**(2020-08-11) Ver 2.0 - 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 - 12 | ABCA4:Cone-rod dystrophy 3 {(NM\_000350.3) "c.1648G>A", "c.2791G>T", "c.3607G>A", "c.3608G>A", "c.5460+1G>A", "c.834delT"} |
| 13 - 14 | ABCB11:Cholestasis, progressive familial intrahepatic type 2 {(NM\_003742.4) "c.1409G>A", "c.3268C>T"} |
| 15 - 19 | ABCC8:Hyperinsulinemic hypoglycemia, familial, 1 (PHHI/HHF1) {(NM\_000352.4) "c.1116dupT", "c.2506C>T", "c.3989-9G>A", "c.4160\_4162delTCT", "c.560T>A"} |
| 20 - 20 | ABCD1:X-linked adrenoleukodystrophy {(NM\_000033.4) "c.686T>C"} |
| 21 - 22 | ABHD5:Chanarin-Dorfman syndrome {(NM\_016006.6) "c.412T>C", "c.934C>T"} |
| 23 - 25 | ACADM:Medium-chain Acyl-CoA dehydrogenase deficiency {(NM\_000016.5) "c.616C>T", "c.799G>A" | (NM\_001127328) "c.374C>T"} |
| 26 - 34 | ACADVL:Acyl-CoA dehydrogenase, very long-chain, VLCAD deficiency {(NM\_000018.4) "c.1096C>T", "c.1748C>T", "c.260T>C", "c.367G>A", "c.637G>A", "c.65C>A", "c.779C>T", "c.799\_802delGTTA", "c.894G>A"} |
| 35 - 35 | ACO2:Infantile cerebellar-retinal degeneration {(NM\_001098.3) "c.336C>G"} |
| 36 - 36 | ACP5:Spondyloenchondrodysplasia with immune dysregulation {(NM\_001111035.2) "c.325G>A"} |
| 37 - 37 | ACSF3:Combined malonic and methylmalonic aciduria {(NM\_174917) "c.1411C>T"} |
| 38 - 39 | ADAMTS2:Ehlers Danlos syndrome, type VIIC {(NM\_014244.5) "c.2384G>A", "c.673C>T"} |
| 40 - 41 | ADA:Severe combined immunodeficiency due to ADA deficiency {(NM\_000022.4) "c.703C>T", "c.792G>A"} |
| 42 - 45 | ADGRG1:Bilateral Frontoparietal Polymicrogyria (BFPP) {(NM\_005682.7) "c.1046G>C", "c.1167+3G>C", "c.1693C>T", "c.739\_745delCAGGACC"} |
| 46 - 47 | ADGRV1:Usher syndrome, type 2C {(NM\_032119.4) "c.14973-2A>G", "c.15494delA"} |
| 48 - 48 | AGA:Aspartylglucosaminuria {(NM\_000027.4) "c.214T>C"} |
| 49 - 51 | AGL:Glycogen storage disease III {(NM\_000642.3) "c.1078C>T", "c.1222C>T", "c.4455delT"} |
| 52 - 64 | AGXT:Hyperoxaluria, primary, type I {(NM\_000030.3) "c.121G>A", "c.33dupC", "c.466G>A", "c.584T>G", "c.586G>A", "c.680+1G>A", "c.697C>T", "c.727G>C", "c.731T>C", "c.860\_861delGCinsCG", "c.865C>T", "c.893T>C", "c.997A>T"} |
| 65 - 67 | AHI1:Joubert syndrome-3 {(NM\_017651.4) "c.2212C>T", "c.3032C>G", "c.787dupC"} |
| 68 - 68 | AIMP1:Leukodystrophy, hypomyelinating, 3 {(NM\_004757.3) "c.292\_293delCA"} |
| 69 - 71 | AIPL1:Leber congenital amaurosis 4 {(NM\_014336.5) "c.211G>T", "c.215G>A", "c.834G>A"} |
| 72 - 77 | AIRE:Autoimmune polyendocrinopathy syndrome , type I (APS-1) with or without reversible metaphyseal dysplasia {(NM\_000383.4) "c.1163\_1164insA", "c.247A>G", "c.254A>G", "c.44G>A", "c.47C>T", "c.769C>T"} |
| 78 - 78 | ALDH1A3:Microphthalmia, isolated 8 {(NM\_000693.4) "c.211G>A"} |
| 79 - 79 | ALDH7A1:Epilepsy, pyridoxine-dependent {(NM\_001182.5) "c.1489+5G>A"} |
| 80 - 82 | ALDOB:Fructose intolerance {(NM\_000035.4) "c.1005C>G", "c.448G>C", "c.524C>A"} |
| 83 - 84 | ALMS1:Alstrom syndrome {(NM\_015120.4) "c.8008C>T", "c.808C>T"} |
| 85 - 86 | ALPL:Hypophosphatasia, infantile {(NM\_000478.6) "c.1348C>T", "c.141C>A"} |
| 87 - 87 | AMT:Glycine encephalopathy, AMT-related {(NM\_000481.3) "c.125A>G"} |
| 88 - 88 | ANO5:Limb-girdle muscular dystrophy {(NM\_213599.2) "c.191dupA"} |
| 89 - 89 | AP4B1:Spastic paraplegia 47, Autosomal Recessive {(NM\_006594.4) "c.664delC"} |
| 90 - 90 | APTX:Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia {(NM\_175073.2) "c.837G>A"} |
| 91 - 92 | AQP2:Diabetes insipidus, nephrogenic {(NM\_000486.5) "c.298G>T", "c.83T>C"} |
| 93 - 93 | ARFGEF2:Periventricular heterotopia with microcephaly {(NM\_006420.3) "c.1958+1G>A"} |
| 94 - 94 | ARHGDIA:Nephrotic syndrome, type 8 {(NM\_004309.6) "c.518G>T"} |
| 95 - 95 | ARL6:Bardet-Biedl syndrome 3 {(NM\_032146.5) "c.364C>T"} |
| 96 - 108 | ARSA:Metachromatic leukodystrophy - MLD {(NM\_000487.6) "c.1136C>T", "c.1174C>T", "c.1283C>T", "c.211T>G", "c.263G>A", "c.292\_293delTCinsCT", "c.465+1G>A", "c.47G>A", "c.542T>G", "c.576G>C", "c.827C>T", "c.937C>T" | (NM\_001085425.3) "c.449C>T"} |
| 109 - 109 | ASL:Argininosuccinic aciduria {(NM\_000048.4) "c.346C>T"} |
| 110 - 110 | ASNS:Asparagine synthetase deficiency {(NM\_183356.3) "c.1084T>G"} |
| 111 - 114 | ASPA:Canavan Disease {(NM\_001128085.1) "c.433-2A>G", "c.693C>A", "c.854A>C", "c.914C>A"} |
| 115 - 115 | ASS1:Citrullinemia,classic {(NM\_000050) "c.1168G>A"} |
| 116 - 128 | ATM:Ataxia-tyelangiectasia {(NM\_000051.3) "c.103C>T", "c.1339C>T", "c.1547T>C", "c.2284\_2285delCT", "c.2839-579\_2839-576del4", "c.3245\_3247delATCinsTGAT", "c.3576G>A", "c.368delA", "c.497del7514", "c.5763-1050A>G", "c.6672\_6680delGGCTCTACGinsCTC", "c.7241\_7244delAAGC", "deletion\_exon\_3-4"} |
| 129 - 129 | ATP6V0A2:Cutis laxa, Autosomal Recessive, type IIA {(NM\_012463.4) "c.2375C>G"} |
| 130 - 150 | ATP7B:Wilson disease {(NM\_000053.3) "c.122A>G", "c.1340\_1343delAAAC", "c.1544G>A", "c.1639delC", "c.1703T>G", "c.1934T>G", "c.2293G>A", "c.2333G>T", "c.2337G>A", "c.2817G>T", "c.2906G>A", "c.3191A>C", "c.3207C>A", "c.3451C>T", "c.3551T>C", "c.3638G>T", "c.3649\_3654delGTTCTG", "c.3659C>T", "c.3842G>A", "c.4152T>G", "c.845delT"} |
| 151 - 151 | AVP:Familial neurohypophyseal diabetes insipidus {(NM\_000490) "c.77C>T"} |
| 152 - 152 | B4GALT1:Congenital disorder of glycosylation, type IId {(NM\_001497) "c.61C>T"} |
| 153 - 155 | BBS10:Bardet-Biedl syndrome 10 {(NM\_024685.4) "c.1091delA", "c.1399delA", "c.271dupT"} |
| 156 - 157 | BBS1:Bardet-Biedl syndrome 1 {(NM\_024649.5) "c.1169T>G", "c.479G>A"} |
| 158 - 162 | BBS2:Bardet-Biedl syndrome 2 {(NM\_031885.4) "c.1895G>C", "c.224T>G", "c.311A>C", "c.401C>G", "c.98C>A"} |
| 163 - 164 | BBS4:Bardet-Biedl syndrome 4 {(NM\_033028.5) "c.77-1422\_221-753del ", "c.884G>C"} |
| 165 - 165 | BBS7:Bardet-Biedl syndrome 7 {(NM\_176824.3) "c.1786G>A"} |
| 166 - 172 | BCKDHA:Maple syrup urine disease, type Ia {(NM\_000709.4) "c.169delG", "c.718del", "c.792C>G", "c.859C>T", "c.890G>A", "c.909\_910delGT", "c.935\_937del"} |
| 173 - 179 | BCKDHB:Maple syrup urine disease, type Ib {(NM\_000056.4) "c.1016C>T", "c.1114G>T", "c.356T>G", "c.548G>C", "c.670C>T", "c.800\_803delAGGA", "c.832G>A"} |
| 180 - 184 | BLM:Bloom syndrome {(NM\_000057.4) "c.1984\_1985delAA", "c.2207\_2212delATCTGAinsTAGATTC", "c.2407dupT", "c.3510T>A", "c.98+1G>T"} |
| 185 - 185 | BMPER:Diaphanospondylodysostosis {(NM\_133468.5) "c.310C>T"} |
| 186 - 186 | BMPR1B:Brachydactyly type A2 {(NM\_001256793.2) "c.377G>A"} |
| 187 - 188 | BSND:Bartter syndrome, type 4a infantile variant with sensorineuronal deafness {(NM\_057176.3) "c.167\_168insTTTCCC", "c.28G>A"} |
