רשימת מחלות ומוטציות בפאנל MOH09.2024 v2.10 (2024-11-12)
Hybrid Capture-Based Next Generation Sequencing

Mutation #	Gene: Disease Name {(Transcsript) "Mutation name"}
1	ABCA4:CONE-ROD DYSTROPHY 3; CORD3/ RETINITIS PIGMENTOSA 19; RP19/STARGARDT DISEASE 1; STGD1 {(NM_000350.3) "c.834delT"}
2 - 2	ABCB11:Progressive Familial Intrahepatic Cholestasis (PFIC) type II {(NM_003742.4) "c.3268C>T"}
3 - 5	ABCC8:Hyperinsulinemic hypoglycemia, familial, 1 (PHHI/HHF1) {(NM_000352.6) "c.2506C>T", "c.3989-9G>A", "c.4160_4162delTCT"}
6 - 8	ACADVL:ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD {(NM_000018.4) "c.1096C>T", "c.65C>A", "c.799_802delGTTA"}
9 - 9	ACP5:Spondyloenchondrodysplasia with immune dysregulation {(NM_016237.5) "c.325G>A"}
10 - 11	ACSF3:Combined Malonic and Methylmalonic aciduria {(NM_001243279.3) "c.1411C>T", "c.1412G>A"}
12 - 12	ADAMTS2:ADAMTS2:Ehlers Danlos syndrome, type VIIC {(NM_014244.5) "c.673C>T"}
13 - 13	ADGRG1:BFPP-Bilateral frontoparietal polymicrogyria {(NM_201525.4) "c.1167+3G>C"}
14 - 15	AGL:AGL:Glycogen storage disease III {(NM_000642.3) "c.3911dupA", "c.4455delT"}
16 - 22	AGXT:Hyperoxaluria type 1 {(NM_000030.3) "c.121G>A", "c.26C>A", "c.33_34insC", "c.358+1G>A", "c.584T>G", "c.865C>T", "c.997A>T"}
23 - 23	AHSG:Alopecia-mental retardation syndrome 1, Autosomal recessive {(NM_001622.4) "c.950G>A"}
24 - 24	AIMP1:LEUKODYSTROPHY, HYPOMYELINATING, 3; HLD3 {(NM_001142416.2) "c.292_293del"}
25 - 26	AIRE:APECED/APS1 {(NM_000383.4) "c.254A>G", "c.47C>T"}
27 - 28	ALDOB:ALDOB:Fructose intolerance {(NM_000035.4) "c.448G>C", "c.524C>A"}
29 - 30	ALPL:Hypophosphatasia {(NM_000478.6) "c.1171C>T", "c.1348C>T"}
31 - 31	AMT:Non ketotic hyperglycinemia (NKH),GLYCINE ENCEPHALOPATHY 2; GCE2 {(NM_000481.4) "c.125A>G"}
32 - 32	AP4M1:Hereditary spastic paraplegia 50 {(NM_004722.4) "c.203del"}
33 - 33	AQP2:DIABETES INSIPIDUS, NEPHROGENIC, AUTOSOMAL (DIABETES INSIPIDUS, NEPHROGENIC, TYPE II) {(NM_000486.5) "c.83T>C"}
34 - 34	ARL6:BARDET BIEDL SYNDROME 3, BBS3 {(NM_001278293.3) "c.364C>T"}
35 - 35	ARNT2:Webb-Dattani syndrome (3), Autosomal recessive {(NM_014862.4) "c.1000C>T"}

