**(2020-11-21) Ver 2 - Clalit רשימת מחלות ומוטציות בפאנל**  
**Hybrid Capture-Based Next Generation Sequencing**

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| 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.-1500\_-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 - 1697 | 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"} |
| 1698 - 1698 | SNAP29:Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome - CEDNIK Syndrome {(NM\_004782) "c.223delG"} |
| 1699 - 1699 | SNX10:Osteopetrosis, Autosomal Recessive 8 {(NM\_001199835.1) "c.152G>A"} |
| 1700 - 1703 | SPG11:Spastic paraplegia 11, Autosomal Recessive {(NM\_025137) "c.5986dupT" | (NM\_025137.4) "c.118C>T", "c.2471dupT", "c.4339C>T"} |
| 1704 - 1708 | SPINK5:Netherton syndrome {(NM\_001127698.1) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC", "c.995delT"} |
| 1709 - 1709 | ST3GAL3:Early infantile epileptic encephalopathy 15 {(NM\_006279.4) "c.958G>C"} |
| 1710 - 1710 | STRA6:Microphthalmia {(NM\_001142617.1) "c.1678G>C"} |
| 1711 - 1712 | STRC:Deafness, Autosomal Recessive 16 {(NM\_153700.2) "EX7\_EX29DEL", "c.4171C>G"} |
| 1713 - 1713 | SUCLA2:Mitochondrial DNA depletion syndrome 5 {(NM\_003850) "c.788\_802+29del"} |
| 1714 - 1715 | SUMF1:Multiple sulfatase deficiency {(NM\_182760.3) "c.1043C>T", "c.463T>C"} |
| 1716 - 1717 | SURF1:Leigh syndrome, due to COX deficiency {(NM\_003172) "c.312\_321delTCTGCCAGCCinsAT", "c.575\_576insTGCG"} |
| 1718 - 1718 | SYNE4:Deafness, Autosomal Recessive 76 {(NM\_001039876.3) "c.228\_229delAT"} |
| 1719 - 1719 | SZT2:Epileptic encephalopathy, early infantile, 18 {(NM\_015284.3) "c.73C>T"} |
| 1720 - 1721 | SepSecS:Pontocerebellar hypoplasia type 2D {(NM\_016955.4) "c.1001A>G", "c.715G>A"} |
| 1722 - 1722 | TAF2:Mental retardation, Autosomal Recessive 40 {(NM\_003184.4) "c.557C>G"} |
| 1723 - 1723 | TBCD:Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM\_005993.4) "c.1423G>A"} |
| 1724 - 1725 | TBCE:Hypoparathyroidism retardation dysmorphism syndrome {(NM\_003193.5) "c.155\_166delGCCACGAAGGGA", "c.355\_356del"} |
| 1726 - 1726 | TBX19:Adrenocorticotropic hormone deficiency {(NM\_005149.3) "c.574\_577delATAG"} |
| 1727 - 1730 | TCIRG1:Osteopetrosis, Autosomal Recessive 1 {(NM\_006019.4) "c.117+4A>T", "c.1331G>T", "c.1384\_1386delAAC", "c.674delG"} |
| 1731 - 1731 | TCTN2:Meckel syndrome 8 {(NM\_024809.5) "c.1506-2A>G"} |
| 1732 - 1734 | TECPR2:Spastic paraplegia 49, Autosomal Recessive {(NM\_001172631.2) "c.1319delT", "c.3416delT", "c.566C>T"} |
| 1735 - 1735 | TGM1:Ichthyosis, congenital, Autosomal Recessive 1 {(NM\_000359) "c.2290C>T"} |
| 1736 - 1736 | THG1L:Cerebellar ataxia and developmental delay {(NM\_017872.5) "c.164T>C"} |
| 1737 - 1737 | TIMM50:3-methylglutaconic aciduria, type IX {(ENST00000314349.4) "c.649C>T"} |
