רשימת מחלות ומוטציות גירסה - Ver 2.0 (2020-08-11)
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 - 12	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"}
13 - 14	ABCB11:Cholestasis, progressive familial intrahepatic type 2 {(NM_003742.4) "c.1409G>A", "c.3268C>T"}
15 - 19	ABCC8:Hyperinsulinemic hypoglycemia, familial, 1 (PHHI/HHF1) {(NM_000352.4) "c.1116dupT", "c.2506C>T", "c.3989-9G>A", "c.4160_4162delTCT", "c.560T>A"}
20 - 20	ABCD1:X-linked adrenoleukodystrophy {(NM_000033.4) "c.686T>C"}
21 - 22	ABHD5:Chanarin-Dorfman syndrome {(NM_016006.6) "c.412T>C", "c.934C>T"}
23 - 25	ACADM:Medium-chain Acyl-CoA dehydrogenase deficiency {(NM_000016.5) "c.616C>T", "c.799G>A" | (NM_001127328) "c.374C>T"}
26 - 34	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"}
35 - 35	ACO2:Infantile cerebellar-retinal degeneration {(NM_001098.3) "c.336C>G"}
36 - 36	ACP5:Spondyloenchondrodysplasia with immune dysregulation {(NM_001111035.2) "c.325G>A"}
37 - 37	ACSF3:Combined malonic and methylmalonic aciduria {(NM_174917) "c.1411C>T"}
38 - 39	ADAMTS2:Ehlers Danlos syndrome, type VIIC {(NM_014244.5) "c.2384G>A", "c.673C>T"}
40 - 41	ADA:Severe combined immunodeficiency due to ADA deficiency {(NM_000022.4) "c.703C>T", "c.792G>A"}
42 - 45	ADGRG1:Bilateral Frontoparietal Polymicrogyria (BFPP) {(NM_005682.7) "c.1046G>C", "c.1167+3G>C", "c.1693C>T", "c.739_745delCAGGACC"}
46 - 47	ADGRV1:Usher syndrome, type 2C {(NM_032119.4) "c.14973-2A>G", "c.15494delA"}
48 - 48	AGA:Aspartylglucosaminuria {(NM_000027.4) "c.214T>C"}
49 - 51	AGL:Glycogen storage disease III {(NM_000642.3) "c.1078C>T", "c.1222C>T", "c.4455delT"}
52 - 64	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"}
65 - 67	AHI1:Joubert syndrome-3 {(NM_017651.4) "c.2212C>T", "c.3032C>G", "c.787dupC"}
68 - 68	AIMP1:Leukodystrophy, hypomyelinating, 3 {(NM_004757.3) "c.292_293delCA"}
69 - 71	AIPL1:Leber congenital amaurosis 4 {(NM_014336.5) "c.211G>T",

	"c.215G>A", "c.834G>A"}
72 - 77	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"}
78 - 78	ALDH1A3:Microphthalmia, isolated 8 {(NM_000693.4) "c.211G>A"}
79 - 79	ALDH7A1:Epilepsy, pyridoxine-dependent {(NM_001182.5) "c.1489+5G>A"}
80 - 82	ALDOB:Fructose intolerance {(NM_000035.4) "c.1005C>G", "c.448G>C", "c.524C>A"}
83 - 84	ALMS1:Alstrom syndrome {(NM_015120.4) "c.8008C>T", "c.808C>T"}
85 - 86	ALPL:Hypophosphatasia, infantile {(NM_000478.6) "c.1348C>T", "c.141C>A"}
87 - 87	AMT:Glycine encephalopathy, AMT-related {(NM_000481.3) "c.125A>G"}
88 - 88	ANO5:Limb-girdle muscular dystrophy {(NM_213599.2) "c.191dupA"}
89 - 89	AP4B1:Spastic paraplegia 47, Autosomal Recessive {(NM_006594.4) "c.664delC"}
90 - 90	APTX:Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia {(NM_175073.2) "c.837G>A"}
91 - 92	AQP2:Diabetes insipidus, nephrogenic {(NM_000486.5) "c.298G>T", "c.83T>C"}
93 - 93	ARFGEF2:Periventricular heterotopia with microcephaly {(NM_006420.3) "c.1958+1G>A"}
94 - 94	ARHGDIA:Nephrotic syndrome, type 8 {(NM_004309.6) "c.518G>T"}
95 - 95	ARL6:Bardet-Biedl syndrome 3 {(NM_032146.5) "c.364C>T"}
96 - 108	ARSA:Metachromatic leukodystrophy - MLD {(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"}
109 - 109	ASL:Argininosuccinic aciduria {(NM_000048.4) "c.346C>T"}
110 - 110	ASNS:Asparagine synthetase deficiency {(NM_183356.3) "c.1084T>G"}
111 - 114	ASPA:Canavan Disease {(NM_001128085.1) "c.433-2A>G", "c.693C>A", "c.854A>C", "c.914C>A"}
115 - 115	ASS1:Citrullinemia,classic {(NM_000050) "c.1168G>A"}
116 - 128	ATM:Ataxia-tyelangiectasia {(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"}
129 - 129	ATP6V0A2:Cutis laxa, Autosomal Recessive, type IIA {(NM_012463.4) "c.2375C>G"}
130 - 150	ATP7B:Wilson disease {(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"}
151 - 151	AVP:Familial neurohypophyseal diabetes insipidus {(NM_000490)

	"c.77C>T"}
152 - 152	B4GALT1:Congenital disorder of glycosylation, type IId {(NM_001497) "c.61C>T"}
153 - 155	BBS10:Bardet-Biedl syndrome 10 {(NM_024685.4) "c.1091delA", "c.1399delA", "c.271dupT"}
156 - 157	BBS1:Bardet-Biedl syndrome 1 {(NM_024649.5) "c.1169T>G", "c.479G>A"}
158 - 162	BBS2:Bardet-Biedl syndrome 2 {(NM_031885.4) "c.1895G>C", "c.224T>G", "c.311A>C", "c.401C>G", "c.98C>A"}
163 - 164	BBS4:Bardet-Biedl syndrome 4 {(NM_033028.5) "c.77-1422_221-753del ", "c.884G>C"}
165 - 165	BBS7:Bardet-Biedl syndrome 7 {(NM_176824.3) "c.1786G>A"}
166 - 172	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"}
173 - 179	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"}
180 - 184	BLM:Bloom syndrome {(NM_000057.4) "c.1984_1985delAA", "c.2207_2212delATCTGAinsTAGATTC", "c.2407dupT", "c.3510T>A", "c.98+1G>T"}
185 - 185	BMPER:Diaphanospondylodysostosis {(NM_133468.5) "c.310C>T"}
186 - 186	BMPR1B:Brachydactyly type A2 {(NM_001256793.2) "c.377G>A"}
187 - 188	BSND:Bartter syndrome, type 4a infantile variant with sensorineuronal deafness {(NM_057176.3) "c.167_168insTTTCCC", "c.28G>A"}
189 - 189	BTD:Biotinidase deficiency {(NM_000060.4) "c.100G>A"}
190 - 191	C12ORF65:Spastic paraplegia 55, Autosomal Recessive {(NM_152269.5) "c.282+2T>A", "c.413_417delAACAA"}
192 - 192	C21orf59:Ciliary dyskinesia, primary, 26 {(NM_021254.4) "c.735C>G"}
193 - 195	C8orf37:Retinitis pigmentosa 64 {(NM_177965.4) "c.497T>A", "c.529C>T", "c.545A>G"}
196 - 196	CAPN3:Muscular dystrophy, limb-girdle, type 2A {(NM_000070) "c.1469G>A"}
197 - 197	CASQ2:Ventricular tachycardia, catecholaminergic polymorphic, 2 {(NM_001232.3) "c.919G>C"}
198 - 201	CBS:Homocystinuria, thrombosis, hyperhomocysteinemic {(NM_000071.2) "c.1006C>T", "c.1224-2A>C", "c.1261delG", "c.785C>G"}
202 - 202	CC2D1A:Mental retardation, Autosomal Recessive 3 - MRT3 {(NM_017721.5) "c.1468+1_1824-1del"}
203 - 203	CCDC174:Birk Volodarsky PMR Synderome Hypotonia and psychomotor developmental delay {(NM_016474.5) "c.1404A>G"}
204 - 204	CCDC65:Ciliary dyskinesia, primary, 27 {(NM_033124.5) "c.877_878delAT"}
205 - 205	CCDC88C:Hydrocephalus, nonsyndromic, Autosomal Recessive {(NM_001080414.4) "c.934C>T"}
206 - 206	CD59:Hemolytic anemia & immune-mediated polyneuropathy, CD59-related {(NM_203330.2) "c.266G>A"}
207 - 207	CDAN1:Dyserythropoietic anemia, congenital, type Ia {(NM_138477.4) "c.3124C>T"}