| 189 - 189 | BTD:Biotinidase deficiency {(NM\_000060.4) "c.100G>A"} |
| 190 - 191 | C12ORF65:Spastic paraplegia 55, Autosomal Recessive {(NM\_152269.5) "c.282+2T>A", "c.413\_417delAACAA"} |
| 192 - 192 | C21orf59:Ciliary dyskinesia, primary, 26 {(NM\_021254.4) "c.735C>G"} |
| 193 - 195 | C8orf37:Retinitis pigmentosa 64 {(NM\_177965.4) "c.497T>A", "c.529C>T", "c.545A>G"} |
| 196 - 196 | CAPN3:Muscular dystrophy, limb-girdle, type 2A {(NM\_000070) "c.1469G>A"} |
| 197 - 197 | CASQ2:Ventricular tachycardia, catecholaminergic polymorphic, 2 {(NM\_001232.3) "c.919G>C"} |
| 198 - 201 | CBS:Homocystinuria, thrombosis, hyperhomocysteinemic {(NM\_000071.2) "c.1006C>T", "c.1224-2A>C", "c.1261delG", "c.785C>G"} |
| 202 - 202 | CC2D1A:Mental retardation, Autosomal Recessive 3 - MRT3 {(NM\_017721.5) "c.1468+1\_1824-1del"} |
| 203 - 203 | CCDC174:Birk Volodarsky PMR Synderome Hypotonia and psychomotor developmental delay {(NM\_016474.5) "c.1404A>G"} |
| 204 - 204 | CCDC65:Ciliary dyskinesia, primary, 27 {(NM\_033124.5) "c.877\_878delAT"} |
| 205 - 205 | CCDC88C:Hydrocephalus, nonsyndromic, Autosomal Recessive {(NM\_001080414.4) "c.934C>T"} |
| 206 - 206 | CD59:Hemolytic anemia & immune-mediated polyneuropathy, CD59-related {(NM\_203330.2) "c.266G>A"} |
| 207 - 207 | CDAN1:Dyserythropoietic anemia, congenital, type Ia {(NM\_138477.4) "c.3124C>T"} |
| 208 - 208 | CDH23:Usher Syndrome Type ID {(NM\_022124.6) "c.7903G>T"} |
| 209 - 209 | CDK5:Lissencephaly 7 with cerebellar hypoplasia {(NM\_004935.4) "c.580+1G>A"} |
| 210 - 210 | CECR1:Polyarteritis nodosa, childhood-onset {(NM\_017424.2) "c.139G>A"} |
| 211 - 211 | CEP104:Joubert syndrome (JBTS) {(NM\_014704.4) "c.1328\_1329insT"} |
| 212 - 212 | CEP152:Microcephaly 9, primary, Autosomal Recessive {(NM\_014985.3) "c.2281-2A>G"} |
| 213 - 218 | CEP290:Meckel syndrome 4 {(NM\_025114.3) "c.1225delA", "c.164\_167delCTCA", "c.1666delA", "c.4393C>T", "c.4771C>T", "c.5788A>T"} |
| 219 - 219 | CERKL:Retinitis pigmentosa 26 {(NM\_001030311.2) "c.238+1G>A"} |
| 220 - 220 | CFH:Hemolytic uremic syndrome, complement factor H deficiency {(NM\_000186.3) ":c.3677\_\*4del"} |
| 221 - 465 | CFTR:Cystic fibrosis {(NM\_000492.3) "c.1000C>T", "c.1001G>A", "c.1007T>A", "c.1013C>T", "c.1021\_1022dupTC", "c.1021T>C", "c.1040G>A", "c.1040G>C", "c.1040G>T", "c.1055G>A", "c.1075C>A", "c.[1075C>A;1079C>A]", "c.1081delT", "c.1116+1G>A", "c.1130dupA", "c.1155\_1156dupTA", "c.115C>T", "c.11C>A", "c.1202G>A", "c.1203G>A", "c.1209+1G>A", "c.1240C>T", "c.1327\_1330dupGATA", "c.1340delA", "c.1364C>A", "c.137C>A", "c.1393-1G>A", "c.1397C>A", "c.1397C>G", "c.1400T>C", "c.1418delG", "c.1438G>T", "c.1439G>A", "c.1466C>A", "c.1475C>T", "c.1477C>T", "c.1521\_1523delCTT", "c.1545\_1546delTA", "c.1558G>A", "c.1558G>T", "c.1572C>A", "c.1573C>T", "c.1585-1G>A", "c.1585-8G>A", "c.1624G>T", "c.1625G>A", "c.1645A>C", "c.1646G>A", "c.1646G>T", "c.1647T>G", "c.1650delA", "c.1651G>A", "c.165-1G>A", "c.1652G>A", "c.1654C>T", "c.1657C>T", "c.166G>A", "c.1673T>C", "c.1675G>A", "c.1679+1634A>G", "c.1679+1G>C", "c.1679G>A", "c.1679G>C", "c.1680-1G>A", "c.1682C>A", "c.1692delA", "c.1736A>G", "c.1753G>T", "c.175dupA", "c.1766+1G>A", "c.1766+1G>C", "c.1766+3A>G", "c.1766+5G>T", "c.178G>T", "c.1817\_1900del84", "c.1841A>G", "c.1911delG", "c.1923\_1931delCTCAAAACTinsA", "c.1973\_1985delGAAATTCAATCCTinsAGAAA", "c.1976delA", "c.1986\_1989delAACT", "c.1A>G", "c.200C>T", "c.2012delT", "c.2051\_2052delAAinsG", "c.2052\_2053insA", "c.2052delA", "c.2089dupA", "c.2125C>T", "c.2128A>T", "c.2175dupA", "c.2195T>G", "c.2215delG", "c.223C>T", "c.2290C>T", "c.233dupT", "c.2353C>T", "c.2374C>T", "c.2423\_2424dupAT", "c.2453delT", "c.2462\_2463delGT", "c.2464G>T", "c.2490+1G>A", "c.2491G>T", "c.2537G>A", "c.2547C>A", "c.254G>A", "c.2551C>T", "c.2583delT", "c.2619+1G>A", "c.2619+2dupT", "c.262\_263delTT", "c.2657+2\_2657+3insA", "c.2657+5G>A", "c.2658-1G>C", "c.2668C>T", "c.273+1G>A", "c.273+3A>C", "c.2737\_2738insG", "c.2739T>A", "c.274-1G>A", "c.274G>A", "c.274G>T", "c.2763\_2764dupAG", "c.2780T>C", "c.2834C>T", "c.2856G>C", "c.2875delG", "c.2908G>C", "c.292C>T", "c.2930C>T", "c.2988+1173\_c.3468+2111del8898", "c.2988+1G>A", "c.2988G>A", "c.2989-1G>A", "c.2989-977\_3367+248del", "c.3002\_3003delTG", "c.3039delC", "c.3041A>G", "c.3067\_3072delATAGTG", "c.3139+10T>C", "c.313delA", "c.3140-26A>G", "c.3154T>G", "c.3160C>G", "c.3181G>C", "c.3194T>C", "c.3196C>T", "c.3197G>A", "c.3205G>A", "c.3208C>T", "c.3209G>A", "c.3222T>A", "c.3230T>C", "c.325\_327delTATinsG", "c.3266G>A", "c.3276C>G", "c.328G>C", "c.3292T>C", "c.3299A>C", "c.3302T>A", "c.3310G>T", "c.343G>T", "c.3454G>C", "c.3469-2A>G", "c.3472C>T", "c.3484C>T", "c.349C>T", "c.350G>A", "c.3528delC", "c.3529A>T", "c.3532\_3535dupTCAA", "c.3535\_3538delACCA", "c.3587C>G", "c.3600delA", "c.3605delA", "c.3611G>A", "c.3612G>A", "c.3659delC", "c.366T>A", "c.3691delT", "c.3700A>G", "c.3712C>T", "c.3717+12191C>T", "c.3718-1G>A", "c.3731G>A", "c.3744delA", "c.3752G>A", "c.3763T>C", "c.3764C>A", "c.3764C>T", "c.3773dupT", "c.3793G>A", "c.3808delG", "c.3846G>A", "c.3873+1G>A", "c.3883\_3884insG", "c.3883\_3886delATTT", "c.3883delA", "c.3889dupT", "c.3909C>G", "c.3937C>T", "c.3964-78\_4242+577del", "c.4046G>A", "c.4077\_4080delTGTTinsAA", "c.4111G>T", "c.413\_415dupTAC", "c.416A>C", "c.416A>T", "c.4196\_4197delTC", "c.422C>A", "c.4234C>T", "c.4242+1G>T", "c.4251delA", "c.4297G>A", "c.4300\_4301dupAG", "c.4364C>G", "c.442delA", "c.487A>G", "c.489+1G>T", "c.523A>G", "c.531delT", "c.532G>A", "c.543\_546delTAGT", "c.54-5940\_273+10250del21Kb", "c.575A>G", "c.579+1G>T", "c.579+3A>G", "c.579+5G>A", "c.580-1G>T", "c.595C>T", "c.613C>T", "c.617T>G", "c.658C>T", "c.675T>A", "c.761delA", "c.803delA", "c.805\_806delAT", "c.828C>A", "c.870-2A>G", "c.892delA", "c.933\_935delCTT", "c.948delT", "c.988G>T" | (NM\_000492.4) "c.1079C>A"} |
| 466 - 467 | CLCN1:Myotonia congenita, Autosomal Recessive {(NM\_000083) "c.1444G>A", "c.1586C>T"} |
| 468 - 470 | CLCN5:Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis {(NM\_000084.4) "c.1399C>T", "c.258delA", "c.82C>T"} |
| 471 - 471 | CLCNKB:Bartter syndrome, type 3 and Gitelman syndrome {(NM\_000085.4) "c.1313G>A"} |
| 472 - 472 | CLN5:Ceroid lipofuscinosis, neuronal, 5 {(NM\_006493.3) "c.672delG"} |
| 473 - 473 | CLN6:Ceroid lipofuscinosis, neuronal, 6 {(NM\_017882.3) "c.214G>T"} |
| 474 - 474 | CLN8:Neuronal ceroid lipofuscinosis type 8, including northern epilepsy {(NM\_018941.3) "c.766C>G"} |
