {(NM_002591.4) "c.134T>C"}
1481 - 1482	PCNT:Microcephalic osteodysplastic primordial dwarfism, type II {(NM_006031) "c.2984_2994delCAGACTTTGAG" | (NM_006031.5) "c.3465-1G>A"}
1483 - 1487	PDE6A:Retinitis pigmentosa 43 {(NM_000440) "c.1957C>T", "c.1960C>T", "c.2081_2085delAACAG", "c.409delGinsCT", "c.769C>T"}
1488 - 1488	PDE6B:Retinitis pigmentosa-40 {(NM_001145291) "c.1417delC"}
1489 - 1489	PDE6G:Retinitis pigmentosa 57 {(NM_002602.4) "c.187+1G>T"}
1490 - 1492	PEPD:Prolidase deficiency {(NM_000285.4) "c.1103T>G", "c.605C>T", "c.634G>C"}
1493 - 1495	PEX1:Peroxisome biogenesis disorder 1A (Zellweger) {(NM_000466) "c.2916delA" | (NM_000466.3) "c.2097dupT", "c.2528G>A"}
1496 - 1498	PEX2:Peroxisome biogenesis disorder 5A (Zellweger) {(NM_001079867.1) "c.355C>T", "c.550delT", "c.669G>A"}
1499 - 1503	PEX6:Peroxisome biogenesis disorder 4B (Zellweger syndrome) {(NM_000287.4) "c.1715C>T", "c.1944delC", "c.1947delG", "c.2094+2T>C", "c.2534T>C"}
1504 - 1504	PEX7:Rhizomelic chondrodysplasia punctata type 1 {(NM_000288.4) "c.283T>G"}
1505 - 1505	PGAP3:Hyperphosphatasia with mental retardation syndrome 4 {(NM_033419.5) "c.845A>G"}
1506 - 1506	PGM1:Congenital disorder of glycosylation, type It {(NM_002633) "c.112A>T"}
1507 - 1507	PHGDH:Phosphoglycerate dehydrogenase deficiency {(NM_006623.3)

	"c.1468G>A"}
1508 - 1508	PHKG2:Glycogen storage disease IXc {(NM_000294.3) "c.71A>G"}
1509 - 1509	PHYH:Refsum disease {(NM_001037537.1) "c.523C>T"}
1510 - 1511	PIGN:Multiple congenital anomalies-hypotonia-seizures syndrome 1 {(NM_012327.5) "c.2126G>A", "c.755A>T"}
1512 - 1513	PIGT:Multiple congenital anomalies-hypotonia-seizures syndrome 3 {(NM_015937.6) "c.1564T>G", "c.761delG"}
1514 - 1514	PIP5K1C:Lethal congenital contractural syndrome 3 {(NM_012398.2) "c.757G>A"}
1515 - 1515	PJVK:Deafness, Autosomal Recessive 59 {(NM_001042702.4) "c.406C>T"}
1516 - 1526	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"}
1527 - 1531	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"}
1532 - 1532	PLAA:Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies {(NM_001031689.3) "c.2254C>T"}
1533 - 1533	PLEKHG2:Leukodystrophy and acquired microcephaly with or without dystonia {(NM_022835.3) "c.610C>T"}
1534 - 1537	PMM2:Congenital disorder of glycosylation Ia {(NM_000303) "c.338C>T", "c.357C>A", "c.422G>A", "c.691G>A"}
1538 - 1538	POC1A:Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis {(NM_015426.5) "c.512T>C"}
1539 - 1539	POMGNT2:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8) {(NM_032806.6) "c.1232_1233delAG"}
1540 - 1541	POMT1:Walker-Warburg Syndrome, type A, 1 {(NM_007171) "c.2167dupG", "c.428-1G>C"}
1542 - 1542	POMT2:Walker-Warburg Syndrome, type A, 2 {(NM_013382) "c.924-2A>C"}
1543 - 1543	POR:Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis {(NM_000941.3) "c.1615G>A"}