208 - 208	CDH23:Usher Syndrome Type ID {(NM_022124.6) "c.7903G>T"}
209 - 209	CDK5:Lissencephaly 7 with cerebellar hypoplasia {(NM_004935.4) "c.580+1G>A"}
210 - 210	CECR1:Polyarteritis nodosa, childhood-onset {(NM_017424.2) "c.139G>A"}
211 - 211	CEP104:Joubert syndrome (JBTS) {(NM_014704.4) "c.1328_1329insT"}
212 - 212	CEP152:Microcephaly 9, primary, Autosomal Recessive {(NM_014985.3) "c.2281-2A>G"}
213 - 218	CEP290:Meckel syndrome 4 {(NM_025114.3) "c.1225delA", "c.164_167delCTCA", "c.1666delA", "c.4393C>T", "c.4771C>T", "c.5788A>T"}
219 - 219	CERKL:Retinitis pigmentosa 26 {(NM_001030311.2) "c.238+1G>A"}
220 - 220	CFH:Hemolytic uremic syndrome, complement factor H deficiency {(NM_000186.3) ":c.3677_*4del"}
221 - 465	CFTR:Cystic fibrosis {(NM_000492.3) "c.1000C>T", "c.1001G>A", "c.1007T>A", "c.1013C>T", "c.1021_1022dupTC", "c.1021T>C", "c.1040G>A", "c.1040G>C", "c.1040G>T", "c.1055G>A", "c.1075C>A", "c.[1075C>A;1079C>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.1650delA", "c.1651G>A", "c.165-1G>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.2462_2463delGT", "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.3717+12191C>T", "c.3718-1G>A", "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.4196_4197delTC", "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.543_546delTAGT", "c.54-5940_273+10250del21Kb", "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" | (NM_000492.4) "c.1079C>A"}
466 - 467	CLCN1:Myotonia congenita, Autosomal Recessive {(NM_000083) "c.1444G>A", "c.1586C>T"}
468 - 470	CLCN5:Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis {(NM_000084.4) "c.1399C>T", "c.258delA", "c.82C>T"}
471 - 471	CLCNKB:Bartter syndrome, type 3 and Gitelman syndrome {(NM_000085.4) "c.1313G>A"}
472 - 472	CLN5:Ceroid lipofuscinosis, neuronal, 5 {(NM_006493.3) "c.672delG"}
473 - 473	CLN6:Ceroid lipofuscinosis, neuronal, 6 {(NM_017882.3) "c.214G>T"}
474 - 474	CLN8:Neuronal ceroid lipofuscinosis type 8, including northern epilepsy {(NM_018941.3) "c.766C>G"}
475 - 478	CLRN1:Usher syndrome, type 3A {(NM_174878.2) "c.144T>G", "c.349_358del", "c.433+1G>A", "c.528T>G"}
479 - 484	CNGA3:Achromatopsia-2 - total color blindness {(NM_001298.2) "c.1114C>T", "c.1585G>A", "c.1640T>G", "c.67C>T", "c.940_942delATC", "c.985G>T"}
485 - 485	CNGB1:Retinitis pigmentosa 45 {(NM_001297.5) "c.2284C>T"}
486 - 487	CNGB3:Achromatopsia-3,macular degeneration, juvenile {(NM_019098.4) "c.1148delC", "c.644-1G>C"}
488 - 489	CNNM4:Jalili syndrome {(NM_020184.4) "c.1813C>T", "c.599C>A"}
490 - 490	COL11A2:Otospondylomegaepiphyseal dysplasia (ZW) {(NM_080680.2) "c.3991C>T"}
491 - 494	COL4A3:Alport Syndrome, COL4A3-Related {(NM_000091.4) "c.1791_1793dupTCC", "c.227delG", "c.3518-2A>G", "c.4649T>G"}
495 - 496	COL4A4:Alport syndrome, COL4A4-Related {(NM_000092.4) "c.3933C>G", "c.785_792dupCACCTGAC"}
497 - 501	COL4A5:Alport syndrome, COL4A5-Related {(NM_000495.4) "c.2641G>T", "c.367delG", "c.4691G>C", "c.4946T>G", "c.5030G>A"}
502 - 502	COLEC11:3MC syndrome 2 {(NM_199235.2) "c.627_628delCG"}