| 475 - 478 | CLRN1:Usher syndrome, type 3A {(NM\_174878.2) "c.144T>G", "c.349\_358del", "c.433+1G>A", "c.528T>G"} |
| 479 - 484 | CNGA3:Achromatopsia-2 - total color blindness {(NM\_001298.2) "c.1114C>T", "c.1585G>A", "c.1640T>G", "c.67C>T", "c.940\_942delATC", "c.985G>T"} |
| 485 - 485 | CNGB1:Retinitis pigmentosa 45 {(NM\_001297.5) "c.2284C>T"} |
| 486 - 487 | CNGB3:Achromatopsia-3,macular degeneration, juvenile {(NM\_019098.4) "c.1148delC", "c.644-1G>C"} |
| 488 - 489 | CNNM4:Jalili syndrome {(NM\_020184.4) "c.1813C>T", "c.599C>A"} |
| 490 - 490 | COL11A2:Otospondylomegaepiphyseal dysplasia (ZW) {(NM\_080680.2) "c.3991C>T"} |
| 491 - 494 | COL4A3:Alport Syndrome, COL4A3-Related {(NM\_000091.4) "c.1791\_1793dupTCC", "c.227delG", "c.3518-2A>G", "c.4649T>G"} |
| 495 - 496 | COL4A4:Alport syndrome, COL4A4-Related {(NM\_000092.4) "c.3933C>G", "c.785\_792dupCACCTGAC"} |
| 497 - 501 | COL4A5:Alport syndrome, COL4A5-Related {(NM\_000495.4) "c.2641G>T", "c.367delG", "c.4691G>C", "c.4946T>G", "c.5030G>A"} |
| 502 - 502 | COLEC11:3MC syndrome 2 {(NM\_199235.2) "c.627\_628delCG"} |
| 503 - 506 | COLQ:Myasthenic syndrome, congenital, 5 {(NM\_005677) "c.788dupC" | (NM\_005677.4) "c.1228C>T", "c.718G>T", "c.792dupG"} |
| 507 - 507 | COQ4:Coenzyme Q10 deficiency, primary, 7 {(NM\_016035.5) "c.718C>T"} |
| 508 - 513 | CPS1:Carbamoylphosphate synthetase I deficiency {(NM\_001875.5) "c.1760G>A", "c.3265C>T", "c.3374C>T", "c.3558+1G>C", "c.4101+2T>C", "c.794C>T"} |
| 514 - 514 | CPT1A:Carnitine palmitoyltransferase 1 deficiency {(NM\_001031847) "c.1361A>G"} |
| 515 - 515 | CPT2:Carnitine palmitoyltransferase II deficiency {(NM\_000098) "c.110\_111dupGC"} |
| 516 - 530 | CRB1:Leber congenital amaurosis 8 {(NM\_201253.3) "c.1148G>A", "c.1576C>T", "c.1733T>A", "c.1842delT", "c.1844G>T", "c.2230C>T", "c.2234C>T", "c.2498G>A", "c.2555T>C", "c.2680\_2684delAACCC", "c.3307G>A", "c.4005+1G>A", "c.4121\_4130delCAACTCAGGG", "c.424G>T", "c.455G>A"} |
| 531 - 534 | CRB2:Ventriculomegaly with cystic kidney disease {(NM\_173689.7) "c.1882C>T", "c.1928A>C", "c.2277G>A", "c.2400C>G"} |
| 535 - 535 | CRTAP:Osteogenesis imperfecta, type VII {(NM\_006371.4) "c.793+1G>T"} |
| 536 - 536 | CSTA:Exfoliative ichthyosis, Autosomal Recessive, ichthyosis bullosa of Siemens-like {(NM\_005213.4) "c.67-2A>T"} |
| 537 - 538 | CTNS:Cystinosis,CTNS-related {(NM\_004937.2) "c.1015G>A", "c.530A>C"} |
| 539 - 539 | CTSC:Haim-Munk syndrome {(NM\_001814.6) "c.857A>G"} |
| 540 - 540 | CTSK:Pycnodysostosis {(NM\_000396.4) "c.990A>G"} |
| 541 - 544 | CYBA:Chronic granulomatous disease,autosomal, due to deficiency of CYBA {(NM\_000101.4) "c.164C>G", "c.171dupG", "c.70G>A", "c.71G>A"} |
| 545 - 549 | CYBB:Chronic granulomatous disease, X-linked {(NM\_000397) "c.1016dupC", "c.1081T>C", "c.271C>T", "c.676C>T", "c.90\_92delCCGinsGGT"} |
| 550 - 551 | CYP11A1:Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete {(NM\_000781.3) "c.644T>C", "c.694C>T"} |
| 552 - 552 | CYP11B2:Hypoaldosteronism, congenital, due to CMO II deficiency {(NM\_000498.3) "c.541C>T"} |
| 553 - 554 | CYP1B1:Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset {(NM\_000104.3) "c.1568G>A", "c.182G>A"} |
| 555 - 559 | CYP27A1:Cerebrotendinous xanthomatosis {(NM\_000784.4) "c.1016C>T", "c.1184G>A", "c.355delC", "c.819delT", "c.845-1G>A"} |
| 560 - 560 | CYP7B1:Spastic paraplegia 5A, Autosomal Recessive {(NM\_004820.5) "c.1081C>T"} |
| 561 - 561 | DAG1:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 {(NM\_001165928.3) "c.743delC"} |
| 562 - 562 | DARS2:Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation {(NM\_018122.5) "c.492+2T>C"} |
| 563 - 563 | DBT:Maple syrup urine disease, type II {(NM\_001918) "c.581C>G"} |
| 564 - 564 | DCAF17:Woodhouse-Sakati syndome {(NM\_025000.4) "c.436delC"} |
| 565 - 565 | DCLRE1C:Severe combined immunodeficiency, Athabascan type {(NM\_001033858.2) "c.1307\_1308insAGGATGCT"} |
| 566 - 566 | DDR2:Spondylometaepiphyseal dysplasia, short limb-hand type {(NM\_006182.4) "c.2254C>T"} |
| 567 - 567 | DGAT1:Diarrhea 7, congenital {(NM\_012079.6) "c.751+2T>C"} |
| 568 - 568 | DGUOK:Mitochondrial DNA depletion syndrome (hepatocerebral type) {(NM\_080916.3) "c.255delA"} |
| 569 - 569 | DGUOK:Mitochondrial DNA depletion syndrome {(NM\_080916.3) "c.271delA"} |
| 570 - 570 | DHCR24:Desmosterolosis {(NM\_014762.4) "c.307C>T"} |
| 571 - 585 | DHCR7:Smith Lemli Opitz syndrome {(NM\_001360.2) "c.1054C>T", "c.1210C>T", "c.1228G>A", "c.1A>G", "c.278C>T", "c.452G>A", "c.453G>A", "c.506C>T", "c.724C>T", "c.725G>A", "c.755A>G", "c.906C>G", "c.964-1G>C", "c.964-1G>T", "c.976G>T"} |
| 586 - 586 | DHDDS:Retinitis pigmentosa 59 {(NM\_024887.3) "c.124A>G"} |
| 587 - 590 | DLD:Dihydrolipoamide Dehydrogenase Deficiency {(NM\_000108.5) "c.104dupA", "c.1123G>A", "c.1436A>T", "c.685G>T"} |
| 591 - 591 | DLL3:Spondylocostal dysostosis 1, Autosomal Recessive {(NM\_016941.3) "c.395delG"} |
| 592 - 593 | DNAH11:Ciliary dyskinesia, primary, 7, with or without situs inversus {(NM\_001277115.2) "c.11929G>T", "c.13242\_13245delAAAG"} |
| 594 - 594 | DNAH5:Ciliary dyskinesia, primary, 3, with or without situs inversus (CILD3/PCD) {(NM\_001369.2) "c.8011-2A>G"} |
| 595 - 595 | DNAI1:Ciliary dyskinesia, primary, 1, with or without situs inversus {(NM\_012144.4) "c.1490G>A"} |
| 596 - 597 | DNAI2:Ciliary dyskinesia, primary, 9, with or without situs inversus {(NM\_023036.6) "c.1304G>A", "c.1494+1G>A"} |
| 598 - 598 | DNAL1:Ciliary dyskinesia, primary, 16 {(NM\_031427.4) "c.449A>G"} |
| 599 - 599 | DOCK8:Hyper-IgE recurrent infection syndrome, autosomal recessive {(NM\_203447) "c.5132C>A"} |
| 600 - 601 | DOLK:Congenital disorder of glycosylation, type Im {(NM\_014908.3) "c.1222C>G", "c.912G>T"} |
| 602 - 603 | DSG1:Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE {(NM\_001942.4) "c.1861delG", "c.395C>A"} |
| 604 - 604 | DST:Epidermolysis bullosa simplex, Autosomal Recessive 2 {(NM\_183380.3) "c.14865delA"} |
| 605 - 610 | DYSF:Muscular dystrophy, limb-girdle, type 2B {(NM\_003494.4) "c.2372C>G", "c.2779delG", "c.4741C>T", "c.4872\_4876delGCCCGinsCCCC", "c.5057+5G>A", "c.5429G>A"} |
| 611 - 613 | ELP1:Dysautonomia, familial {(NM\_003640.5) "c.2087G>C", "c.2204+6T>C", "c.2741C>T"} |
| 614 - 614 | EOGT:Adams-Oliver syndrome 4 {(NM\_001278689.2) "c.1074delA"} |
| 615 - 615 | EPCAM:Colorectal cancer, hereditary nonpolyposis, type 1 {(NM\_002354) "c.-358\_\*415del"} |
| 616 - 616 | EPG5:Vici syndrome {(NM\_020964.3) "c.5704dupT"} |
| 617 - 617 | EPM2A:Epilepsy, progressive myoclonic 2A (Lafora) {(NM\_005670) "56\_kb\_incl.\_ex.\_2"} |