1544 - 1544	PPIB:Osteogenesis imperfecta, type IX {(NM_000942.4) "c.563_566delACAG"}
1545 - 1545	PPP1R13L:Cardio-Cutaneous Syndrome DCM {(NM_006663.4) "c.2241C>G"}
1546 - 1546	PPT1:Ceroid lipofuscinosis, neuronal, 1 {(NM_000310.3) "c.169dupA"}
1547 - 1547	PRCD:Retinitis pigmentosa 36 {(NM_001077620) "c.64C>T"}
1548 - 1548	PRICKLE1:Epilepsy, progressive myoclonic 1B {(NM_153026.3) "c.311G>A"}
1549 - 1549	PSMB8:Autoinflammation, lipodystrophy, and dermatosis syndrome {(NM_148919.4) "c.405C>A"}
1550 - 1550	PTPN23:Developmental delay, cognitive impairment, and atopic atrophy {(NM_015466) "c.3886_3888del"}
1551 - 1551	PUS1:Mitochondrial myopathy and sideroblastic anemia 1 {(NM_001002020.3) "c.346C>T"}
1552 - 1552	RAB27A:Griscelli syndrome, type 2 {(NM_004580) "c.148_149delinsC"}
1553 - 1553	RAB28:Cone-rod dystrophy 18 {(NM_001017979) "c.409C>T"}
1554 - 1555	RAG1:Severe combined immudeficiency, B cell-negative, RAG1-related {(NM_000448.2) "c.1361T>A", "c.1410_1413delCTTG"}

1556 - 1560	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"}
1561 - 1564	RAPSN:Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency {(NM_005055.5) "c.-210A>G", "c.-27C>G", "c.264C>A", "c.672_673insACT"}
1565 - 1565	RAPSN:Severe combined immudeficiency, B cell-negative, RAG2-related {(NM_005055) "c.648T>A"}
1566 - 1566	RARS2:Pontocerebellar hypoplasia, type 6 {(NM_020320.5) "c.110+5A>G"}
1567 - 1574	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"}
1575 - 1576	RECQL2:Werner syndrome {(NM_000553.5) "c.1105C>T", "c.2665C>T"}
1577 - 1577	RFX5:Bare lymphocyte syndrome, type II (SCID) {(NM_000449) "c.715C>T"}
1578 - 1578	RIN2:Macrocephaly, alopecia, cutis laxa, and scoliosis {(NM_018993.3) "c.1731delC"}
1579 - 1579	RNASEH2B:Aicardi-Goutieres syndrome 2 {(NM_024570.3) "c.529G>A"}
1580 - 1580	ROGDI:Kohlschutter-Tonz syndrome {(NM_024589.2) "c.469C>T"}
1581 - 1582	RP1:Retinitis pigmentosa 1 {(NM_006269) "c.688G>T" | (NM_006269.2) "c.4941dupT"}
1583 - 1588	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"}
1589 - 1592	RPGRIP1:Cone-rod dystrophy 13 {(NM_020366) "c.1615_1624delGAACTGGAGG", "c.2935C>T", "c.2974delA", "c.3663_3666delAGAA"}
1593 - 1593	RPGRIP1L:Meckel syndrome 5 {(NM_015272.5) "c.118C>T"}
1594 - 1594	RRM2B:Mitochondrial DNA depletion syndrome 8 {(NM_015713.5) "c.215C>G"}
1595 - 1595	RSPH9:Ciliary dyskinesia, primary, 12 {(NM_152732.5) "c.804_806delGAA"}
1596 - 1600	RTEL1:Dyskeratosis congenita {(NM_001283009.1) "c.1476G>T", "c.2848C>T", "c.2869C>T", "c.2920C>T", "c.3791G>A"}
1601 - 1603	RYR1:Minicore myopathy with external ophthalmoplegia {(NM_000540) "c.1366G>A", "c.9047A>G" | (NM_000540.2) "c.9623C>T"}
1604 - 1605	SAMD9:Tumoral calcinosis, familial, normophosphatemic {(NM_017654.4) "c.1030C>T", "c.4483A>G"}
1606 - 1610	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"}