503 - 506	COLQ:Myasthenic syndrome, congenital, 5 {(NM_005677) "c.788dupC" | (NM_005677.4) "c.1228C>T", "c.718G>T", "c.792dupG"}
507 - 507	COQ4:Coenzyme Q10 deficiency, primary, 7 {(NM_016035.5) "c.718C>T"}
508 - 513	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"}
514 - 514	CPT1A:Carnitine palmitoyltransferase 1 deficiency {(NM_001031847) "c.1361A>G"}
515 - 515	CPT2:Carnitine palmitoyltransferase II deficiency {(NM_000098) "c.110_111dupGC"}
516 - 530	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"}
531 - 534	CRB2:Ventriculomegaly with cystic kidney disease {(NM_173689.7) "c.1882C>T", "c.1928A>C", "c.2277G>A", "c.2400C>G"}
535 - 535	CRTAP:Osteogenesis imperfecta, type VII {(NM_006371.4) "c.793+1G>T"}
536 - 536	CSTA:Exfoliative ichthyosis, Autosomal Recessive, ichthyosis bullosa of Siemens-like {(NM_005213.4) "c.67-2A>T"}
537 - 538	CTNS:Cystinosis,CTNS-related {(NM_004937.2) "c.1015G>A", "c.530A>C"}
539 - 539	CTSC:Haim-Munk syndrome {(NM_001814.6) "c.857A>G"}
540 - 540	CTSK:Pycnodysostosis {(NM_000396.4) "c.990A>G"}
541 - 544	CYBA:Chronic granulomatous disease,autosomal, due to deficiency of CYBA {(NM_000101.4) "c.164C>G", "c.171dupG", "c.70G>A", "c.71G>A"}
545 - 549	CYBB:Chronic granulomatous disease, X-linked {(NM_000397) "c.1016dupC", "c.1081T>C", "c.271C>T", "c.676C>T", "c.90_92delCCGinsGGT"}
550 - 551	CYP11A1:Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete {(NM_000781.3) "c.644T>C", "c.694C>T"}
552 - 552	CYP11B2:Hypoaldosteronism, congenital, due to CMO II deficiency {(NM_000498.3) "c.541C>T"}
553 - 554	CYP1B1:Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset {(NM_000104.3) "c.1568G>A", "c.182G>A"}
555 - 559	CYP27A1:Cerebrotendinous xanthomatosis {(NM_000784.4) "c.1016C>T", "c.1184G>A", "c.355delC", "c.819delT", "c.845-1G>A"}
560 - 560	CYP7B1:Spastic paraplegia 5A, Autosomal Recessive {(NM_004820.5) "c.1081C>T"}
561 - 561	DAG1:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 {(NM_001165928.3) "c.743delC"}
562 - 562	DARS2:Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation {(NM_018122.5) "c.492+2T>C"}
563 - 563	DBT:Maple syrup urine disease, type II {(NM_001918) "c.581C>G"}
564 - 564	DCAF17:Woodhouse-Sakati syndome {(NM_025000.4) "c.436delC"}
565 - 565	DCLRE1C:Severe combined immunodeficiency, Athabascan type {(NM_001033858.2) "c.1307_1308insAGGATGCT"}
566 - 566	DDR2:Spondylometaepiphyseal dysplasia, short limb-hand type {(NM_006182.4) "c.2254C>T"}

567 - 567	DGAT1:Diarrhea 7, congenital {(NM_012079.6) "c.751+2T>C"}
568 - 568	DGUOK:Mitochondrial DNA depletion syndrome (hepatocerebral type) {(NM_080916.3) "c.255delA"}
569 - 569	DGUOK:Mitochondrial DNA depletion syndrome {(NM_080916.3) "c.271delA"}
570 - 570	DHCR24:Desmosterolosis {(NM_014762.4) "c.307C>T"}
571 - 585	DHCR7:Smith Lemli Opitz syndrome {(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"}
586 - 586	DHDDS:Retinitis pigmentosa 59 {(NM_024887.3) "c.124A>G"}
587 - 590	DLD:Dihydrolipoamide Dehydrogenase Deficiency {(NM_000108.5) "c.104dupA", "c.1123G>A", "c.1436A>T", "c.685G>T"}
591 - 591	DLL3:Spondylocostal dysostosis 1, Autosomal Recessive {(NM_016941.3) "c.395delG"}
592 - 593	DNAH11:Ciliary dyskinesia, primary, 7, with or without situs inversus {(NM_001277115.2) "c.11929G>T", "c.13242_13245delAAAG"}
594 - 594	DNAH5:Ciliary dyskinesia, primary, 3, with or without situs inversus (CILD3/PCD) {(NM_001369.2) "c.8011-2A>G"}
595 - 595	DNAI1:Ciliary dyskinesia, primary, 1, with or without situs inversus {(NM_012144.4) "c.1490G>A"}
596 - 597	DNAI2:Ciliary dyskinesia, primary, 9, with or without situs inversus {(NM_023036.6) "c.1304G>A", "c.1494+1G>A"}
598 - 598	DNAL1:Ciliary dyskinesia, primary, 16 {(NM_031427.4) "c.449A>G"}
599 - 599	DOCK8:Hyper-IgE recurrent infection syndrome, autosomal recessive {(NM_203447) "c.5132C>A"}
600 - 601	DOLK:Congenital disorder of glycosylation, type Im {(NM_014908.3) "c.1222C>G", "c.912G>T"}
602 - 603	DSG1:Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE {(NM_001942.4) "c.1861delG", "c.395C>A"}
604 - 604	DST:Epidermolysis bullosa simplex, Autosomal Recessive 2 {(NM_183380.3) "c.14865delA"}
605 - 610	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"}
611 - 613	ELP1:Dysautonomia, familial {(NM_003640.5) "c.2087G>C", "c.2204+6T>C", "c.2741C>T"}
614 - 614	EOGT:Adams-Oliver syndrome 4 {(NM_001278689.2) "c.1074delA"}
615 - 615	EPCAM:Colorectal cancer, hereditary nonpolyposis, type 1 {(NM_002354) "c.-358_*415del"}
616 - 616	EPG5:Vici syndrome {(NM_020964.3) "c.5704dupT"}
617 - 617	EPM2A:Epilepsy, progressive myoclonic 2A (Lafora) {(NM_005670) "56_kb_incl._ex._2"}
618 - 618	ERBB3:Lethal congenital contractural syndrome 2 {(NM_001982.3) "c.1184-9A>G"}
619 - 619	ERCC2:Xeroderma pigmentosum, group D {(NM_000400.3) "c.2048G>A"}
620 - 620	ERCC5:Xeroderma pigmentosum/Cockayne {(NM_000123.3) "c.205C>T"}

621 - 621	ERCC6:Cockayne syndrome, type B {(NM_000124.4) "c.1034_1035insT"}
622 - 624	ERCC8:Cockayne syndrome, type A {(NM_000082.3) "c.37G>T", "c.843+1G>C", "c.966C>A"}
625 - 625	ESCO2:Roberts-SC phocomelia syndrome {(NM_001017420.3) "c.1674-2A>G"}
626 - 629	ETFDH:Glutaric acidemia IIC {(NM_004453.4) "c.1074G>C", "c.1084G>A", "c.1425C>A", "c.299T>A"}
630 - 630	EXOSC3:Pontocerebellar hypoplasia, type 1B {(NM_016042.4) "c.571G>T"}
631 - 631	EXOSC8:Pontocerebellar hypoplasia, type 1C {(NM_181503.3) "c.5C>T"}
632 - 635	EYS:Retinitis pigmentosa 25 {(NM_001142800.2) "c.1211dupA", "c.3699delG", "c.3715G>T", "c.8155_8156delCA"}
636 - 636	F7:Factor VII deficiency {(NM_000131.4) "c.1256C>T"}
637 - 637	FA2H:Spastic paraplegia 35, Autosomal Recessive {(NM_024306.5) "c.786+1G>A"}
638 - 639	FAH:Tyrosinemia type 1 {(NM_000137.2) "c.1069G>T", "c.192G>T"}
640 - 644	FAH:Tyrosinemia, type I {(NM_000137.2) "c.1062+5G>A", "c.554-1G>T", "c.707-1G>C", "c.782C>T", "c.786G>A"}
645 - 648	FAM161A:Retinitis pigmentosa 28 {(NM_001201543.2) "c.1309A>T", "c.1355_1356delCA", "c.1567C>T" | (NM_032180) "c.1618C>T"}
649 - 649	FAM20A:Amelogenesis imperfecta, type IG (enamel-renal syndrome) {(NM_017565.4) "c.1523delC"}
650 - 657	FANCA:Fanconi anemia, complementation group A {(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"}
658 - 663	FANCC:Fanconi anemia, complementation group C {(NM_000136.3) "c.1642C>T", "c.1661T>C", "c.37C>T", "c.456+4A>T", "c.553C>T", "c.67delG"}
664 - 665	FANCG:Fanconi Anemia - complementation group G {(NM_004629.1) "c.212T>C", "c.510+3A>G"}
666 - 666	FDX1L:Mitochondrial muscle myopathy {(NM_001031734.4) "c.10A>T"}
667 - 669	FERMT1:Kindler syndrome {(NM_017671.4) "c.137_140delTAGT", "c.-1500_-19+470del", "c.749G>A"}
670 - 670	FGB:Afibrinogenemia congenital {(NM_005141.4) "c.1400G>A"}
671 - 672	FGFR3:Achondroplasia {(NM_000142) "c.1138G>A", "c.1138G>C"}
673 - 674	FGFR3:Hypochondroplasia {(NM_000142) "c.1620C>A", "c.1620C>G"}
675 - 675	FH:Fumarase deficiency, leiomyomatosis and renal cell cancer {(NM_000143.3) "c.905-1G>A"}
676 - 676	FKBP10:Osteogenesis imperfecta, type XI {(NM_021939.3) "c.310C>T"}
677 - 677	FKRP:Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 {(NM_024301.5) "c.160C>T"}
678 - 678	FKTN:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 - Walker Warburg syndrome {(NM_001079802.1) "c.1167dupA"}
679 - 679	FLT4:Autosomal Recessive Hereditary Lymphedema {(NM_182925.5) "c.3704C>G"}
680 - 680	FOXRED1:Mitochondrial encephalomyopathy complex I deficiency {(NM_017547.4) "c.1054C>T"}
681 - 681	FRMD4A:Microcephaly intellectual disability and dysmorphism