| 618 - 618 | ERBB3:Lethal congenital contractural syndrome 2 {(NM\_001982.3) "c.1184-9A>G"} |
| 619 - 619 | ERCC2:Xeroderma pigmentosum, group D {(NM\_000400.3) "c.2048G>A"} |
| 620 - 620 | ERCC5:Xeroderma pigmentosum/Cockayne {(NM\_000123.3) "c.205C>T"} |
| 621 - 621 | ERCC6:Cockayne syndrome, type B {(NM\_000124.4) "c.1034\_1035insT"} |
| 622 - 624 | ERCC8:Cockayne syndrome, type A {(NM\_000082.3) "c.37G>T", "c.843+1G>C", "c.966C>A"} |
| 625 - 625 | ESCO2:Roberts-SC phocomelia syndrome {(NM\_001017420.3) "c.1674-2A>G"} |
| 626 - 629 | ETFDH:Glutaric acidemia IIC {(NM\_004453.4) "c.1074G>C", "c.1084G>A", "c.1425C>A", "c.299T>A"} |
| 630 - 630 | EXOSC3:Pontocerebellar hypoplasia, type 1B {(NM\_016042.4) "c.571G>T"} |
| 631 - 631 | EXOSC8:Pontocerebellar hypoplasia, type 1C {(NM\_181503.3) "c.5C>T"} |
| 632 - 635 | EYS:Retinitis pigmentosa 25 {(NM\_001142800.2) "c.1211dupA", "c.3699delG", "c.3715G>T", "c.8155\_8156delCA"} |
| 636 - 636 | F7:Factor VII deficiency {(NM\_000131.4) "c.1256C>T"} |
| 637 - 637 | FA2H:Spastic paraplegia 35, Autosomal Recessive {(NM\_024306.5) "c.786+1G>A"} |
| 638 - 639 | FAH:Tyrosinemia type 1 {(NM\_000137.2) "c.1069G>T", "c.192G>T"} |
| 640 - 644 | FAH:Tyrosinemia, type I {(NM\_000137.2) "c.1062+5G>A", "c.554-1G>T", "c.707-1G>C", "c.782C>T", "c.786G>A"} |
| 645 - 648 | FAM161A:Retinitis pigmentosa 28 {(NM\_001201543.2) "c.1309A>T", "c.1355\_1356delCA", "c.1567C>T" | (NM\_032180) "c.1618C>T"} |
| 649 - 649 | FAM20A:Amelogenesis imperfecta, type IG (enamel-renal syndrome) {(NM\_017565.4) "c.1523delC"} |
| 650 - 657 | FANCA:Fanconi anemia, complementation group A {(NM\_000135.4) "c.189+1G>A", "c.2172dupG", "c.3788\_3790delTCT", "c.4168-2A>C", "c.4261-2A>C", "c.4275delT", "c.891\_893+1delCTGG", "c.Del\_exon\_31-37"} |
| 658 - 663 | FANCC:Fanconi anemia, complementation group C {(NM\_000136.3) "c.1642C>T", "c.1661T>C", "c.37C>T", "c.456+4A>T", "c.553C>T", "c.67delG"} |
| 664 - 665 | FANCG:Fanconi Anemia - complementation group G {(NM\_004629.1) "c.212T>C", "c.510+3A>G"} |
| 666 - 666 | FDX1L:Mitochondrial muscle myopathy {(NM\_001031734.4) "c.10A>T"} |
| 667 - 669 | FERMT1:Kindler syndrome {(NM\_017671.4) "c.137\_140delTAGT", "c.-1500\_-19+470del", "c.749G>A"} |
| 670 - 670 | FGB:Afibrinogenemia congenital {(NM\_005141.4) "c.1400G>A"} |
| 671 - 672 | FGFR3:Achondroplasia {(NM\_000142​) "c.1138G>A", "c.1138G>C"} |
| 673 - 674 | FGFR3:Hypochondroplasia {(NM\_000142​) "c.1620C>A", "c.1620C>G"} |
| 675 - 675 | FH:Fumarase deficiency, leiomyomatosis and renal cell cancer {(NM\_000143.3) "c.905-1G>A"} |
| 676 - 676 | FKBP10:Osteogenesis imperfecta, type XI {(NM\_021939.3) "c.310C>T"} |
| 677 - 677 | FKRP:Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 {(NM\_024301.5) "c.160C>T"} |
| 678 - 678 | FKTN:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 - Walker Warburg syndrome {(NM\_001079802.1) "c.1167dupA"} |
| 679 - 679 | FLT4:Autosomal Recessive Hereditary Lymphedema {(NM\_182925.5) "c.3704C>G"} |
| 680 - 680 | FOXRED1:Mitochondrial encephalomyopathy complex I deficiency {(NM\_017547.4) "c.1054C>T"} |
| 681 - 681 | FRMD4A:Microcephaly intellectual disability and dysmorphism {(NM\_018027) "c.2134\_2146dup13"} |
| 682 - 682 | FTO:Growth retardation, developmental delay, coarse facies, and early death {(NM\_001080432.3) "c.947G>A"} |
| 683 - 684 | G6PC3:Neutropenia, severe congenital 4, Autosomal Recessive {(NM\_138387.3) "c.765\_766delAG", "c.785G>A"} |
| 685 - 696 | G6PC:Glycogen storage disease Ia - GDS1a {(NM\_000151.4) "c.1039C>T", "c.247C>T", "c.248G>A", "c.379\_380dupTA", "c.497T>G", "c.508C>T", "c.562G>C", "c.648G>T", "c.724C>T", "c.79delC", "c.809G>T", "c.979\_981delTTC"} |
| 697 - 707 | GAA:Pompe (Glycogen storage disease type II) {(NM\_000152.5) "c.1064T>C", "c.1082C>T", "c.1210G>A", "c.1564C>A", "c.1935C>A", "c.1942G>A", "c.2560C>T", "c.340\_341insT", "c.670C>T", "c.896T>C", "c.896T>G"} |
| 708 - 710 | GALC:Krabbe disease {(NM\_000153.4) "c.1630G>A", "c.1748A>C", "c.1796T>G"} |
| 711 - 712 | GALNT3:Tumoral calcinosis, hyperphosphatemic, familial {(NM\_004482.4) "c.1524+1G>A", "c.1524+5G>A"} |
| 713 - 722 | GALT:Galactosemia {(NM\_000155.3) "5.5-KB\_DEL", "c.152G>A", "c.253-2A>G", "c.404C>T", "c.413C>T", "c.512T>C", "c.563A>G", "c.584T>C", "c.626A>G", "c.855G>T"} |
| 723 - 723 | GAN:Giant axonal neuropathy 1 {(NM\_022041.3) "c.973G>A"} |
| 724 - 724 | GATC:Hypertophic Cardiomyopathy {(NM\_176818) "c.233T>G"} |
| 725 - 725 | GATM:Cerebral creatine deficiency syndrome 3 {(NM\_001482.3) "c.1111dupA"} |
| 726 - 737 | GBA:Gaucher disease, , type I {(NM\_001005741.3) "c.115+1G>A", "c.1226A>G", "c.1294T>A", "c.1297G>T", "c.1342G>C", "c.1448T>C", "c.1504C>T", "c.1505G>A", "c.1604G>A", "c.259C>T", "c.703T>C", "c.84dupG"} |
| 738 - 738 | GBE1:Glycogen storage disease IV {(NM\_000158) "c.2053-3358\_\*3188delinsTGTTTTTTACATGACAGGT"} |
| 739 - 750 | GCDH:Glutaricaciduria type I {(NM\_000159.4) "c.1168G>C", "c.1173delG", "c.1204C>T", "c.1205G>A", "c.1247C>T", "c.1262C>T", "c.1306G>T", "c.301G>A", "c.505+1G>A", "c.848T>C", "c.877G>A", "c.914C>T"} |
| 751 - 752 | GH1:Growth hormone deficiency, isolated, type IA {(NM\_000515.5) "c.456+5G>C", "c.67G>T"} |
| 753 - 753 | GHRHR:Growth hormone deficiency, isolated, type IB {(NM\_000823.4) "c.1069C>T"} |
| 754 - 759 | GHR:Laron dwarfism {(NM\_000163.5) "c.11G>A", "c.594A>G", "c.62G>A", "c.703C>T", "c.744delT", "del5,6ex"} |
| 760 - 774 | GJB2:Deafness, autosomal recessive 1A {(NM\_004004.6) "c.109G>A", "c.167delT", "c.229T>C", "c.230G>A", "c.235delC", "c.250G>A", "c.269T>C", "c.358\_360delGAG", "c.35delG", "c.370C>T", "c.51\_62delCACCAGCATTGGinsA", "c.551G>C", "c.614T>C", "c.71G>A", "c.94C>T"} |
| 775 - 775 | GJB6:Deafness, Autosomal Recessive 1B {(NM\_006783.4) "309\_kb"} |
| 776 - 781 | GLB1:GM1-gangliosidosis, type I {(NM\_000404.4) "c.1038G>C", "c.485delT", "c.602G>A", "c.824A>G", "c.827A>C", "c.914+4A>G"} |
| 782 - 785 | GLDC:Glycine encephalopathy and non-ketoic hyperglycinemia, GLDC-related {(NM\_000170.2) "c.2405C>T", "c.2607C>A", "c.2T>C", "c.985C>A"} |
| 786 - 786 | GLRA1:Hyperekplexia, hereditary 1, autosomal dominant or recessive {(NM\_001146040.1) "c.298C>T"} |
| 787 - 787 | GMPPA:Alacrima, achalasia, and mental retardation syndrome {(NM\_013335.3) "c.1000A>C"} |
| 788 - 789 | GMPPB:Muscular dystrophy-dystroglycanopathy {(NM\_013334.3) "c.656T>C", "c.860G>A"} |
| 790 - 790 | GNE:Hereditary inclusion body myopathy (HIBM) {(NM\_005476.6) "c.2135T>C"} |
| 791 - 796 | GNPTAB:Mucolipidosis III alpha/beta {(NM\_024312.5) "c.118-2A>G", "c.2918dupT", "c.3434+1G>A", "c.3434+715G>A", "c.3503\_3504delTC", "c.3613C>T"} |
