**(2020-11-08) Ver 2 - רשימת מחלות ומוטציות גירסה**  
**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 - 1685 | SMN1: Spinal muscular atrophy-1 {(NM\_000344) "c.835\_\*3del"} |
| 1686 - 1698 | SMPD1: Niemann-Pick disease type B, SMPD1-related {(NM\_000543) "c.1092-1G>C", "c.880C>A" | (NM\_000543.5) "c.1493G>T", "c.1705T>C", "c.1758T>G", "c.1805G>C", "c.1828\_1830delCGC", "c.442T>A", "c.573delT", "c.730G>A", "c.911T>C", "c.96G>A", "c.996delC"} |
| 1699 - 1699 | SNAP29: Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome - CEDNIK Syndrome {(NM\_004782) "c.223delG"} |
| 1700 - 1700 | SNX10: Osteopetrosis, Autosomal Recessive 8 {(NM\_001199835.1) "c.152G>A"} |
| 1701 - 1704 | SPG11: Spastic paraplegia 11, Autosomal Recessive {(NM\_025137) "c.5986dupT" | (NM\_025137.4) "c.118C>T", "c.2471dupT", "c.4339C>T"} |
| 1705 - 1709 | SPINK5: Netherton syndrome {(NM\_001127698.1) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC", "c.995delT"} |
| 1710 - 1710 | ST3GAL3: Early infantile epileptic encephalopathy 15 {(NM\_006279.4) "c.958G>C"} |
| 1711 - 1711 | STRA6: Microphthalmia {(NM\_001142617.1) "c.1678G>C"} |
| 1712 - 1713 | STRC: Deafness, Autosomal Recessive 16 {(NM\_153700.2) "EX7\_EX29DEL", "c.4171C>G"} |
| 1714 - 1714 | SUCLA2: Mitochondrial DNA depletion syndrome 5 {(NM\_003850) "c.788\_802+29del"} |
| 1715 - 1716 | SUMF1: Multiple sulfatase deficiency {(NM\_182760.3) "c.1043C>T", "c.463T>C"} |
| 1717 - 1718 | SURF1: Leigh syndrome, due to COX deficiency {(NM\_003172) "c.312\_321delTCTGCCAGCCinsAT", "c.575\_576insTGCG"} |
| 1719 - 1719 | SYNE4: Deafness, Autosomal Recessive 76 {(NM\_001039876.3) "c.228\_229delAT"} |
| 1720 - 1720 | SZT2: Epileptic encephalopathy, early infantile, 18 {(NM\_015284.3) "c.73C>T"} |
| 1721 - 1722 | SepSecS: Pontocerebellar hypoplasia type 2D {(NM\_016955.4) "c.1001A>G", "c.715G>A"} |
| 1723 - 1723 | TAF2: Mental retardation, Autosomal Recessive 40 {(NM\_003184.4) "c.557C>G"} |
| 1724 - 1724 | TBCD: Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM\_005993.4) "c.1423G>A"} |
| 1725 - 1726 | TBCE: Hypoparathyroidism retardation dysmorphism syndrome {(NM\_003193.5) "c.155\_166delGCCACGAAGGGA", "c.355\_356del"} |
| 1727 - 1727 | TBX19: Adrenocorticotropic hormone deficiency {(NM\_005149.3) "c.574\_577delATAG"} |
| 1728 - 1731 | TCIRG1: Osteopetrosis, Autosomal Recessive 1 {(NM\_006019.4) "c.117+4A>T", "c.1331G>T", "c.1384\_1386delAAC", "c.674delG"} |
| 1732 - 1732 | TCTN2: Meckel syndrome 8 {(NM\_024809.5) "c.1506-2A>G"} |
| 1733 - 1735 | TECPR2: Spastic paraplegia 49, Autosomal Recessive {(NM\_001172631.2) "c.1319delT", "c.3416delT", "c.566C>T"} |
| 1736 - 1736 | TGM1: Ichthyosis, congenital, Autosomal Recessive 1 {(NM\_000359) "c.2290C>T"} |
| 1737 - 1737 | THG1L: Cerebellar ataxia and developmental delay {(NM\_017872.5) "c.164T>C"} |
| 1738 - 1738 | TIMM50: 3-methylglutaconic aciduria, type IX {(ENST00000314349.4) "c.649C>T"} |
| 1739 - 1741 | TK2: Mitochondrial DNA depletion syndrome 2 (myopathic type) {(NM\_004614.5) "c.360\_361delGCinsAA", "c.361C>A", "c.635T>A"} |
| 1742 - 1742 | TKT: Short stature, developmental delay, and congenital heart defects {(NM\_001135055.2) "c.769\_770insCTACCTCCTTATCTTCTG"} |