	{(NM_018027) "c.2134_2146dup13"}
682 - 682	FTO:Growth retardation, developmental delay, coarse facies, and early death {(NM_001080432.3) "c.947G>A"}
683 - 684	G6PC3:Neutropenia, severe congenital 4, Autosomal Recessive {(NM_138387.3) "c.765_766delAG", "c.785G>A"}
685 - 696	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"}
697 - 707	GAA:Pompe (Glycogen storage disease type II) {(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"}
708 - 710	GALC:Krabbe disease {(NM_000153.4) "c.1630G>A", "c.1748A>C", "c.1796T>G"}
711 - 712	GALNT3:Tumoral calcinosis, hyperphosphatemic, familial {(NM_004482.4) "c.1524+1G>A", "c.1524+5G>A"}
713 - 722	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"}
723 - 723	GAN:Giant axonal neuropathy 1 {(NM_022041.3) "c.973G>A"}
724 - 724	GATC:Hypertophic Cardiomyopathy {(NM_176818) "c.233T>G"}
725 - 725	GATM:Cerebral creatine deficiency syndrome 3 {(NM_001482.3) "c.1111dupA"}
726 - 737	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"}
738 - 738	GBE1:Glycogen storage disease IV {(NM_000158) "c.2053-3358_*3188delinsTGTTTTTTACATGACAGGT"}
739 - 750	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"}
751 - 752	GH1:Growth hormone deficiency, isolated, type IA {(NM_000515.5) "c.456+5G>C", "c.67G>T"}
753 - 753	GHRHR:Growth hormone deficiency, isolated, type IB {(NM_000823.4) "c.1069C>T"}
754 - 759	GHR:Laron dwarfism {(NM_000163.5) "c.11G>A", "c.594A>G", "c.62G>A", "c.703C>T", "c.744delT", "del5,6ex"}
760 - 774	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"}
775 - 775	GJB6:Deafness, Autosomal Recessive 1B {(NM_006783.4) "309_kb"}
776 - 781	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"}
782 - 785	GLDC:Glycine encephalopathy and non-ketoic hyperglycinemia, GLDC-related {(NM_000170.2) "c.2405C>T", "c.2607C>A", "c.2T>C", "c.985C>A"}

786 - 786	GLRA1:Hyperekplexia, hereditary 1, autosomal dominant or recessive {(NM_001146040.1) "c.298C>T"}
787 - 787	GMPPA:Alacrima, achalasia, and mental retardation syndrome {(NM_013335.3) "c.1000A>C"}
788 - 789	GMPPB:Muscular dystrophy-dystroglycanopathy {(NM_013334.3) "c.656T>C", "c.860G>A"}
790 - 790	GNE:Hereditary inclusion body myopathy (HIBM) {(NM_005476.6) "c.2135T>C"}
791 - 796	GNPTAB:Mucolipidosis III alpha/beta {(NM_024312.5) "c.118-2A>G", "c.2918dupT", "c.3434+1G>A", "c.3434+715G>A", "c.3503_3504delTC", "c.3613C>T"}
797 - 797	GNPTG:Mucolipidosis III gamma {(NM_032520.5) "c.499dupC"}
798 - 798	GPSM2:Chudley-McCullough syndrome {(NM_013296.5) "c.379C>T"}
799 - 799	GRHPR:Hyperoxaluria, primary, type II {(NM_012203.2) "c.975A>G"}
800 - 807	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"}
808 - 808	HACD1:Congenital myopathy {(NM_014241.4) "c.744C>A"}
809 - 809	HADHA:Long-Chain hydroxylacyl-CoA dehydrogenase deficiency (LCHAD) {(NM_000182.5) "c.1528G>C"}
810 - 818	HBB:Hemoglobipathies (Including sickle-cell anemia and beta thalassemia, Hb C, D, E, O) {(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"}
819 - 839	HEXA:Tay-Sachs disease {(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"}
840 - 840	HEXB:Sandhoff disease, infantile, juvenile, and adult forms {(NM_000521) "c.1082+5G>A"}
841 - 841	HGD:Alkaptonuria {(NM_000187) "c.16-272_87+305del"}
842 - 842	HGSNAT:Retinitis pigmentosa 73 {(NM_152419.3) "c.370A>T"}
843 - 843	HIKESHI:Leukodystrophy, early onset spastic paraparesis,acquired microcephaly, optic atrophy and risk of early death {(NM_016401.4) "c.160G>C"}
844 - 846	HMGCL:HMG-CoA lyase deficiency {(NM_000191.3) "c.122G>A", "c.125A>G", "c.521G>A"}
847 - 847	HOGA1:Hyperoxaluria, primary, type III {(NM_138413) "c.944_946delAGG"}
848 - 850	HPD:Thyrosinemia type III {(NM_002150.3) "c.325-1G>A", "c.415-1G>A", "c.481G>C"}
851 - 851	HPS1:Hermansky-Pudlak syndrome 1 {(NM_000195.5) "c.972delC"}
852 - 855	HPS3:Hermansky-Pudlak syndrome 3 {(NM_032383.5) "c.1163+1G>A", "c.1691+2T>G", "c.2482-2A>G", "c.-2993_217+690del3900"}
856 - 856	HPS6:Hermansky-Pudlak syndrome 6 {(NM_024747.5) "c.1065dupG"}
857 - 857	HSPD1:Leukodystrophy, hypomyelinating, 4, HLD4 (HSP60) {(NM_199440.1) "c.86A>G"}