| 797 - 797 | GNPTG:Mucolipidosis III gamma {(NM\_032520.5) "c.499dupC"} |
| 798 - 798 | GPSM2:Chudley-McCullough syndrome {(NM\_013296.5) "c.379C>T"} |
| 799 - 799 | GRHPR:Hyperoxaluria, primary, type II {(NM\_012203.2) "c.975A>G"} |
| 800 - 807 | GUCY2D:Leber congenital amaurosis 1 , Cone-rod dystrophy 6 {(NM\_000180.3) "c.1992T>G", "c.2129C>T", "c.2513G>A", "c.2618C>G", "c.389delC", "c.529C>T", "c.620delC", "c.693delG"} |
| 808 - 808 | HACD1:Congenital myopathy {(NM\_014241.4) "c.744C>A"} |
| 809 - 809 | HADHA:Long-Chain hydroxylacyl-CoA dehydrogenase deficiency (LCHAD) {(NM\_000182.5) "c.1528G>C"} |
| 810 - 818 | HBB:Hemoglobipathies (Including sickle-cell anemia and beta thalassemia, Hb C, D, E, O) {(NM\_000518.5) "1.78\_Mb", "c.17\_18delCT", "c.20A>T", "c.25\_26delAA", "c.27dupG", "c.316-106C>G", "c.47G>A", "c.92+1G>A", "c.93-21G>A"} |
| 819 - 839 | HEXA:Tay-Sachs disease {(NM\_000520.5) "c.1073+1G>A", "c.1274\_1277dupTATC", "c.1351C>G", "c.1421+1G>C", "c.1444G>A", "c.316C>T", "c.459+2dupT", "c.496delC", "c.509G>A", "c.532C>T", "c.533G>A", "c.533G>T", "c.540C>G", "c.571-2A>G", "c.749G>A", "c.749G>T", "c.78G>A", "c.805+1G>A", "c.805G>A", "c.835T>C", "c.910\_912delTTC"} |
| 840 - 840 | HEXB:Sandhoff disease, infantile, juvenile, and adult forms {(NM\_000521) "c.1082+5G>A"} |
| 841 - 841 | HGD:Alkaptonuria {(NM\_000187) "c.16-272\_87+305del"} |
| 842 - 842 | HGSNAT:Retinitis pigmentosa 73 {(NM\_152419.3) "c.370A>T"} |
| 843 - 843 | HIKESHI:Leukodystrophy, early onset spastic paraparesis,acquired microcephaly, optic atrophy and risk of early death {(NM\_016401.4) "c.160G>C"} |
| 844 - 846 | HMGCL:HMG-CoA lyase deficiency {(NM\_000191.3) "c.122G>A", "c.125A>G", "c.521G>A"} |
| 847 - 847 | HOGA1:Hyperoxaluria, primary, type III {(NM\_138413) "c.944\_946delAGG"} |
| 848 - 850 | HPD:Thyrosinemia type III {(NM\_002150.3) "c.325-1G>A", "c.415-1G>A", "c.481G>C"} |
| 851 - 851 | HPS1:Hermansky-Pudlak syndrome 1 {(NM\_000195.5) "c.972delC"} |
| 852 - 855 | HPS3:Hermansky-Pudlak syndrome 3 {(NM\_032383.5) "c.1163+1G>A", "c.1691+2T>G", "c.2482-2A>G", "c.-2993\_217+690del3900"} |
| 856 - 856 | HPS6:Hermansky-Pudlak syndrome 6 {(NM\_024747.5) "c.1065dupG"} |
| 857 - 857 | HSPD1:Leukodystrophy, hypomyelinating, 4, HLD4 (HSP60) {(NM\_199440.1) "c.86A>G"} |
| 858 - 858 | IBA57:Spastic paraplegia 74, Autosomal Recessive {(NM\_001010867.4) "c.678A>G"} |
| 859 - 861 | IDUA:Mucopolysaccharidosis Type IH - Hurler syndrome {(NM\_000203.5) "c.1096A>C", "c.208C>T", "c.928C>T"} |
| 862 - 863 | IGHMBP2:Neuronopathy, distal hereditary motor, type VI {(NM\_002180.2) "c.114delA", "c.707T>G"} |
| 864 - 864 | IL10RA:Inflammatory bowel disease 28, early onset, autosomal recessive {(NM\_001558) "c.537G>A"} |
| 865 - 867 | INSR:Leprechaunism, Donohue syndrome {(NM\_000208.4) "c.167T>C", "c.3079C>T", "c.857G>A"} |
| 868 - 868 | INVS:Nephronophthisis 2, infantile {(NM\_014425.5) "c.2719C>T"} |
| 869 - 869 | ISPD:Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), {(NM\_001101426.4) "c.165dupG"} |
| 870 - 872 | ITGA2B:Glanzmann thrombasthenia, ITGA2B-related {(NM\_000419.4) "c.1947-1G>A", "c.818G>A", "c.97A>G"} |
| 873 - 874 | ITGB3:Glanzmann thrombasthenia, ITGB3-related {(NM\_000212.2) "11.2kbincl.ex.10-partex.13", "c.428T>G"} |
| 875 - 875 | ITGB4:Epidermolysis bullosa, junctional, with pyloric atresia - Carmi syndrome {(NM\_000213.5) "c.3224\_3793+120del"} |
| 876 - 876 | IVD:Isovaleric academia {(NM\_002225.5) "c.941C>T"} |
| 877 - 877 | KCNJ10:SESAME syndrome {(NM\_002241.5) "c.524G>A"} |
| 878 - 878 | KLHL40:Nemaline myopathy 8, Autosomal Recessive {(NM\_152393.4) "c.581T>A"} |
| 879 - 879 | KRT14:Epidermolysis bullosa simplex {(NM\_000526) "c.915G>A"} |
| 880 - 884 | LAMA2:Muscular dystrophy, congenital, due to partial LAMA2 deficiency {(NM\_000426.3) "c.3718C>T", "c.5260delG", "c.828C>G", "c.8665G>A", "c.8689C>T"} |
| 885 - 888 | LAMA3:Laryngoonychocutaneous Syndrome {(NM\_000227.4) "c.1981C>T", "c.2975delA", "c.4815G>T", "c.893\_894insT"} |
| 889 - 898 | LAMB3:Epidermolysis bullosa, junctional, non-Herlitz type {(NM\_000228.3) "c.124C>T", "c.1295dupA", "c.1903C>T", "c.1978C>T", "c.2528delA", "c.2914C>T", "c.3024delT", "c.3247C>T", "c.430C>T", "c.727C>T"} |
| 899 - 900 | LAMC2:Epidermolysis bullosa, junctional, Herlitz type {(NM\_018891.2) "c.1756C>T", "c.368\_373delinsACCAC"} |
| 901 - 905 | LCA5:Leber congenital amaurosis 5 {(NM\_181714.3) "c.1062\_1068delCGAAAAC", "c.1714C>T", "c.238C>T", "c.835C>T", "c.94delT"} |
| 906 - 907 | LIFR:Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome-LIFR related {(NM\_002310.5) "c.1601-1G>A", "c.2472\_2476delTATGT"} |
| 908 - 909 | LIPA:Wolman disease {(NM\_001127605.2) "c.260G>T", "c.398delC"} |
| 910 - 910 | LOXHD1:Deafness, Autosomal Recessive 77 {(NM\_144612.6) "c.4714C>T"} |
| 911 - 911 | LRBA:Immunodeficiency, common variable, 8, with autoimmunity {(NM\_001199282.2) "c.7937T>G"} |
| 912 - 912 | MAK:Retinitis pigmentosa 62 {(NM\_001242957.2) "c.497G>A"} |
| 913 - 913 | MAN1B1:Mental retardation, Autosomal Recessive 15 {(NM\_016219.5) "c.1863G>A"} |
| 914 - 914 | MATN3:Spondyloepimetaphyseal dysplasia {(NM\_002381.5) "c.910T>A"} |
| 915 - 915 | MCIDAS:Mucociliary clearance disorder {(NM\_001190787.2) "c.1142G>A"} |
| 916 - 919 | MCOLN1:Mucolipidosis type IV - ML4 {(NM\_020533.3) "c.-1015\_788del6433", "c.1207C>T", "c.406-2A>G", "c.964C>T"} |
| 920 - 920 | MED17:Microcephaly, postnatal progressive, with seizures and brain atrophy ((ICCA) {(NM\_004268.5) "c.1112T>C"} |
| 921 - 921 | MED25:Basel-Vanagaite-Smirin-Yosef syndrome {(NM\_030973.3) "c.116A>G"} |
| 922 - 922 | MEGF10:Myopathy, areflexia, respiratory distress, and dysphagia, early-onset {(NM\_001256545.2) "c.1325delC"} |
| 923 - 923 | MFSD8:Ceroid lipofuscinosis, neuronal, 7 {(NM\_152778.2) "c.472G>A"} |
| 924 - 924 | MKS1:Meckel syndrome 1 {(NM\_017777.3) "c.1048C>T"} |
| 925 - 927 | MLC1:Megalencephalic leukoencephalopathy with subcortical cysts {(NM\_015166.3) "c.176G>A", "c.274C>T", "c.278C>T"} |
| 928 - 928 | MLPH:Griscelli syndrome, type 3 {(NM\_024101.7) "c.103C>T"} |
| 929 - 929 | MMACHC:Methylmalonic aciduria and homocystinuria, cblC type {(NM\_015506.3) "c.271dupA"} |
| 930 - 931 | MOCS1:Molybdenum cofactor deficiency A {(NM\_001075098.3) "c.722delT", "c.971G>A"} |
| 932 - 933 | MOCS2:Molybdenum cofactor deficiency Type B {(NM\_004531.5) "c.226G>A", "c.377+1G>A"} |
| 934 - 936 | MPDU1:Congenital disorder of glycosylation, type If {(NM\_004870.4) "c.218G>A", "c.2T>C", "c.356T>C"} |