| 1743 - 1747 | TMC1: Deafness, Autosomal Recessive 7 {(NM\_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"} |
| 1748 - 1748 | TMEM165: Congenital disorder of glycosylation {(NM\_018475.4) "c.792+182G>A"} |
| 1749 - 1751 | TMEM216: Joubert syndrome 2 (MKS2) {(NM\_001173990.3) "c.218G>A", "c.218G>T", "c.230G>C"} |
| 1752 - 1752 | TMEM231: Meckel syndrome 11 {(NM\_001077418.3) "c.664+4A>G"} |
| 1753 - 1753 | TMEM260: Neurodevelopmental, Cardiac, and Renal Syndrome {(NM\_017799.3) "c.1393C>T"} |
| 1754 - 1755 | TMEM38B: Osteogenesis imperfecta, type XIV {(NM\_018112) "c.455\_542del", "c.507G>A"} |
| 1756 - 1758 | TMEM67: Joubert syndrome type 6 (MSK3) {(NM\_153704) "c.1065+1delG" | (NM\_153704.5) "c.1975C>T", "c.725A>G"} |
| 1759 - 1761 | TMEM70: ATPase deficiency, nuclear encoded {(NM\_017866) "c.336T>A" | (NM\_017866.6) "c.238C>T", "c.316+1G>T"} |
| 1762 - 1763 | TMPRSS3: Deafness, Autosomal Recessive 8/10 {(NM\_024022) "c.1177\_1184delins" | (NM\_024022.2) "c.989delA"} |
| 1764 - 1764 | TNNT1: Nemaline myopathy 5, Amish type {(NM\_003283) "c.574\_577delinsTAGTGCTGT"} |
| 1765 - 1765 | TPP1: Ceroid lipofuscinosis, neuronal, 2 {(NM\_000391) "c.775delC"} |
| 1766 - 1766 | TRAK1: Encephalopathy, fatal {(NM\_001042646.2) "c.287-2A>C"} |
| 1767 - 1767 | TRAPPC9: Mental retardation, Autosomal Recessive 13 {(NM\_031466.7) "c.1423C>T"} |
| 1768 - 1768 | TRIM32: Bardet-Biedl syndrome 11 {(NM\_012210) "c.388C>T"} |
| 1769 - 1769 | TRIOBP: Deafness, Autosomal Recessive 28 {(NM\_001039141) "c.1741C>T"} |
| 1770 - 1771 | TRMT10A: Microcephaly, short stature, and impaired glucose metabolism {(NM\_152292.4) "c.616G>A", "c.727C>T"} |
| 1772 - 1774 | TRMU: LIFT, Liver failure infantile transient {(NM\_018006) "c.500\_509del10", "c.835G>A" | (NM\_018006.5) "c.229T>C"} |
| 1775 - 1778 | TRPM1: Night blindness, congenital stationary (complete), 1C, Autosomal Recessive {(NM\_002420.5) "36.4-KB\_DEL,\_EX2-7", "c.2567G>A", "c.2629C>T", "c.880A>T"} |
| 1779 - 1780 | TRPM6: Hypomagnesemia 1, intestinal {(NM\_017662.5) "c.1010+5G>C", "c.2009+1G>A"} |
| 1781 - 1782 | TSHR: Hypothyroidism, congenital, nongoitrous, 1 {(NM\_000369) "c.202C>T" | (NM\_000369.2) "c.1825C>T"} |
| 1783 - 1783 | TSPAN12: Vitroretinal vascular malformations, congenital {(NM\_012338) "c.542G>T"} |
| 1784 - 1785 | TTN: Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis {(NM\_003319.4) "c.58881dupA" | (NM\_133432) "c.36122delC"} |
| 1786 - 1786 | TUBGCP4: Microcephaly, primary, Autosomal Recessive {(NM\_014444.5) "c.579dupT"} |
| 1787 - 1789 | TULP1: Retinitis pigmentosa 14 {(NM\_003322) "c.849\_852dup" | (NM\_003322.6) "c.1349G>A", "c.1495+2dupT"} |
| 1790 - 1791 | TYMP: Mitochondrial DNA depletion syndrome 1 (MNGIE type) {(NM\_001113755.2) "c.433G>A", "c.866A>C"} |
| 1792 - 1808 | TYR: Albinism, oculocutaneous, type IA (OCA1A) {(NM\_000372) "c.1A>G" | (NM\_000372.5) "c.1037-1G>A", "c.1037-7T>A", "c.1118C>A", "c.1204C>T", "c.1217C>T", "c.1357C>T", "c.140G>A", "c.149C>G", "c.454C>T", "c.649C>T", "c.649delC", "c.74dupT", "c.757G>A", "c.832C>T", "c.880G>A", "c.896G>A"} |
| 1809 - 1809 | UNC13D: Hemophagocytic lymphohistiocytosis, familial, 3 {(NM\_199242.2) "c.753+1G>T"} |