858 - 858	IBA57:Spastic paraplegia 74, Autosomal Recessive {(NM_001010867.4) "c.678A>G"}
859 - 861	IDUA:Mucopolysaccharidosis Type IH - Hurler syndrome {(NM_000203.5) "c.1096A>C", "c.208C>T", "c.928C>T"}
862 - 863	IGHMBP2:Neuronopathy, distal hereditary motor, type VI {(NM_002180.2) "c.114delA", "c.707T>G"}
864 - 864	IL10RA:Inflammatory bowel disease 28, early onset, autosomal recessive {(NM_001558) "c.537G>A"}
865 - 867	INSR:Leprechaunism, Donohue syndrome {(NM_000208.4) "c.167T>C", "c.3079C>T", "c.857G>A"}
868 - 868	INVS:Nephronophthisis 2, infantile {(NM_014425.5) "c.2719C>T"}
869 - 869	ISPD:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), {(NM_001101426.4) "c.165dupG"}
870 - 872	ITGA2B:Glanzmann thrombasthenia, ITGA2B-related {(NM_000419.4) "c.1947-1G>A", "c.818G>A", "c.97A>G"}
873 - 874	ITGB3:Glanzmann thrombasthenia, ITGB3-related {(NM_000212.2) "11.2kbincl.ex.10-partex.13", "c.428T>G"}
875 - 875	ITGB4:Epidermolysis bullosa, junctional, with pyloric atresia - Carmi syndrome {(NM_000213.5) "c.3224_3793+120del"}
876 - 876	IVD:Isovaleric academia {(NM_002225.5) "c.941C>T"}
877 - 877	KCNJ10:SESAME syndrome {(NM_002241.5) "c.524G>A"}
878 - 878	KLHL40:Nemaline myopathy 8, Autosomal Recessive {(NM_152393.4) "c.581T>A"}
879 - 879	KRT14:Epidermolysis bullosa simplex {(NM_000526) "c.915G>A"}
880 - 884	LAMA2:Muscular dystrophy, congenital, due to partial LAMA2 deficiency {(NM_000426.3) "c.3718C>T", "c.5260delG", "c.828C>G", "c.8665G>A", "c.8689C>T"}
885 - 888	LAMA3:Laryngoonychocutaneous Syndrome {(NM_000227.4) "c.1981C>T", "c.2975delA", "c.4815G>T", "c.893_894insT"}
889 - 898	LAMB3:Epidermolysis bullosa, junctional, non-Herlitz type {(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"}
899 - 900	LAMC2:Epidermolysis bullosa, junctional, Herlitz type {(NM_018891.2) "c.1756C>T", "c.368_373delinsACCAC"}
901 - 905	LCA5:Leber congenital amaurosis 5 {(NM_181714.3) "c.1062_1068delCGAAAAC", "c.1714C>T", "c.238C>T", "c.835C>T", "c.94delT"}
906 - 907	LIFR:Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome-LIFR related {(NM_002310.5) "c.1601-1G>A", "c.2472_2476delTATGT"}
908 - 909	LIPA:Wolman disease {(NM_001127605.2) "c.260G>T", "c.398delC"}
910 - 910	LOXHD1:Deafness, Autosomal Recessive 77 {(NM_144612.6) "c.4714C>T"}
911 - 911	LRBA:Immunodeficiency, common variable, 8, with autoimmunity {(NM_001199282.2) "c.7937T>G"}
912 - 912	MAK:Retinitis pigmentosa 62 {(NM_001242957.2) "c.497G>A"}
913 - 913	MAN1B1:Mental retardation, Autosomal Recessive 15 {(NM_016219.5) "c.1863G>A"}
914 - 914	MATN3:Spondyloepimetaphyseal dysplasia {(NM_002381.5) "c.910T>A"}

915 - 915	MCIDAS:Mucociliary clearance disorder {(NM_001190787.2) "c.1142G>A"}
916 - 919	MCOLN1:Mucolipidosis type IV - ML4 {(NM_020533.3) "c.-1015_788del6433", "c.1207C>T", "c.406-2A>G", "c.964C>T"}
920 - 920	MED17:Microcephaly, postnatal progressive, with seizures and brain atrophy ((ICCA) {(NM_004268.5) "c.1112T>C"}
921 - 921	MED25:Basel-Vanagaite-Smirin-Yosef syndrome {(NM_030973.3) "c.116A>G"}
922 - 922	MEGF10:Myopathy, areflexia, respiratory distress, and dysphagia, early-onset {(NM_001256545.2) "c.1325delC"}
923 - 923	MFSD8:Ceroid lipofuscinosis, neuronal, 7 {(NM_152778.2) "c.472G>A"}
924 - 924	MKS1:Meckel syndrome 1 {(NM_017777.3) "c.1048C>T"}
925 - 927	MLC1:Megalencephalic leukoencephalopathy with subcortical cysts {(NM_015166.3) "c.176G>A", "c.274C>T", "c.278C>T"}
928 - 928	MLPH:Griscelli syndrome, type 3 {(NM_024101.7) "c.103C>T"}
929 - 929	MMACHC:Methylmalonic aciduria and homocystinuria, cblC type {(NM_015506.3) "c.271dupA"}
930 - 931	MOCS1:Molybdenum cofactor deficiency A {(NM_001075098.3) "c.722delT", "c.971G>A"}
932 - 933	MOCS2:Molybdenum cofactor deficiency Type B {(NM_004531.5) "c.226G>A", "c.377+1G>A"}
934 - 936	MPDU1:Congenital disorder of glycosylation, type If {(NM_004870.4) "c.218G>A", "c.2T>C", "c.356T>C"}
937 - 940	MPL:Thrombocytopenia, congenital amegakaryocytic {(NM_005373.2) "c.1031T>A", "c.127C>T", "c.460T>C", "c.79+2T>A"}
941 - 941	MPV17:Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) {(NM_002437.5) "c.278A>C"}
942 - 942	MRE11A:Ataxia Telangiectasia like disorder {(NM_005591.3) "c.290A>G"}
943 - 944	MTHFR:Homocystinuria due to MTHFR deficiency {(NM_005957.4) "c.16delA", "c.474A>T"}
945 - 948	MTTP:Abetalipoproteinemia ABL {(NM_000253.3) "c.2212delT", "c.2593G>T", "c.307A>T", "c.62-2A>G"}
949 - 949	MUT:Methylmalonic acidemia, mut(0) type {(NM_000255.4) "c.655A>T"}
950 - 950	MVK:Hyper-IgD syndrome {(NM_000431.4) "c.1129G>A"}
951 - 952	MYBPC1:Lethal congenital contracture syndrome 4 {(NM_002465.4) "c.556G>A", "c.952C>T"}
953 - 954	MYH2:Proximal myopathy and ophthalmoplegia {(NM_017534.6) "c.2400delG", "c.706G>A"}
955 - 959	MYO15A:Deafness, Autosomal Recessive 3 {(NM_016239.4) "c.373_374delCG", "c.4240G>A", "c.7207G>T", "c.8183G>A", "c.8467G>A"}
960 - 978	MYO7A:Usher syndrome, type 1B {(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"}
979 - 979	NAGLU:Mucopolysaccharidosis type IIIB (Sanfilippo B) {(NM_000263.4) "c.2021G>A"}
980 - 980	NARS2:Combined oxidative phosphorylation deficiency 24 (COXPD24)