| 1738 - 1740 | TK2:Mitochondrial DNA depletion syndrome 2 (myopathic type) {(NM\_004614.5) "c.360\_361delGCinsAA", "c.361C>A", "c.635T>A"} |
| 1741 - 1741 | TKT:Short stature, developmental delay, and congenital heart defects {(NM\_001135055.2) "c.769\_770insCTACCTCCTTATCTTCTG"} |
| 1742 - 1746 | TMC1:Deafness, Autosomal Recessive 7 {(NM\_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"} |
| 1747 - 1747 | TMEM165:Congenital disorder of glycosylation {(NM\_018475.4) "c.792+182G>A"} |
| 1748 - 1750 | TMEM216:Joubert syndrome 2 (MKS2) {(NM\_001173990.3) "c.218G>A", "c.218G>T", "c.230G>C"} |
| 1751 - 1751 | TMEM231:Meckel syndrome 11 {(NM\_001077418.3) "c.664+4A>G"} |
| 1752 - 1752 | TMEM260:Neurodevelopmental, Cardiac, and Renal Syndrome {(NM\_017799.3) "c.1393C>T"} |
| 1753 - 1754 | TMEM38B:Osteogenesis imperfecta, type XIV {(NM\_018112) "c.455\_542del", "c.507G>A"} |
| 1755 - 1757 | TMEM67:Joubert syndrome type 6 (MSK3) {(NM\_153704) "c.1065+1delG" | (NM\_153704.5) "c.1975C>T", "c.725A>G"} |
| 1758 - 1760 | TMEM70:ATPase deficiency, nuclear encoded {(NM\_017866) "c.336T>A" | (NM\_017866.6) "c.238C>T", "c.316+1G>T"} |
| 1761 - 1762 | TMPRSS3:Deafness, Autosomal Recessive 8/10 {(NM\_024022) "c.1177\_1184delins" | (NM\_024022.2) "c.989delA"} |
| 1763 - 1763 | TNNT1:Nemaline myopathy 5, Amish type {(NM\_003283) "c.574\_577delinsTAGTGCTGT"} |
| 1764 - 1764 | TPP1:Ceroid lipofuscinosis, neuronal, 2 {(NM\_000391) "c.775delC"} |
| 1765 - 1765 | TRAK1:Encephalopathy, fatal {(NM\_001042646.2) "c.287-2A>C"} |
| 1766 - 1766 | TRAPPC9:Mental retardation, Autosomal Recessive 13 {(NM\_031466.7) "c.1423C>T"} |
| 1767 - 1767 | TRIM32:Bardet-Biedl syndrome 11 {(NM\_012210) "c.388C>T"} |
| 1768 - 1768 | TRIOBP:Deafness, Autosomal Recessive 28 {(NM\_001039141) "c.1741C>T"} |
| 1769 - 1770 | TRMT10A:Microcephaly, short stature, and impaired glucose metabolism {(NM\_152292.4) "c.616G>A", "c.727C>T"} |
| 1771 - 1773 | TRMU:LIFT, Liver failure infantile transient {(NM\_018006) "c.500\_509del10", "c.835G>A" | (NM\_018006.5) "c.229T>C"} |
| 1774 - 1777 | 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"} |
| 1778 - 1779 | TRPM6:Hypomagnesemia 1, intestinal {(NM\_017662.5) "c.1010+5G>C", "c.2009+1G>A"} |
| 1780 - 1781 | TSHR:Hypothyroidism, congenital, nongoitrous, 1 {(NM\_000369) "c.202C>T" | (NM\_000369.2) "c.1825C>T"} |
| 1782 - 1782 | TSPAN12:Vitroretinal vascular malformations, congenital {(NM\_012338) "c.542G>T"} |
| 1783 - 1784 | TTN:Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis {(NM\_003319.4) "c.58881dupA" | (NM\_133432) "c.36122delC"} |
| 1785 - 1785 | TUBGCP4:Microcephaly, primary, Autosomal Recessive {(NM\_014444.5) "c.579dupT"} |
| 1786 - 1788 | TULP1:Retinitis pigmentosa 14 {(NM\_003322) "c.849\_852dup" | (NM\_003322.6) "c.1349G>A", "c.1495+2dupT"} |