1611 - 1611	SARS2:Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis {(NM_017827.3) "c.1169A>G"}
1612 - 1612	SCAPER:Retinitis pigmentosa with intellectual disability {(NM_020843) "c.2806delC"}
1613 - 1614	SCN9A:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_002977.3) "c.1124delG", "c.2687G>A"}
1615 - 1615	SCNN1A:Pseudohypoaldosteronism type I - SCNN1A gene {(NM_001038) "c.1522C>T"}
1616 - 1617	SCNN1B:Pseudohypoaldosteronism type I - SCNN1B gene {(NM_000336)

	"c.648dupA", "c.915delC"}
1618 - 1619	SDHA:Cardiomyopathy, dilated , 1GG neonatal isolated {(NM_004168) "c.1A>G" | (NM_004168.4) "c.1664G>A"}
1620 - 1621	SEC23B:Dyserythropoietic anemia, congenital, type II {(NM_006363.6) "c.2129C>T", "c.325G>A"}
1622 - 1626	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"}
1627 - 1627	SGCG:Muscular dystrophy, limb-girdle, type 2C {(NM_000231.2) "c.525delT"}
1628 - 1635	SGSH:Mucopolysaccharidisis type IIIA (Sanfilippo A) {(NM_000199) "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"}
1636 - 1636	SLC12A3:Bartter Syndrome, Gitelman Variant {(NM_000339.3) "c.1313G>A"}
1637 - 1637	SLC17A5:Sialic acid storage disorder, infantile (ISSD) {(NM_012434.5) "c.983G>A"}
1638 - 1638	SLC18A3:Myasthenia gravis, congenital {(NM_003055) "c.1078G>C"}
1639 - 1640	SLC19A2:Thiamine-responsive megaloblastic anemia syndrome {(NM_006996.3) "c.1223+1G>A", "c.725delC"}
1641 - 1643	SLC1A4:Spastic tetraplegia, thin corpus callosum, and progressive microcephaly {(NM_003038.5) "c.1369C>T", "c.766G>A", "c.944_945del"}
1644 - 1644	SLC22A5:Carnitine deficiency, systemic primary {(NM_003060.3) "c.1196G>A"}
1645 - 1646	SLC25A15:Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome {(NM_014252) "c.562_564delTTC" | (NM_014252.3) "c.706A>G"}
1647 - 1648	SLC25A1:Combined D-2- and L-2-hydroxyglutaric aciduria {(NM_005984.5) "c.389G>A", "c.845G>A"}
1649 - 1650	SLC25A20:Carnitine-acylcarnitine translocase deficiency - CACT {(NM_000387.6) "c.609-3C>G", "c.713A>G"}
1651 - 1651	SLC26A3:Congenital chloride diarhhea (CLD) {(NM_000111.2) "c.559G>T"}
1652 - 1662	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"}
1663 - 1666	SLC29A3:Histiocytosis-lymphadenopathy plus syndrome {(NM_018344.5) "c.1157G>A" | (NM_018344.6) "c.1045delC", "c.1279G>A", "c.1309G>A"}
1667 - 1669	SLC2A2:Fanconi-Bickel syndrome {(NM_000340.2) "c.372A>C", "c.734A>C", "c.901C>T"}
1670 - 1670	SLC30A9:Birk-Landau-Perez cerebro-renal syndrome {(NM_016474.5) "c.1047_1049delCAG"}
1671 - 1672	SLC35A3:Arthrogryposis, mental retardation, and seizures {(NM_012243.3) "c.514C>T", "c.886A>G"}
1673 - 1673	SLC35C1:Congenital disorder of glycosylation, type IIc {(NM_018389.4) "c.923C>G"}
1674 - 1677	SLC37A4:Glycogen storage disease Ib {(NM_001164277.1) "c.1042_1043delCT", "c.1179G>A", "c.446G>A", "c.83G>A"}
1678 - 1678	SLC39A4:Acrodermatitis enteropathica {(NM_130849.3) "c.1224delC"}