	{(NM_024678) "c.500A>G"}
981 - 982	NBEAL2:Gray platelet syndrome {(NM_015175.2) "c.2701C>T", "c.5413dupG"}
983 - 984	NCF1:Chronic granulomatous disease due to deficiency of NCF-1 {(NM_000265.6) "c.153+1G>A", "c.579G>A"}
985 - 987	NCF2:Chronic granulomatous disease due to deficiency of NCF-2 {(NM_000433.3) "c.1171_1175delAAGCT", "c.196C>T", "c.304C>T"}
988 - 988	NDUFA11:Mitochondrial complex I deficiency - NDUFA11 gene {(NM_001193375.1) "c.97+5G>A"}
989 - 989	NDUFAF5:Mitochondrial complex I deficiency - NDUFAF5 gene {(NM_024120.5) "c.749G>T"}
990 - 990	NDUFS2:Mitochondrial complex I deficiency-NDUFS2 gene {(NM_004550.4) "c.1237T>C"}
991 - 991	NDUFS4:Leigh syndrome {(NM_002495.4) "c.462delA"}
992 - 992	NDUFS6:Mitochondrial complex I deficiency - NDUFS6 gene {(NM_004553.4) "c.344G>A"}
993 - 996	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"}
997 - 997	NGLY1:Congenital disorder of deglycosylation {(NM_018297.4) "c.1294G>T"}
998 - 998	NNT:Glucocorticoid deficiency 4 {(NM_182977.3) "c.598G>A"}
999 - 1016	NPC1:Niemann-Pick disease type C1 {(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"}
1017 - 1025	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"}
1026 - 1026	NPHS2:Nephrotic syndrome {(NM_014625.3) "c.412C>T"}
1027 - 1029	NTRK1:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_001012331.1) "c.1842_1843insT", "c.2066C>T", "c.207_208delTG"}
1030 - 1030	NUP62:Striatonigral degeneration, Infantile Bilateral Striatal Necrosis (IBSN) {(NM_016553.4) "c.1172A>C"}
1031 - 1031	OCA2:Albinism, oculocutaneous, type II {(NM_000275.3) "c.1327G>A"}
1032 - 1032	OPA3:3-methylglutaconic aciduria, type III - Costeff {(NM_025136.3) "c.143-1G>C"}
1033 - 1035	OTC:Ornithine transcarbamylase deficiency {(NM_000531.6) "c.717+1G>T", "c.829C>T", "c.958C>T"}
1036 - 1036	OTOA:Deafness, Autosomal Recessive 22 {(NM_144672) "c.1025A>T"}
1037 - 1037	OTOF:Deafness, Autosomal Recessive 9 {(NM_194248.2) "c.2866+1G>A"}
1038 - 1055	PAH:Phenylketonuria {(NM_000277.3) "c.1045T>C", "c.1066-11G>A", "c.1208C>T", "c.1222C>T", "c.1315+1G>A", "c.143T>C", "c.165delT", "c.165T>G", "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.898G>T"}
1056 - 1056	PCCA:Propionic acidemia, PCCA-related {(NM_000282.4) "c.923dupT"}
1057 - 1057	PCCB:Propionic acidemia, PCCB-related {(NM_000532.5) "c.1173dupT"}

1058 - 1059	PCDH12:Microcephaly, seizures, spasticity, and brain calcification (MISSBC) {(NM_016580) "c.2515C>T", "c.995delT"}
1060 - 1060	PCDH15:Usher syndrome, type 1F {(NM_033056.3) "c.733C>T"}
1061 - 1061	PCK1:Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency {(NM_002591.4) "c.134T>C"}
1062 - 1062	PCNT:Microcephalic osteodysplastic primordial dwarfism type II (MOPDII) {(NM_006031.5) "c.3465-1G>A"}
1063 - 1063	PDE6G:Retinitis pigmentosa 57 {(NM_002602.4) "c.187+1G>T"}
1064 - 1066	PEPD:Prolidase deficiency {(NM_000285.4) "c.1103T>G", "c.605C>T", "c.634G>C"}
1067 - 1068	PEX1:Peroxisome biogenesis disorder 1A (Zellweger) {(NM_000466.3) "c.2097dupT", "c.2528G>A"}
1069 - 1071	PEX2:Peroxisome biogenesis disorder 5A (Zellweger) {(NM_001079867.1) "c.355C>T", "c.550delC", "c.669G>A"}
1072 - 1075	PEX6:Peroxisome biogenesis disorder 4B (Zellweger syndrome) {(NM_000287.4) "c.1715C>T", "c.1947delG", "c.2094+2T>C", "c.2534T>C"}
1076 - 1076	PEX7:Rhizomelic chondrodysplasia punctata type 1 {(NM_000288.4) "c.283T>G"}
1077 - 1077	PGAP3:Hyperphosphatasia with mental retardation syndrome 4 {(NM_033419.5) "c.845A>G"}
1078 - 1078	PHGDH:Phosphoglycerate dehydrogenase deficiency {(NM_006623.3) "c.1468G>A"}
1079 - 1079	PHKG2:Glycogen storage disease IXc {(NM_000294.3) "c.71A>G"}
1080 - 1080	PHYH:Refsum disease {(NM_001037537.1) "c.523C>T"}
1081 - 1082	PIGN:Multiple congenital anomalies-hypotonia-seizures syndrome 1 {(NM_012327.5) "c.2126G>A", "c.755A>T"}
1083 - 1084	PIGT:Multiple congenital anomalies-hypotonia-seizures syndrome 3 {(NM_015937.6) "c.1564T>G", "c.761delG"}
1085 - 1085	PIP5K1C:Lethal congenital contractural syndrome 3 {(NM_012398.2) "c.757G>A"}
1086 - 1086	PJVK:Deafness, Autosomal Recessive 59 {(NM_001042702.4) "c.406C>T"}
1087 - 1092	PKHD1:Polycystic kidney & hepatic disease, PKHD1-related {(NM_138694.4) "c.107C>T", "c.1350delC", "c.2279G>A", "c.3761_3762delCCinsG", "c.6122-12G>A", "c.824C>T"}
1093 - 1096	PLA2G6:Infantile neuroaxonal dystrophy 1 (INAD) {(NM_003560.4) "c.1040G>C", "c.2070_2072delTGT", "c.2251G>A", "c.668C>A"}
1097 - 1097	PLAA:Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies {(NM_001031689.3) "c.2254C>T"}
1098 - 1101	PMM2:Congenital disorder of glycosylation Ia {(NM_000303) "c.338C>T", "c.357C>A", "c.422G>A", "c.691G>A"}
1102 - 1102	POC1A:Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis {(NM_015426.5) "c.512T>C"}
1103 - 1103	POR:Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis {(NM_000941.3) "c.1615G>A"}
1104 - 1104	PPIB:Osteogenesis imperfecta, type IX {(NM_000942.4) "c.563_566delACAG"}
1105 - 1105	PPP1R13L:Cardio-Cutaneous Syndrome DCM {(NM_006663.4) "c.2241C>G"}