| 1789 - 1790 | TYMP:Mitochondrial DNA depletion syndrome 1 (MNGIE type) {(NM\_001113755.2) "c.433G>A", "c.866A>C"} |
| 1791 - 1807 | 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"} |
| 1808 - 1808 | UNC13D:Hemophagocytic lymphohistiocytosis, familial, 3 {(NM\_199242.2) "c.753+1G>T"} |
| 1809 - 1810 | UNC80:Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM\_032504) "c.7183C>T" | (NM\_032504.1) "c.151C>T"} |
| 1811 - 1811 | UPB1:Beta-ureidopropionase deficiency {(NM\_016327.3) "c.917-1G>A"} |
| 1812 - 1812 | UQCRQ:Mitochondrial complex III deficiency, nuclear type 4 {(NM\_014402.5) "c.134C>T"} |
| 1813 - 1815 | USH1C:Usher syndrome, type 1C {(NM\_005709.3) "c.1220delG", "c.238dupC", "c.497-2delA"} |
| 1816 - 1816 | USH1G:Usher syndrome, type 1G {(NM\_173477) "c.205dup"} |
| 1817 - 1842 | 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"} |
| 1843 - 1843 | USMG5:Leigh syndrome realted to USMG5 {(NM\_032747) "c.87+1G>C"} |
| 1844 - 1845 | VDR:Rickets, vitamin D-resistant, type IIA {(NM\_001017535.1) "c.277+1G>T", "c.885C>A"} |
| 1846 - 1846 | VIPAS39:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome 2 {(NM\_022067) "c.808C>T"} |
| 1847 - 1847 | VPS11:Hypomyelination and developmental delay {(NM\_021729.5) "c.2536T>G"} |
| 1848 - 1851 | VPS13A:Choreoacanthocytosis {(NM\_033305.3) "c.2343delA", "c.6059delC", "c.9446\_9449dup", "delexon70-73"} |
| 1852 - 1853 | VPS13B:Cohen syndrome {(NM\_017890) "c.4894C>T" | (NM\_017890.4) "c.6732+1G>A"} |
| 1854 - 1855 | VPS33B:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome {(NM\_018668.4) "c.403+1G>A", "c.700G>C"} |
| 1856 - 1856 | VPS37A:Spastic paraplegia 53, Autosomal Recessive {(NM\_152415.3) "c.1146A>T"} |
| 1857 - 1857 | VPS45:Neutropenia, severe congenital, 5, Autosomal Recessive {(NM\_007259.5) "c.671C>A"} |
| 1858 - 1859 | VPS53:Pontocerebellar hypoplasia, type 2E (PCCA2) {(NM\_001128159.3) "c.1556+5G>A", "c.2084A>G"} |
| 1860 - 1860 | VRK1:Pontocerebellar hypoplasia type 1A {(NM\_003384.3) "c.1072C>T"} |
| 1861 - 1863 | WFS1:Wolfram-like syndrome, Autosomal Dominant {(NM\_006005) "c.1230\_1233delCTCT", "c.1770\_1773delGTCT", "c.2590G>A"} |
| 1864 - 1865 | WISP3:Arthropathy, progressive pseudorheumatoid, of childhood {(NM\_003880.3) "c.156C>A ", "c.536\_537delGT"} |
| 1866 - 1866 | XPC:Xeroderma pigmentosum, group C {(NM\_004628.4) "c.566\_567delAT"} |
| 1867 - 1867 | XRCC2:Fanconi Anemia {(NM\_005431.1) "c.643C>T"} |
| 1868 - 1868 | ZBTB24:Immunodeficiency-centromeric instability-facial anomalies syndrome-2 {(NM\_014797.2) "c.501dupA"} |
| 1869 - 1869 | ZMPSTE24:Mandibuloacral dysplasia with type B lipodystrophy {(NM\_005857) "c.1085dupT"} |
| 1870 - 1871 | 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.**