| 1810 - 1811 | UNC80: Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM\_032504) "c.7183C>T" | (NM\_032504.1) "c.151C>T"} |
| 1812 - 1812 | UPB1: Beta-ureidopropionase deficiency {(NM\_016327.3) "c.917-1G>A"} |
| 1813 - 1813 | UQCRQ: Mitochondrial complex III deficiency, nuclear type 4 {(NM\_014402.5) "c.134C>T"} |
| 1814 - 1816 | USH1C: Usher syndrome, type 1C {(NM\_005709.3) "c.1220delG", "c.238dupC", "c.497-2delA"} |
| 1817 - 1817 | USH1G: Usher syndrome, type 1G {(NM\_173477) "c.205dup"} |
| 1818 - 1843 | USH2A: Usher syndrome, type 2A {(NM\_206933.3) "c.1000C>T", "c.10211delC", "c.12052G>A", "c.12067-2A>G", "c.12575G>A", "c.13274C>T", "c.14020A>G", "c.14023A>T", "c.14413G>A", "c.14424C>A", "c.2167+5G>A", "c.2209C>T", "c.236\_239dupGTAC", "c.3368A>G", "c.377delG", "c.3959C>T", "c.4544C>T", "c.5078G>A", "c.5519G>T", "c.5776+1G>A", "c.6159delA", "c.6937G>T", "c.802G>A", "c.8558+1G>T", "c.8719A>C", "c.9685delG"} |
| 1844 - 1844 | USMG5: Leigh syndrome realted to USMG5 {(NM\_032747) "c.87+1G>C"} |
| 1845 - 1846 | VDR: Rickets, vitamin D-resistant, type IIA {(NM\_001017535.1) "c.277+1G>T", "c.885C>A"} |
| 1847 - 1847 | VIPAS39: Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome 2 {(NM\_022067) "c.808C>T"} |
| 1848 - 1848 | VPS11: Hypomyelination and developmental delay {(NM\_021729.5) "c.2536T>G"} |
| 1849 - 1852 | VPS13A: Choreoacanthocytosis {(NM\_033305.3) "c.2343delA", "c.6059delC", "c.9446\_9449dup", "delexon70-73"} |
| 1853 - 1854 | VPS13B: Cohen syndrome {(NM\_017890) "c.4894C>T" | (NM\_017890.4) "c.6732+1G>A"} |
| 1855 - 1856 | VPS33B: Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome {(NM\_018668.4) "c.403+1G>A", "c.700G>C"} |
| 1857 - 1857 | VPS37A: Spastic paraplegia 53, Autosomal Recessive {(NM\_152415.3) "c.1146A>T"} |
| 1858 - 1858 | VPS45: Neutropenia, severe congenital, 5, Autosomal Recessive {(NM\_007259.5) "c.671C>A"} |
| 1859 - 1860 | VPS53: Pontocerebellar hypoplasia, type 2E (PCCA2) {(NM\_001128159.3) "c.1556+5G>A", "c.2084A>G"} |
| 1861 - 1861 | VRK1: Pontocerebellar hypoplasia type 1A {(NM\_003384.3) "c.1072C>T"} |
| 1862 - 1864 | WFS1: Wolfram-like syndrome, Autosomal Dominant {(NM\_006005) "c.1230\_1233delCTCT", "c.1770\_1773delGTCT", "c.2590G>A"} |
| 1865 - 1866 | WISP3: Arthropathy, progressive pseudorheumatoid, of childhood {(NM\_003880.3) "c.156C>A ", "c.536\_537delGT"} |
| 1867 - 1867 | XPC: Xeroderma pigmentosum, group C {(NM\_004628.4) "c.566\_567delAT"} |
| 1868 - 1868 | XRCC2: Fanconi Anemia {(NM\_005431.1) "c.643C>T"} |
| 1869 - 1869 | ZBTB24: Immunodeficiency-centromeric instability-facial anomalies syndrome-2 {(NM\_014797.2) "c.501dupA"} |
| 1870 - 1870 | ZMPSTE24: Mandibuloacral dysplasia with type B lipodystrophy {(NM\_005857) "c.1085dupT"} |
| 1871 - 1872 | ZNF469: Brittle cornea syndrome 1 {(NM\_001127464.2) "c.5943delA", "c.9531delG"} |

**\* לפי החלטת איגוד הגנטיקאים הישראלי, המוטציה מדווחת חיובית רק ליהודים ממוצא קווקזי**

,De boer M, Gavrieli R, Van leeuwen K, et al. A false-carrier state for the c.579G>A mutation in the NCF1 gene in Ashkenazi Jews. J Med Genet. 2018;55(3):166-172.

**\*\* לפי החלטת איגוד הגנטיקאים הישראלי, המוטציה מדווחת חיובית רק למוצא הדרוזי**

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