1106 - 1106	PPT1:Ceroid lipofuscinosis, neuronal, 1 {(NM_000310.3) "c.169dupA"}
1107 - 1107	PRCD:Retinitis pigmentosa 36 {(NM_001077620) "c.64C>T"}
1108 - 1108	PRICKLE1:Epilepsy, progressive myoclonic 1B {(NM_153026.3) "c.311G>A"}
1109 - 1109	PUS1:Mitochondrial myopathy and sideroblastic anemia 1 {(NM_001002020.2) "c.346C>T"}
1110 - 1111	RAG1:Severe combined immudeficiency, B cell-negative, RAG1-related {(NM_000448.2) "c.1361T>A", "c.1410_1413delCTTG"}
1112 - 1116	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"}
1117 - 1120	RAPSN:Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency {(NM_005055.5) "c.-210A>G", "c.264C>A", "c.-27C>G", "c.672_673insACT"}
1121 - 1121	RARS2:Pontocerebellar hypoplasia, type 6 {(NM_020320.5) "c.110+5A>G"}
1122 - 1129	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"}
1130 - 1131	RECQL2:Werner syndrome {(NM_000553.5) "c.1105C>T", "c.2665C>T"}
1132 - 1132	RFX5:Bare lymphocyte syndrome, type II (SCID) {(NM_000449) "c.715C>T"}
1133 - 1133	RIN2:Macrocephaly, alopecia, cutis laxa, and scoliosis {(NM_018993.3) "c.1731delC"}
1134 - 1134	RNASEH2B:Aicardi-Goutieres syndrome 2 {(NM_024570.3) "c.529G>A"}
1135 - 1135	ROGDI:Kohlschutter-Tonz syndrome {(NM_024589.2) "c.469C>T"}
1136 - 1136	RP1:Retinitis pigmentosa 1 {(NM_006269.2) "c.4941dupT"}
1137 - 1142	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"}
1143 - 1143	RPGRIP1L:Meckel syndrome 5 {(NM_015272.5) "c.118C>T"}
1144 - 1144	RSPH9:Ciliary dyskinesia, primary, 12 {(NM_152732.5) "c.804_806delGAA"}
1145 - 1147	RTEL1:Dyskeratosis congenita {(NM_001283009.1) "c.3791G>A" | (NM_032957.4) "c.1548G>T", "c.2992C>T"}
1148 - 1148	RYR1:Minicore myopathy with external ophthalmoplegia {(NM_000540.2) "c.9623C>T"}
1149 - 1150	SAMD9:Tumoral calcinosis, familial, normophosphatemic {(NM_017654.4) "c.1030C>T", "c.4483A>G"}
1151 - 1154	SAMHD1:Aicardi Goutieres syndrome {(NM_015474.3) "9.1-KB_DEL", "c.1106T>C", "c.649_650insG", "c.676C>G"}
1155 - 1155	SARS2:Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis {(NM_017827.3) "c.1169A>G"}
1156 - 1156	SCAPER:Retinitis pigmentosa with intellectual disability {(NM_020843) "c.2806delC"}
1157 - 1157	SCN9A:Insensitivity to pain, congenital (CIPA) {(NM_002977.3) "c.2687G>A"}
1158 - 1158	SCN9A:Insensitivity to pain, congenital {(NM_002977.3) "c.1124delG"}
1159 - 1159	SDHA:Cardiomyopathy, dilated , 1GG neonatal isolated {(NM_004168.4) "c.1664G>A"}
1160 - 1161	SEC23B:Dyserythropoietic anemia, congenital, type II {(NM_006363.6) "c.2129C>T", "c.325G>A"}

1162 - 1163	SepSecS:Pontocerebellar hypoplasia type 2D {(NM_016955.4) "c.1001A>G", "c.715G>A"}
1164 - 1166	SERAC1:3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome {(NM_032861.4) "c.1018delT", "c.128+4A>G", "c.698_699delinsAGTATA"}
1167 - 1167	SGCG:Muscular dystrophy, limb-girdle, type 2C {(NM_000231.2) "c.525delT"}
1168 - 1173	SGSH:Mucopolysaccharidisis type IIIA (Sanfilippo A) {(NM_000199.5) "c.1093C>T", "c.1298G>A", "c.332T>C", "c.416C>T", "c.544C>T", "c.812C>T"}
1174 - 1174	SLC12A3:Bartter and Gitelman syndrome {(NM_000339.3) "c.1313G>A"}
1175 - 1175	SLC17A5:Sialic acid storage disorder, infantile (ISSD) {(NM_012434.5) "c.983G>A"}
1176 - 1177	SLC19A2:Thiamine-responsive megaloblastic anemia syndrome {(NM_006996.3) "c.1223+1G>A", "c.725delC"}
1178 - 1180	SLC1A4:Spastic tetraplegia, thin corpus callosum, and progressive microcephaly {(NM_003038.5) "c.1369C>T", "c.766G>A", "c.944_945del"}
1181 - 1181	SLC22A5:Carnitine deficiency, systemic primary {(NM_003060.3) "c.1196G>A"}
1182 - 1183	SLC25A1:Combined D-2- and L-2-hydroxyglutaric aciduria {(NM_005984.5) "c.389G>A", "c.845G>A"}
1184 - 1185	SLC25A20:Carnitine-acylcarnitine translocase deficiency - CACT {(NM_000387.6) "c.609-3C>G", "c.713A>G"}
1186 - 1186	SLC26A3:Congenital chloride diarhhea (CLD) {(NM_000111.2) "c.559G>T"}
1187 - 1197	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"}
1198 - 1200	SLC29A3:Histiocytosis-lymphadenopathy plus syndrome {(NM_018344.6) "c.1045delC", "c.1279G>A", "c.1309G>A"}
1201 - 1203	SLC2A2:Fanconi-Bickel syndrome {(NM_000340.2) "c.372A>C", "c.734A>C", "c.901C>T"}
1204 - 1204	SLC30A9:Birk-Landau-Perez cerebro-renal syndrome {(NM_016474.5) "c.1047_1049delCAG"}
1205 - 1206	SLC35A3:Arthrogryposis, mental retardation, and seizures {(NM_012243.3) "c.514C>T", "c.886A>G"}
1207 - 1207	SLC35C1:Congenital disorder of glycosylation, type IIc {(NM_018389.4) "c.923C>G"}
1208 - 1211	SLC37A4:Glycogen storage disease Ib {(NM_001164277.1) "c.1042_1043delCT", "c.1179G>A", "c.446G>A", "c.83G>A"}
1212 - 1212	SLC39A4:Acrodermatitis enteropathica {(NM_130849.3) "c.1224delC"}
1213 - 1213	SLC4A4:Renal tubular acidosis (RTA), proximal, with ocular abnormalities and mental retardation {(NM_003759.3) "c.2321G>A"}
1214 - 1214	SLCO2A1:Hypertrophic osteoarthropathy, primary, Autosomal Recessive 2 {(NM_005630.2) "c.1292delC"}
1215 - 1216	SMARCAL1:Schimke immunoosseous dysplasia {(NM_014140.3) "c.2542G>T", "c.863-2A>G"}
1217 - 1217	SMN1:Spinal muscular atrophy-1 {(NM_000344) "c.835_*3del"}
1218 - 1228	SMPD1:Niemann-Pick disease type B, SMPD1-related {(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"}