1679 - 1679	SLC45A2:Albinism, oculocutaneous, type IV {(NM_001012509) "c.1076_1077delAG"}

1680 - 1680	SLC46A1:Folate malabsorption, hereditary {(NM_080669) "c.337C>T"}
1681 - 1681	SLC4A4:Renal tubular acidosis (RTA), proximal, with ocular abnormalities and mental retardation {(NM_003759.3) "c.2321G>A"}
1682 - 1682	SLCO2A1:Hypertrophic osteoarthropathy, primary, Autosomal Recessive 2 {(NM_005630.2) "c.1292delC"}
1683 - 1684	SMARCAL1:Schimke immunoosseous dysplasia {(NM_014140.3) "c.2542G>T", "c.863-2A>G"}
1685 - 1685	SMN1:Spinal muscular atrophy-1 {(NM_000344) "c.835_*3del"}
1686 - 1698	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"}
1699 - 1699	SNAP29:Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome - CEDNIK Syndrome {(NM_004782) "c.223delG"}
1700 - 1700	SNX10:Osteopetrosis, Autosomal Recessive 8 {(NM_001199835.1) "c.152G>A"}
1701 - 1704	SPG11:Spastic paraplegia 11, Autosomal Recessive {(NM_025137) "c.5986dupT" | (NM_025137.4) "c.118C>T", "c.2471dupT", "c.4339C>T"}
1705 - 1709	SPINK5:Netherton syndrome {(NM_001127698.1) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC", "c.995delT"}
1710 - 1710	ST3GAL3:Early infantile epileptic encephalopathy 15 {(NM_006279.4) "c.958G>C"}
1711 - 1711	STRA6:Microphthalmia {(NM_001142617.1) "c.1678G>C"}
1712 - 1713	STRC:Deafness, Autosomal Recessive 16 {(NM_153700.2) "EX7_EX29DEL", "c.4171C>G"}
1714 - 1714	SUCLA2:Mitochondrial DNA depletion syndrome 5 {(NM_003850) "c.788_802+29del"}
1715 - 1716	SUMF1:Multiple sulfatase deficiency {(NM_182760.3) "c.1043C>T", "c.463T>C"}
1717 - 1718	SURF1:Leigh syndrome, due to COX deficiency {(NM_003172) "c.312_321delTCTGCCAGCCinsAT", "c.575_576insTGCG"}
1719 - 1719	SYNE4:Deafness, Autosomal Recessive 76 {(NM_001039876.3) "c.228_229delAT"}
1720 - 1720	SZT2:Epileptic encephalopathy, early infantile, 18 {(NM_015284.3) "c.73C>T"}
1721 - 1722	SepSecS:Pontocerebellar hypoplasia type 2D {(NM_016955.4) "c.1001A>G", "c.715G>A"}
1723 - 1723	TAF2:Mental retardation, Autosomal Recessive 40 {(NM_003184.4) "c.557C>G"}
1724 - 1724	TBCD:Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM_005993.4) "c.1423G>A"}
1725 - 1726	TBCE:Hypoparathyroidism retardation dysmorphism syndrome {(NM_003193.5) "c.155_166delGCCACGAAGGGA", "c.355_356del"}
1727 - 1727	TBX19:Adrenocorticotropic hormone deficiency {(NM_005149.3) "c.574_577delATAG"}
1728 - 1731	TCIRG1:Osteopetrosis, Autosomal Recessive 1 {(NM_006019.4) "c.117+4A>T", "c.1331G>T", "c.1384_1386delAAC", "c.674delG"}
1732 - 1732	TCTN2:Meckel syndrome 8 {(NM_024809.5) "c.1506-2A>G"}

1733 - 1735	TECPR2:Spastic paraplegia 49, Autosomal Recessive {(NM_001172631.2) "c.1319delT", "c.3416delT", "c.566C>T"}
1736 - 1736	TGM1:Ichthyosis, congenital, Autosomal Recessive 1 {(NM_000359) "c.2290C>T"}
1737 - 1737	THG1L:Cerebellar ataxia and developmental delay {(NM_017872.5) "c.164T>C"}