| 937 - 940 | MPL:Thrombocytopenia, congenital amegakaryocytic {(NM\_005373.2) "c.1031T>A", "c.127C>T", "c.460T>C", "c.79+2T>A"} |
| 941 - 941 | MPV17:Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) {(NM\_002437.5) "c.278A>C"} |
| 942 - 942 | MRE11A:Ataxia Telangiectasia like disorder {(NM\_005591.3) "c.290A>G"} |
| 943 - 944 | MTHFR:Homocystinuria due to MTHFR deficiency {(NM\_005957.4) "c.16delA", "c.474A>T"} |
| 945 - 948 | MTTP:Abetalipoproteinemia ABL {(NM\_000253.3) "c.2212delT", "c.2593G>T", "c.307A>T", "c.62-2A>G"} |
| 949 - 949 | MUT:Methylmalonic acidemia, mut(0) type {(NM\_000255.4) "c.655A>T"} |
| 950 - 950 | MVK:Hyper-IgD syndrome {(NM\_000431.4) "c.1129G>A"} |
| 951 - 952 | MYBPC1:Lethal congenital contracture syndrome 4 {(NM\_002465.4) "c.556G>A", "c.952C>T"} |
| 953 - 954 | MYH2:Proximal myopathy and ophthalmoplegia {(NM\_017534.6) "c.2400delG", "c.706G>A"} |
| 955 - 959 | MYO15A:Deafness, Autosomal Recessive 3 {(NM\_016239.4) "c.373\_374delCG", "c.4240G>A", "c.7207G>T", "c.8183G>A", "c.8467G>A"} |
| 960 - 978 | MYO7A:Usher syndrome, type 1B {(NM\_000260.4) "c.1190C>A", "c.1996C>T", "c.2065delC", "c.2187+1G>A", "c.2307delC", "c.2476G>A", "c.2777T>A", "c.29T>C", "c.3892G>A", "c.4153-2A>G", "c.470+1G>A", "c.5068dupC", "c.5581C>T", "c.5660C>T", "c.6196delC", "c.620A>G", "c.640G>A", "c.6487G>A", "c.700C>T"} |
| 979 - 979 | NAGLU:Mucopolysaccharidosis type IIIB (Sanfilippo B) {(NM\_000263.4) "c.2021G>A"} |
| 980 - 980 | NARS2:Combined oxidative phosphorylation deficiency 24 (COXPD24) {(NM\_024678) "c.500A>G"} |
| 981 - 982 | NBEAL2:Gray platelet syndrome {(NM\_015175.2) "c.2701C>T", "c.5413dupG"} |
| 983 - 984 | NCF1:Chronic granulomatous disease due to deficiency of NCF-1 {(NM\_000265.6) "c.153+1G>A", "c.579G>A"} |
| 985 - 987 | NCF2:Chronic granulomatous disease due to deficiency of NCF-2 {(NM\_000433.3) "c.1171\_1175delAAGCT", "c.196C>T", "c.304C>T"} |
| 988 - 988 | NDUFA11:Mitochondrial complex I deficiency - NDUFA11 gene {(NM\_001193375.1) "c.97+5G>A"} |
| 989 - 989 | NDUFAF5:Mitochondrial complex I deficiency - NDUFAF5 gene {(NM\_024120.5) "c.749G>T"} |
| 990 - 990 | NDUFS2:Mitochondrial complex I deficiency-NDUFS2 gene {(NM\_004550.4) "c.1237T>C"} |
| 991 - 991 | NDUFS4:Leigh syndrome {(NM\_002495.4) "c.462delA"} |
| 992 - 992 | NDUFS6:Mitochondrial complex I deficiency - NDUFS6 gene {(NM\_004553.4) "c.344G>A"} |
| 993 - 996 | NEB:Nemaline myopathy 2 {(NM\_001271208.2) "c.17118+1G>A", "c.18808C>T", "c.9619-2A>G" | (NM\_004543.4) "c.7431+1917\_7536+372del"} |
| 997 - 997 | NGLY1:Congenital disorder of deglycosylation {(NM\_018297.4) "c.1294G>T"} |
| 998 - 998 | NNT:Glucocorticoid deficiency 4 {(NM\_182977.3) "c.598G>A"} |
| 999 - 1016 | NPC1:Niemann-Pick disease type C1 {(NM\_000271.5) "c.1211G>A", "c.1241\_1242delTC", "c.1437\_1442delCACCAT", "c.1552C>T", "c.1761delT", "c.2279\_2281delTCT", "c.2780C>T", "c.2972\_2973delAG", "c.2974G>A", "c.2974G>C", "c.2974G>T", "c.3007C>T", "c.3347\_3348delTC", "c.3467A>G", "c.3557G>A", "c.3614C>A", "c.3637T>G", "c.3673T>G"} |
| 1017 - 1025 | NPHS1:Nephrotic syndrome type 1 {(NM\_004646.3) "c.1138C>T", "c.121\_122delCT", "c.1707C>G", "c.2104G>A", "c.2160dupC", "c.3325C>T", "c.3478C>T", "c.514\_516delACC", "c.532C>T"} |
| 1026 - 1026 | NPHS2:Nephrotic syndrome {(NM\_014625.3) "c.412C>T"} |
| 1027 - 1029 | NTRK1:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM\_001012331.1) "c.1842\_1843insT", "c.2066C>T", "c.207\_208delTG"} |
| 1030 - 1030 | NUP62:Striatonigral degeneration, Infantile Bilateral Striatal Necrosis (IBSN) {(NM\_016553.4) "c.1172A>C"} |
| 1031 - 1031 | OCA2:Albinism, oculocutaneous, type II {(NM\_000275.3) "c.1327G>A"} |
| 1032 - 1032 | OPA3:3-methylglutaconic aciduria, type III - Costeff {(NM\_025136.3) "c.143-1G>C"} |
| 1033 - 1035 | OTC:Ornithine transcarbamylase deficiency {(NM\_000531.6) "c.717+1G>T", "c.829C>T", "c.958C>T"} |
| 1036 - 1036 | OTOA:Deafness, Autosomal Recessive 22 {(NM\_144672) "c.1025A>T"} |
| 1037 - 1037 | OTOF:Deafness, Autosomal Recessive 9 {(NM\_194248.2) "c.2866+1G>A"} |
| 1038 - 1055 | PAH:Phenylketonuria {(NM\_000277.3) "c.1045T>C", "c.1066-11G>A", "c.1208C>T", "c.1222C>T", "c.1315+1G>A", "c.143T>C", "c.165delT", "c.165T>G", "c.441+5G>T", "c.473G>A", "c.689T>C", "c.722G>A", "c.727C>T", "c.754C>T", "c.782G>A", "c.782G>C", "c.842C>T", "c.898G>T"} |
| 1056 - 1056 | PCCA:Propionic acidemia, PCCA-related {(NM\_000282.4) "c.923dupT"} |
| 1057 - 1057 | PCCB:Propionic acidemia, PCCB-related {(NM\_000532.5) "c.1173dupT"} |
| 1058 - 1059 | PCDH12:Microcephaly, seizures, spasticity, and brain calcification (MISSBC) {(NM\_016580) "c.2515C>T", "c.995delT"} |
| 1060 - 1060 | PCDH15:Usher syndrome, type 1F {(NM\_033056.3) "c.733C>T"} |
| 1061 - 1061 | PCK1:Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency {(NM\_002591.4) "c.134T>C"} |
| 1062 - 1062 | PCNT:Microcephalic osteodysplastic primordial dwarfism type II (MOPDII) {(NM\_006031.5) "c.3465-1G>A"} |
| 1063 - 1063 | PDE6G:Retinitis pigmentosa 57 {(NM\_002602.4) "c.187+1G>T"} |
| 1064 - 1066 | PEPD:Prolidase deficiency {(NM\_000285.4) "c.1103T>G", "c.605C>T", "c.634G>C"} |
| 1067 - 1068 | PEX1:Peroxisome biogenesis disorder 1A (Zellweger) {(NM\_000466.3) "c.2097dupT", "c.2528G>A"} |
| 1069 - 1071 | PEX2:Peroxisome biogenesis disorder 5A (Zellweger) {(NM\_001079867.1) "c.355C>T", "c.550delC", "c.669G>A"} |
| 1072 - 1075 | PEX6:Peroxisome biogenesis disorder 4B (Zellweger syndrome) {(NM\_000287.4) "c.1715C>T", "c.1947delG", "c.2094+2T>C", "c.2534T>C"} |
| 1076 - 1076 | PEX7:Rhizomelic chondrodysplasia punctata type 1 {(NM\_000288.4) "c.283T>G"} |
| 1077 - 1077 | PGAP3:Hyperphosphatasia with mental retardation syndrome 4 {(NM\_033419.5) "c.845A>G"} |
| 1078 - 1078 | PHGDH:Phosphoglycerate dehydrogenase deficiency {(NM\_006623.3) "c.1468G>A"} |
| 1079 - 1079 | PHKG2:Glycogen storage disease IXc {(NM\_000294.3) "c.71A>G"} |
| 1080 - 1080 | PHYH:Refsum disease {(NM\_001037537.1) "c.523C>T"} |
| 1081 - 1082 | PIGN:Multiple congenital anomalies-hypotonia-seizures syndrome 1 {(NM\_012327.5) "c.2126G>A", "c.755A>T"} |
| 1083 - 1084 | PIGT:Multiple congenital anomalies-hypotonia-seizures syndrome 3 {(NM\_015937.6) "c.1564T>G", "c.761delG"} |
| 1085 - 1085 | PIP5K1C:Lethal congenital contractural syndrome 3 {(NM\_012398.2) "c.757G>A"} |
| 1086 - 1086 | PJVK:Deafness, Autosomal Recessive 59 {(NM\_001042702.4) "c.406C>T"} |
| 1087 - 1092 | PKHD1:Polycystic kidney & hepatic disease, PKHD1-related {(NM\_138694.4) "c.107C>T", "c.1350delC", "c.2279G>A", "c.3761\_3762delCCinsG", "c.6122-12G>A", "c.824C>T"} |