1229 - 1229	SNAP29:Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome - CEDNIK Syndrome {(NM_004782) "c.223delG"}
1230 - 1230	SNX10:Osteopetrosis, Autosomal Recessive 8 {(NM_001199835.1) "c.152G>A"}
1231 - 1233	SPG11:Spastic paraplegia 11, Autosomal Recessive {(NM_025137.4) "c.118C>T", "c.2471dupT", "c.4339C>T"}
1234 - 1237	SPINK5:Netherton syndrome {(NM_001127698.1) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC"}
1238 - 1238	ST3GAL3:Early infantile epileptic encephalopathy 15 {(NM_006279.4) "c.958G>C"}
1239 - 1239	STRA6:Microphthalmia {(NM_001142617.1) "c.1678G>C"}
1240 - 1241	STRC:Deafness, Autosomal Recessive 16 {(NM_153700.2) "c.4171C>G", "EX7_EX29DEL"}
1242 - 1243	SUMF1:Multiple sulfatase deficiency {(NM_182760.3) "c.1043C>T", "c.463T>C"}
1244 - 1244	SYNE4:Deafness, Autosomal Recessive 76 {(NM_001039876.3) "c.228_229delAT"}
1245 - 1245	SZT2:Epileptic encephalopathy, early infantile, 18 {(NM_015284.3) "c.73C>T"}
1246 - 1246	TAF2:Mental retardation, Autosomal Recessive 40 {(NM_003184.4) "c.557C>G"}
1247 - 1247	TBCD:Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM_005993.4) "c.1423G>A"}
1248 - 1249	TBCE:Hypoparathyroidism retardation dysmorphism syndrome {(NM_003193.5) "c.155_166delGCCACGAAGGGA", "c.354_355del"}
1250 - 1250	TBX19:Adrenocorticotropic hormone deficiency {(NM_005149.3) "c.574_577delATAG"}
1251 - 1254	TCIRG1:Osteopetrosis, Autosomal Recessive 1 {(NM_006019.4) "c.117+4A>T", "c.1331G>T", "c.1384_1386delAAC", "c.674delG"}
1255 - 1255	TCTN2:Meckel syndrome 8 {(NM_024809.5) "c.1506-2A>G"}
1256 - 1258	TECPR2:Spastic paraplegia 49, Autosomal Recessive {(NM_001172631.2) "c.1319delT", "c.3416delT", "c.566C>T"}
1259 - 1259	THG1L:Cerebellar ataxia and developmental delay {(NM_017872.5) "c.164T>C"}
1260 - 1262	TK2:Mitochondrial DNA depletion syndrome 2 (myopathic type) {(NM_004614.5) "c.360_361delGCinsAA", "c.361C>A", "c.635T>A"}
1263 - 1267	TMC1:Deafness, Autosomal Recessive 7 {(NM_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"}
1268 - 1270	TMEM216:Joubert syndrome 2 (MKS2) {(NM_001173990.3) "c.218G>A", "c.218G>T", "c.230G>C"}
1271 - 1271	TMEM231:Meckel syndrome 11 {(NM_001077418.3) "c.664+4A>G"}
1272 - 1272	TMEM38B:Osteogenesis imperfecta, type XIV {(NM_018112) "c.455_542del"}
1273 - 1275	TMEM67:Joubert syndrome type 6 (MSK3) {(NM_153704) "c.1065+1delG" | (NM_153704.5) "c.1975C>T", "c.725A>G"}
1276 - 1277	TMEM70:ATPase deficiency, nuclear encoded {(NM_017866.6) "c.238C>T", "c.316+1G>T"}
1278 - 1278	TMPRSS3:Deafness, Autosomal Recessive 8/10 {(NM_024022.2)

	"c.989delA"}
1279 - 1279	TRAPPC9:Mental retardation, Autosomal Recessive 13 {(NM_031466.7) "c.1423C>T"}
1280 - 1280	TRIM32:Bardet-Biedl syndrome 11 {(NM_012210) "c.388C>T"}
1281 - 1282	TRMT10A:Microcephaly, short stature, and impaired glucose metabolism {(NM_152292.4) "c.616G>A", "c.727C>T"}
1283 - 1283	TRMU:LIFT, Liver failure infantile transient {(NM_018006.5) "c.229T>C"}
1284 - 1284	TRPM1:Night blindness, congenital stationary (complete), 1C, Autosomal Recessive {(NM_002420.5) "36.4-KB_DEL,_EX2-7"}
1285 - 1286	TRPM6:Hypomagnesemia 1, intestinal {(NM_017662.5) "c.1010+5G>C", "c.2009+1G>A"}
1287 - 1287	TSHR:Hypothyroidism, congenital, nongoitrous, 1 {(NM_000369.2) "c.1825C>T"}
1288 - 1289	TTN:Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis {(NM_003319.4) "c.58881dupA" | (NM_133432) "c.36122delC"}
1290 - 1291	TULP1:Retinitis pigmentosa 14 {(NM_003322.6) "c.1349G>A", "c.1495+2dupT"}
1292 - 1293	TYMP:Mitochondrial DNA depletion syndrome 1 (MNGIE type) {(NM_001113755.2) "c.433G>A", "c.866A>C"}
1294 - 1309	TYR:Albinism, oculocutaneous, type IA (OCA1A) {(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"}
1310 - 1310	UNC13D:Hemophagocytic lymphohistiocytosis, familial, 3 {(NM_199242.2) "c.753+1G>T"}
1311 - 1311	UNC80:Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM_032504.1) "c.151C>T"}
1312 - 1312	UQCRQ:Mitochondrial complex III deficiency, nuclear type 4 {(NM_014402.5) "c.134C>T"}
1313 - 1315	USH1C:Usher syndrome, type 1C {(NM_005709.3) "c.1220delG", "c.238dupC", "c.497-2delA"}
1316 - 1339	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.5519G>T", "c.6159delA", "c.6937G>T", "c.802G>A", "c.8558+1G>T", "c.8719A>C", "c.9685delG"}
1340 - 1341	VDR:Rickets, vitamin D-resistant, type IIA {(NM_001017535.1) "c.277+1G>T", "c.885C>A"}
1342 - 1342	VPS11:Hypomyelination and developmental delay {(NM_021729.5) "c.2536T>G"}
1343 - 1344	VPS13A:Choreoacanthocytosis {(NM_033305.3) "c.6059delC", "delexon70-73"}
1345 - 1345	VPS13B:Cohen syndrome {(NM_017890.4) "c.6732+1G>A"}
1346 - 1347	VPS33B:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome {(NM_018668.4) "c.403+1G>A", "c.700G>C"}
1348 - 1348	VPS37A:Spastic paraplegia 53, Autosomal Recessive {(NM_152415.3) "c.1146A>T"}

1349 - 1349	VPS45:Neutropenia, severe congenital, 5, Autosomal Recessive {(NM_007259.5) "c.671C>A"}
1350 - 1351	VPS53:Pontocerebellar hypoplasia, type 2E (PCCA2) {(NM_001128159.3) "c.1556+5G>A", "c.2084A>G"}
1352 - 1352	VRK1:Pontocerebellar hypoplasia type 1A {(NM_003384.3) "c.1072C>T"}
1353 - 1353	WISP3:Arthropathy, progressive pseudorheumatoid, of childhood {(NM_003880.3) "c.536_537delGT"}
1354 - 1354	XPC:Xeroderma pigmentosum, group C {(NM_004628.4) "c.566_567delAT"}
1355 - 1355	ZBTB24:Immunodeficiency-centromeric instability-facial anomalies syndrome-2 {(NM_014797.2) "c.501dupA"}
1356 - 1357	ZNF469:Brittle cornea syndrome 1 {(NM_001127464.2) "c.5943delA", "c.9531delG"}