1738 - 1738	TIMM50:3-methylglutaconic aciduria, type IX {(ENST00000314349.4) "c.649C>T"}
1739 - 1741	TK2:Mitochondrial DNA depletion syndrome 2 (myopathic type) {(NM_004614.5) "c.360_361delGCinsAA", "c.361C>A", "c.635T>A"}
1742 - 1742	TKT:Short stature, developmental delay, and congenital heart defects {(NM_001135055.2) "c.769_770insCTACCTCCTTATCTTCTG"}
1743 - 1747	TMC1:Deafness, Autosomal Recessive 7 {(NM_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"}
1748 - 1748	TMEM165:Congenital disorder of glycosylation {(NM_018475.4) "c.792+182G>A"}
1749 - 1751	TMEM216:Joubert syndrome 2 (MKS2) {(NM_001173990.3) "c.218G>A", "c.218G>T", "c.230G>C"}
1752 - 1752	TMEM231:Meckel syndrome 11 {(NM_001077418.3) "c.664+4A>G"}
1753 - 1753	TMEM260:Neurodevelopmental, Cardiac, and Renal Syndrome {(NM_017799.3) "c.1393C>T"}
1754 - 1755	TMEM38B:Osteogenesis imperfecta, type XIV {(NM_018112) "c.455_542del", "c.507G>A"}
1756 - 1758	TMEM67:Joubert syndrome type 6 (MSK3) {(NM_153704) "c.1065+1delG" | (NM_153704.5) "c.1975C>T", "c.725A>G"}
1759 - 1761	TMEM70:ATPase deficiency, nuclear encoded {(NM_017866) "c.336T>A" | (NM_017866.6) "c.238C>T", "c.316+1G>T"}
1762 - 1763	TMPRSS3:Deafness, Autosomal Recessive 8/10 {(NM_024022) "c.1177_1184delins" | (NM_024022.2) "c.989delA"}
1764 - 1764	TNNT1:Nemaline myopathy 5, Amish type {(NM_003283) "c.574_577delinsTAGTGCTGT"}
1765 - 1765	TPP1:Ceroid lipofuscinosis, neuronal, 2 {(NM_000391) "c.775delC"}
1766 - 1766	TRAK1:Encephalopathy, fatal {(NM_001042646.2) "c.287-2A>C"}
1767 - 1767	TRAPPC9:Mental retardation, Autosomal Recessive 13 {(NM_031466.7) "c.1423C>T"}
1768 - 1768	TRIM32:Bardet-Biedl syndrome 11 {(NM_012210) "c.388C>T"}
1769 - 1769	TRIOBP:Deafness, Autosomal Recessive 28 {(NM_001039141) "c.1741C>T"}
1770 - 1771	TRMT10A:Microcephaly, short stature, and impaired glucose metabolism {(NM_152292.4) "c.616G>A", "c.727C>T"}
1772 - 1774	TRMU:LIFT, Liver failure infantile transient {(NM_018006) "c.500_509del10", "c.835G>A" | (NM_018006.5) "c.229T>C"}
1775 - 1778	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"}
1779 - 1780	TRPM6:Hypomagnesemia 1, intestinal {(NM_017662.5) "c.1010+5G>C", "c.2009+1G>A"}
1781 - 1782	TSHR:Hypothyroidism, congenital, nongoitrous, 1 {(NM_000369) "c.202C>T"

	| (NM_000369.2) "c.1825C>T"}
1783 - 1783	TSPAN12:Vitroretinal vascular malformations, congenital {(NM_012338) "c.542G>T"}
1784 - 1785	TTN:Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis {(NM_003319.4) "c.58881dupA" | (NM_133432) "c.36122delC"}
1786 - 1786	TUBGCP4:Microcephaly, primary, Autosomal Recessive {(NM_014444.5) "c.579dupT"}
1787 - 1789	TULP1:Retinitis pigmentosa 14 {(NM_003322) "c.849_852dup" | (NM_003322.6) "c.1349G>A", "c.1495+2dupT"}
1790 - 1791	TYMP:Mitochondrial DNA depletion syndrome 1 (MNGIE type) {(NM_001113755.2) "c.433G>A", "c.866A>C"}