| 1093 - 1096 | PLA2G6:Infantile neuroaxonal dystrophy 1 (INAD) {(NM\_003560.4) "c.1040G>C", "c.2070\_2072delTGT", "c.2251G>A", "c.668C>A"} |
| 1097 - 1097 | PLAA:Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies {(NM\_001031689.3) "c.2254C>T"} |
| 1098 - 1101 | PMM2:Congenital disorder of glycosylation Ia {(NM\_000303) "c.338C>T", "c.357C>A", "c.422G>A", "c.691G>A"} |
| 1102 - 1102 | POC1A:Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis {(NM\_015426.5) "c.512T>C"} |
| 1103 - 1103 | POR:Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis {(NM\_000941.3) "c.1615G>A"} |
| 1104 - 1104 | PPIB:Osteogenesis imperfecta, type IX {(NM\_000942.4) "c.563\_566delACAG"} |
| 1105 - 1105 | PPP1R13L:Cardio-Cutaneous Syndrome DCM {(NM\_006663.4) "c.2241C>G"} |
| 1106 - 1106 | PPT1:Ceroid lipofuscinosis, neuronal, 1 {(NM\_000310.3) "c.169dupA"} |
| 1107 - 1107 | PRCD:Retinitis pigmentosa 36 {(NM\_001077620) "c.64C>T"} |
| 1108 - 1108 | PRICKLE1:Epilepsy, progressive myoclonic 1B {(NM\_153026.3) "c.311G>A"} |
| 1109 - 1109 | PUS1:Mitochondrial myopathy and sideroblastic anemia 1 {(NM\_001002020.2) "c.346C>T"} |
| 1110 - 1111 | RAG1:Severe combined immudeficiency, B cell-negative, RAG1-related {(NM\_000448.2) "c.1361T>A", "c.1410\_1413delCTTG"} |
| 1112 - 1116 | RAG2:Severe combined immudeficiency, B cell-negative, RAG2-related {(NM\_000536.3) "c.1438G>T", "c.193G>T", "c.379A>T", "c.470G>T", "c.685C>T"} |
| 1117 - 1120 | RAPSN:Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency {(NM\_005055.5) "c.-210A>G", "c.264C>A", "c.-27C>G", "c.672\_673insACT"} |
| 1121 - 1121 | RARS2:Pontocerebellar hypoplasia, type 6 {(NM\_020320.5) "c.110+5A>G"} |
| 1122 - 1129 | RDH12:Leber congenital amaurosis 13 {(NM\_152443.3) "c.146C>T", "c.164C>T", "c.295C>A", "c.377C>T", "c.481C>T", "c.658+1G>A", "c.716G>A", "c.740T>C"} |
| 1130 - 1131 | RECQL2:Werner syndrome {(NM\_000553.5) "c.1105C>T", "c.2665C>T"} |
| 1132 - 1132 | RFX5:Bare lymphocyte syndrome, type II (SCID) {(NM\_000449) "c.715C>T"} |
| 1133 - 1133 | RIN2:Macrocephaly, alopecia, cutis laxa, and scoliosis {(NM\_018993.3) "c.1731delC"} |
| 1134 - 1134 | RNASEH2B:Aicardi-Goutieres syndrome 2 {(NM\_024570.3) "c.529G>A"} |
| 1135 - 1135 | ROGDI:Kohlschutter-Tonz syndrome {(NM\_024589.2) "c.469C>T"} |
| 1136 - 1136 | RP1:Retinitis pigmentosa 1 {(NM\_006269.2) "c.4941dupT"} |
| 1137 - 1142 | RPE65:Leber congenital amaurosis 2 {(NM\_000329.3) "c.1301C>G", "c.227A>C", "c.361dupT", "c.722A>T", "c.886dupA", "c.95-2A>T"} |
| 1143 - 1143 | RPGRIP1L:Meckel syndrome 5 {(NM\_015272.5) "c.118C>T"} |
| 1144 - 1144 | RSPH9:Ciliary dyskinesia, primary, 12 {(NM\_152732.5) "c.804\_806delGAA"} |
| 1145 - 1147 | RTEL1:Dyskeratosis congenita {(NM\_001283009.1) "c.3791G>A" | (NM\_032957.4) "c.1548G>T", "c.2992C>T"} |
| 1148 - 1148 | RYR1:Minicore myopathy with external ophthalmoplegia {(NM\_000540.2) "c.9623C>T"} |
| 1149 - 1150 | SAMD9:Tumoral calcinosis, familial, normophosphatemic {(NM\_017654.4) "c.1030C>T", "c.4483A>G"} |
| 1151 - 1154 | SAMHD1:Aicardi Goutieres syndrome {(NM\_015474.3) "9.1-KB\_DEL", "c.1106T>C", "c.649\_650insG", "c.676C>G"} |
| 1155 - 1155 | SARS2:Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis {(NM\_017827.3) "c.1169A>G"} |
| 1156 - 1156 | SCAPER:Retinitis pigmentosa with intellectual disability {(NM\_020843) "c.2806delC"} |
| 1157 - 1157 | SCN9A:Insensitivity to pain, congenital (CIPA) {(NM\_002977.3) "c.2687G>A"} |
| 1158 - 1158 | SCN9A:Insensitivity to pain, congenital {(NM\_002977.3) "c.1124delG"} |
| 1159 - 1159 | SDHA:Cardiomyopathy, dilated , 1GG neonatal isolated {(NM\_004168.4) "c.1664G>A"} |
| 1160 - 1161 | SEC23B:Dyserythropoietic anemia, congenital, type II {(NM\_006363.6) "c.2129C>T", "c.325G>A"} |
| 1162 - 1163 | SepSecS:Pontocerebellar hypoplasia type 2D {(NM\_016955.4) "c.1001A>G", "c.715G>A"} |
| 1164 - 1166 | SERAC1:3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome {(NM\_032861.4) "c.1018delT", "c.128+4A>G", "c.698\_699delinsAGTATA"} |
| 1167 - 1167 | SGCG:Muscular dystrophy, limb-girdle, type 2C {(NM\_000231.2) "c.525delT"} |
| 1168 - 1173 | SGSH:Mucopolysaccharidisis type IIIA (Sanfilippo A) {(NM\_000199.5) "c.1093C>T", "c.1298G>A", "c.332T>C", "c.416C>T", "c.544C>T", "c.812C>T"} |
| 1174 - 1174 | SLC12A3:Bartter and Gitelman syndrome {(NM\_000339.3) "c.1313G>A"} |
| 1175 - 1175 | SLC17A5:Sialic acid storage disorder, infantile (ISSD) {(NM\_012434.5) "c.983G>A"} |
| 1176 - 1177 | SLC19A2:Thiamine-responsive megaloblastic anemia syndrome {(NM\_006996.3) "c.1223+1G>A", "c.725delC"} |
| 1178 - 1180 | SLC1A4:Spastic tetraplegia, thin corpus callosum, and progressive microcephaly {(NM\_003038.5) "c.1369C>T", "c.766G>A", "c.944\_945del"} |
| 1181 - 1181 | SLC22A5:Carnitine deficiency, systemic primary {(NM\_003060.3) "c.1196G>A"} |
| 1182 - 1183 | SLC25A1:Combined D-2- and L-2-hydroxyglutaric aciduria {(NM\_005984.5) "c.389G>A", "c.845G>A"} |
| 1184 - 1185 | SLC25A20:Carnitine-acylcarnitine translocase deficiency - CACT {(NM\_000387.6) "c.609-3C>G", "c.713A>G"} |
| 1186 - 1186 | SLC26A3:Congenital chloride diarhhea (CLD) {(NM\_000111.2) "c.559G>T"} |
| 1187 - 1197 | SLC26A4:Pendred syndrome {(NM\_000441.2) "c.1001G>T", "c.1151A>G", "c.1198delT", "c.1246A>C", "c.1341+1delG", "c.1458dupT", "c.2000T>G", "c.2168A>G", "c.349C>T", "c.707T>C", "c.716T>A"} |
| 1198 - 1200 | SLC29A3:Histiocytosis-lymphadenopathy plus syndrome {(NM\_018344.6) "c.1045delC", "c.1279G>A", "c.1309G>A"} |
| 1201 - 1203 | SLC2A2:Fanconi-Bickel syndrome {(NM\_000340.2) "c.372A>C", "c.734A>C", "c.901C>T"} |
| 1204 - 1204 | SLC30A9:Birk-Landau-Perez cerebro-renal syndrome {(NM\_016474.5) "c.1047\_1049delCAG"} |
| 1205 - 1206 | SLC35A3:Arthrogryposis, mental retardation, and seizures {(NM\_012243.3) "c.514C>T", "c.886A>G"} |
| 1207 - 1207 | SLC35C1:Congenital disorder of glycosylation, type IIc {(NM\_018389.4) "c.923C>G"} |
| 1208 - 1211 | SLC37A4:Glycogen storage disease Ib {(NM\_001164277.1) "c.1042\_1043delCT", "c.1179G>A", "c.446G>A", "c.83G>A"} |
| 1212 - 1212 | SLC39A4:Acrodermatitis enteropathica {(NM\_130849.3) "c.1224delC"} |
| 1213 - 1213 | SLC4A4:Renal tubular acidosis (RTA), proximal, with ocular abnormalities and mental retardation {(NM\_003759.3) "c.2321G>A"} |
| 1214 - 1214 | SLCO2A1:Hypertrophic osteoarthropathy, primary, Autosomal Recessive 2 {(NM\_005630.2) "c.1292delC"} |
| 1215 - 1216 | SMARCAL1:Schimke immunoosseous dysplasia {(NM\_014140.3) "c.2542G>T", "c.863-2A>G"} |