1792 - 1808	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"}
1809 - 1809	UNC13D:Hemophagocytic lymphohistiocytosis, familial, 3 {(NM_199242.2) "c.753+1G>T"}
1810 - 1811	UNC80:Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM_032504) "c.7183C>T" | (NM_032504.1) "c.151C>T"}
1812 - 1812	UPB1:Beta-ureidopropionase deficiency {(NM_016327.3) "c.917-1G>A"}
1813 - 1813	UQCRQ:Mitochondrial complex III deficiency, nuclear type 4 {(NM_014402.5) "c.134C>T"}
1814 - 1816	USH1C:Usher syndrome, type 1C {(NM_005709.3) "c.1220delG", "c.238dupC", "c.497-2delA"}
1817 - 1817	USH1G:Usher syndrome, type 1G {(NM_173477) "c.205dup"}
1818 - 1843	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"}
1844 - 1844	USMG5:Leigh syndrome realted to USMG5 {(NM_032747) "c.87+1G>C"}
1845 - 1846	VDR:Rickets, vitamin D-resistant, type IIA {(NM_001017535.1) "c.277+1G>T", "c.885C>A"}
1847 - 1847	VIPAS39:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome 2 {(NM_022067) "c.808C>T"}
1848 - 1848	VPS11:Hypomyelination and developmental delay {(NM_021729.5) "c.2536T>G"}
1849 - 1852	VPS13A:Choreoacanthocytosis {(NM_033305.3) "c.2343delA", "c.6059delC", "c.9446_9449dup", "delexon70-73"}
1853 - 1854	VPS13B:Cohen syndrome {(NM_017890) "c.4894C>T" | (NM_017890.4) "c.6732+1G>A"}
1855 - 1856	VPS33B:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome {(NM_018668.4) "c.403+1G>A", "c.700G>C"}
1857 - 1857	VPS37A:Spastic paraplegia 53, Autosomal Recessive {(NM_152415.3) "c.1146A>T"}
1858 - 1858	VPS45:Neutropenia, severe congenital, 5, Autosomal Recessive {(NM_007259.5) "c.671C>A"}

1859 - 1860	VPS53:Pontocerebellar hypoplasia, type 2E (PCCA2) {(NM_001128159.3) "c.1556+5G>A", "c.2084A>G"}
1861 - 1861	VRK1:Pontocerebellar hypoplasia type 1A {(NM_003384.3) "c.1072C>T"}
1862 - 1864	WFS1:Wolfram-like syndrome, Autosomal Dominant {(NM_006005) "c.1230_1233delCTCT", "c.1770_1773delGTCT", "c.2590G>A"}
1865 - 1866	WISP3:Arthropathy, progressive pseudorheumatoid, of childhood {(NM_003880.3) "c.156C>A ", "c.536_537delGT"}
1867 - 1867	XPC:Xeroderma pigmentosum, group C {(NM_004628.4) "c.566_567delAT"}
1868 - 1868	XRCC2:Fanconi Anemia {(NM_005431.1) "c.643C>T"}
1869 - 1869	ZBTB24:Immunodeficiency-centromeric instability-facial anomalies syndrome-2 {(NM_014797.2) "c.501dupA"}
1870 - 1870	ZMPSTE24:Mandibuloacral dysplasia with type B lipodystrophy {(NM_005857) "c.1085dupT"}
1871 - 1872	ZNF469:Brittle cornea syndrome 1 {(NM_001127464.2) "c.5943delA", "c.9531delG"}

*** 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.**

**** לפי החלטת איגוד הגנטיקאים הישראלי, המוטציה מדווחת חיובית רק לאנשים ממוצא דרוזי, Belostotsky R, Seboun E, Idelson GH, et al. Mutations in DHDPSL are responsible for primary hyperoxaluria type III. Am J Hum Genet. 2010;87(3):392-399. doi:10.1016/j.ajhg.2010.07.023.**