| 1217 - 1217 | SMN1:Spinal muscular atrophy-1 {(NM\_000344) "c.835\_\*3del"} |
| 1218 - 1228 | SMPD1:Niemann-Pick disease type B, SMPD1-related {(NM\_000543.5) "c.1493G>T", "c.1705T>C", "c.1758T>G", "c.1805G>C", "c.1828\_1830delCGC", "c.442T>A", "c.573delT", "c.730G>A", "c.911T>C", "c.96G>A", "c.996delC"} |
| 1229 - 1229 | SNAP29:Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome - CEDNIK Syndrome {(NM\_004782) "c.223delG"} |
| 1230 - 1230 | SNX10:Osteopetrosis, Autosomal Recessive 8 {(NM\_001199835.1) "c.152G>A"} |
| 1231 - 1233 | SPG11:Spastic paraplegia 11, Autosomal Recessive {(NM\_025137.4) "c.118C>T", "c.2471dupT", "c.4339C>T"} |
| 1234 - 1237 | SPINK5:Netherton syndrome {(NM\_001127698.1) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC"} |
| 1238 - 1238 | ST3GAL3:Early infantile epileptic encephalopathy 15 {(NM\_006279.4) "c.958G>C"} |
| 1239 - 1239 | STRA6:Microphthalmia {(NM\_001142617.1) "c.1678G>C"} |
| 1240 - 1241 | STRC:Deafness, Autosomal Recessive 16 {(NM\_153700.2) "c.4171C>G", "EX7\_EX29DEL"} |
| 1242 - 1243 | SUMF1:Multiple sulfatase deficiency {(NM\_182760.3) "c.1043C>T", "c.463T>C"} |
| 1244 - 1244 | SYNE4:Deafness, Autosomal Recessive 76 {(NM\_001039876.3) "c.228\_229delAT"} |
| 1245 - 1245 | SZT2:Epileptic encephalopathy, early infantile, 18 {(NM\_015284.3) "c.73C>T"} |
| 1246 - 1246 | TAF2:Mental retardation, Autosomal Recessive 40 {(NM\_003184.4) "c.557C>G"} |
| 1247 - 1247 | TBCD:Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM\_005993.4) "c.1423G>A"} |
| 1248 - 1249 | TBCE:Hypoparathyroidism retardation dysmorphism syndrome {(NM\_003193.5) "c.155\_166delGCCACGAAGGGA", "c.354\_355del"} |
| 1250 - 1250 | TBX19:Adrenocorticotropic hormone deficiency {(NM\_005149.3) "c.574\_577delATAG"} |
| 1251 - 1254 | TCIRG1:Osteopetrosis, Autosomal Recessive 1 {(NM\_006019.4) "c.117+4A>T", "c.1331G>T", "c.1384\_1386delAAC", "c.674delG"} |
| 1255 - 1255 | TCTN2:Meckel syndrome 8 {(NM\_024809.5) "c.1506-2A>G"} |
| 1256 - 1258 | TECPR2:Spastic paraplegia 49, Autosomal Recessive {(NM\_001172631.2) "c.1319delT", "c.3416delT", "c.566C>T"} |
| 1259 - 1259 | THG1L:Cerebellar ataxia and developmental delay {(NM\_017872.5) "c.164T>C"} |
| 1260 - 1262 | TK2:Mitochondrial DNA depletion syndrome 2 (myopathic type) {(NM\_004614.5) "c.360\_361delGCinsAA", "c.361C>A", "c.635T>A"} |
| 1263 - 1267 | TMC1:Deafness, Autosomal Recessive 7 {(NM\_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"} |
| 1268 - 1270 | TMEM216:Joubert syndrome 2 (MKS2) {(NM\_001173990.3) "c.218G>A", "c.218G>T", "c.230G>C"} |
| 1271 - 1271 | TMEM231:Meckel syndrome 11 {(NM\_001077418.3) "c.664+4A>G"} |
| 1272 - 1272 | TMEM38B:Osteogenesis imperfecta, type XIV {(NM\_018112) "c.455\_542del"} |
| 1273 - 1275 | TMEM67:Joubert syndrome type 6 (MSK3) {(NM\_153704) "c.1065+1delG" | (NM\_153704.5) "c.1975C>T", "c.725A>G"} |
| 1276 - 1277 | TMEM70:ATPase deficiency, nuclear encoded {(NM\_017866.6) "c.238C>T", "c.316+1G>T"} |
| 1278 - 1278 | TMPRSS3:Deafness, Autosomal Recessive 8/10 {(NM\_024022.2) "c.989delA"} |
| 1279 - 1279 | TRAPPC9:Mental retardation, Autosomal Recessive 13 {(NM\_031466.7) "c.1423C>T"} |
| 1280 - 1280 | TRIM32:Bardet-Biedl syndrome 11 {(NM\_012210) "c.388C>T"} |
| 1281 - 1282 | TRMT10A:Microcephaly, short stature, and impaired glucose metabolism {(NM\_152292.4) "c.616G>A", "c.727C>T"} |
| 1283 - 1283 | TRMU:LIFT, Liver failure infantile transient {(NM\_018006.5) "c.229T>C"} |
| 1284 - 1284 | TRPM1:Night blindness, congenital stationary (complete), 1C, Autosomal Recessive {(NM\_002420.5) "36.4-KB\_DEL,\_EX2-7"} |
| 1285 - 1286 | TRPM6:Hypomagnesemia 1, intestinal {(NM\_017662.5) "c.1010+5G>C", "c.2009+1G>A"} |
| 1287 - 1287 | TSHR:Hypothyroidism, congenital, nongoitrous, 1 {(NM\_000369.2) "c.1825C>T"} |
| 1288 - 1289 | TTN:Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis {(NM\_003319.4) "c.58881dupA" | (NM\_133432) "c.36122delC"} |
| 1290 - 1291 | TULP1:Retinitis pigmentosa 14 {(NM\_003322.6) "c.1349G>A", "c.1495+2dupT"} |
| 1292 - 1293 | TYMP:Mitochondrial DNA depletion syndrome 1 (MNGIE type) {(NM\_001113755.2) "c.433G>A", "c.866A>C"} |
| 1294 - 1309 | TYR:Albinism, oculocutaneous, type IA (OCA1A) {(NM\_000372.5) "c.1037-1G>A", "c.1037-7T>A", "c.1118C>A", "c.1204C>T", "c.1217C>T", "c.1357C>T", "c.140G>A", "c.149C>G", "c.454C>T", "c.649C>T", "c.649delC", "c.74dupT", "c.757G>A", "c.832C>T", "c.880G>A", "c.896G>A"} |
| 1310 - 1310 | UNC13D:Hemophagocytic lymphohistiocytosis, familial, 3 {(NM\_199242.2) "c.753+1G>T"} |
| 1311 - 1311 | UNC80:Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM\_032504.1) "c.151C>T"} |
| 1312 - 1312 | UQCRQ:Mitochondrial complex III deficiency, nuclear type 4 {(NM\_014402.5) "c.134C>T"} |
| 1313 - 1315 | USH1C:Usher syndrome, type 1C {(NM\_005709.3) "c.1220delG", "c.238dupC", "c.497-2delA"} |
| 1316 - 1339 | USH2A:Usher syndrome, type 2A {(NM\_206933.3) "c.1000C>T", "c.10211delC", "c.12052G>A", "c.12067-2A>G", "c.12575G>A", "c.13274C>T", "c.14020A>G", "c.14023A>T", "c.14413G>A", "c.14424C>A", "c.2167+5G>A", "c.2209C>T", "c.236\_239dupGTAC", "c.3368A>G", "c.377delG", "c.3959C>T", "c.4544C>T", "c.5519G>T", "c.6159delA", "c.6937G>T", "c.802G>A", "c.8558+1G>T", "c.8719A>C", "c.9685delG"} |
| 1340 - 1341 | VDR:Rickets, vitamin D-resistant, type IIA {(NM\_001017535.1) "c.277+1G>T", "c.885C>A"} |
| 1342 - 1342 | VPS11:Hypomyelination and developmental delay {(NM\_021729.5) "c.2536T>G"} |
| 1343 - 1344 | VPS13A:Choreoacanthocytosis {(NM\_033305.3) "c.6059delC", "delexon70-73"} |
| 1345 - 1345 | VPS13B:Cohen syndrome {(NM\_017890.4) "c.6732+1G>A"} |
| 1346 - 1347 | VPS33B:Arthrogryposis renal dysfunction and cholestasis (ARC) syndrome {(NM\_018668.4) "c.403+1G>A", "c.700G>C"} |
| 1348 - 1348 | VPS37A:Spastic paraplegia 53, Autosomal Recessive {(NM\_152415.3) "c.1146A>T"} |
| 1349 - 1349 | VPS45:Neutropenia, severe congenital, 5, Autosomal Recessive {(NM\_007259.5) "c.671C>A"} |
| 1350 - 1351 | VPS53:Pontocerebellar hypoplasia, type 2E (PCCA2) {(NM\_001128159.3) "c.1556+5G>A", "c.2084A>G"} |
| 1352 - 1352 | VRK1:Pontocerebellar hypoplasia type 1A {(NM\_003384.3) "c.1072C>T"} |
| 1353 - 1353 | WISP3:Arthropathy, progressive pseudorheumatoid, of childhood {(NM\_003880.3) "c.536\_537delGT"} |
| 1354 - 1354 | XPC:Xeroderma pigmentosum, group C {(NM\_004628.4) "c.566\_567delAT"} |
| 1355 - 1355 | ZBTB24:Immunodeficiency-centromeric instability-facial anomalies syndrome-2 {(NM\_014797.2) "c.501dupA"} |
| 1356 - 1357 | ZNF469:Brittle cornea syndrome 1 {(NM\_001127464.2) "c.5943delA", "c.9531delG"} |