36 - 42	ARSA:Metachromatic leukodystrophy - MLD {(NM_000487.6) "c.1136C>T", "c.211T>G", "c.292_293delTCinsCT", "c.465+1G>A", "c.47G>A", "c.542T>G", "c.827C>T"}
43 - 43	ASL:Argininosuccinic aciduria {(NM_000048.4) "c.346C>T"}
44 - 44	ASNS:Asparagine Synthetase Deficiency {(NM_001673.5) "c.1084T>G"}
45 - 47	ASPA:Canavan disease {(NM_000049.4) "c.693C>A", "c.854A>C", "c.914C>A"}
48 - 56	ATM:Ataxia-Telangiectasia; AT {(NM_000051.4) "c.103C>T", "c.1339C>T", "c.2284_2285delCT", "c.3245_3247delinsTGAT", "c.3576G>A", "c.368del", "c.497del17514bp", "c.6672_6680delGGCTCTACGinsCTC", "c.7241_7244delAAGC"}
57 - 72	ATP7B:Wilson disease {(NM_000053.4) "c.1340_1343delAAAC", "c.1544G>A", "c.1639delC", "c.1934T>G", "c.2293G>A", "c.2336G>A", "c.2337G>A", "c.2817G>T", "c.3191A>C", "c.3207C>A", "c.3451C>T", "c.3551T>C", "c.3649_3654delGTTCTG", "c.3659C>T", "c.3784G>T", "c.845delT"}
73 - 73	B4GALT1:CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IId; CDG2D {(NM_001497) "c.61C>T"}
74 - 77	BBS2:BARDET-BIEDL SYNDROME 2; BBS2 {(NM_031885.4) "c.1895G>C", "c.224T>G", "c.311A>C", "c.401C>G"}
78 - 79	BBS4:BARDET-BIEDL SYNDROME 4; BBS4 {(NM_033028.5) "c.884G>C", "exon 3-4 del"}
80 - 80	BCKDHA:Maple syrup urine disease, type Ia {(NM_000709.4) "c.859C>T"}
81 - 83	BCKDHB:Maple syrup urine disease, type Ib {(NM_000056.4) "c.1016C>T", "c.356T>G", "c.548G>C"}
84 - 85	BLM:Bloom syndrome {(NM_000057.4) "c.2207_2212delATCTGAinsTAGATTC", "c.2407dupT"}
86 - 86	BLOC1S6:Hermansky-pudlak syndrome {(NM_001311255.1) "c.318_320delinsAT"}
87 - 87	BSND:BARTTER SYNDROME, TYPE 4A {(NM_057176.3) "c.28G>A"}
88 - 88	BTD:Biotinidase deficiency {(NM_000060.4) "c.100G>A"}
89 - 89	C8ORF37:Retinitis pigmentosa 64 {(NM_177965.4) "c.545A>G"}
90 - 91	CAPN3:Muscular dystrophy, limb-girdle, autosomal recessive 1 {(NM_000070) "c.2257G>A", "c.367C>A"}
92 - 92	CASQ2:Ventricular tachycardia {(NM_001232.3) "c.919G>C"}
93 - 93	CC2D1A:Mental retardation non syndromic {(NM_017721.5) "IVS13-16DEL"}
94 - 94	CCDC174:HYPOTONIA, INFANTILE, WITH PSYCHOMOTOR RETARDATION; IHPMR {(NM_016474.5) "c.1404A>G"}
95 - 95	CDAN1:Anemia, congenital dyserythropoietic Type Ia {(NM_138477.4) "c.3124C>T"}
96 - 97	CDH23:Usher Syndrome Type ID {(NM_022124) "c.5749G>A" | (NM_022124.2) "c.7903G>T"}
98 - 98	CEP152:Primary microcephaly hereditary (MCPH) type 9

	{(NM_014985.3) "c.2281-2A>G"}
99 - 100	CEP290:Meckel-Gruber Syndrome type4 {(NM_025114.3) "c.1225delA", "c.1666delA"}
101 - 101	CERKL:CERKL:Retinitis pigmentosa 26 {(NM_001030311.2) "c.238+1G>A IVS1_G-A_+1"}
102 - 102	CFH:HEMOLYTIC UREMIC SYNDROME ATYPICAL/ COMPLEMENT FACTOR H DEFICIENCY; CFHD {(NM_000186.3) "c.3673_3696del"}
103 - 174	CFTR:Cystic fibrosis (CF) {(NM_000492.3) "c.349C>T" | (NM_000492.4) "c.1000C>T", "c.1001G>A", "c.1040G>C", "c.1079C>A/c.1075C>A", "c.1327G>T", "c.1364C>A", "c.1367T>C", "c.1418delG", "c.1466C>A", "c.1516_1518delATC", "c.1521_1523delCTT", "c.1545_1546delTA", "c.1558G>T", "c.1585-1G>A", "c.1624G>T", "c.1646G>T", "c.1647T>G", "c.1652G>A", "c.1657C>T", "c.1679G>C", "c.1680-1G>A", "c.1736A>G", "c.1766+1G>A", "c.2051_2052delAAinsG", "c.2052del", "c.2052dupA", "c.2125C>T", "c.254G>A", "c.2657+5G>A", "c.273+1G>A", "c.274G>T", "c.2834C>T", "c.2856G>C", "c.2988+1G>A", "c.2989-1G>A", "c.3205G>A", "c.3266G>A", "c.3276C>A", "c.3276C>G", "c.3299A>C", "c.3472C>T", "c.3484C>T", "c.3485G>T", "c.350G>A", "c.3528delC", "c.3587C>G", "c.358G>A", "c.3700A>G", "c.3717+12191C>T", "c.3731G>A", "c.3764C>A", "c.3773dupT", "c.3846G>A", "c.3883_3886delATTT", "c.3909C>G", "c.4046G>A", "c.413_415dup", "c.416A>C", "c.4251del", "c.4364C>G", "c.4426C>T", "c.489+1G>T", "c.523A>G", "c.54-5940_273+10250del21kB", "c.579+1G>T", "c.658C>T", "c.675T>A", "c.761delA", "c.892delA", "c.935_937delTCT", "Exon 19-21 del"}
175 - 177	CHRNE:Myasthenic syndrome, congenital, 4C {(NM_000080.4) "c.1161_1162insT", "c.1319_1326+15del", "c.1353dupG"}
178 - 178	CLCN1:Myotonia congenita, Autosomal Recessive {(NM_000083) "c.1444G>A"}
179 - 179	CLCN1:MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE {(NM_000083) "c.1586C>T"}
180 - 181	CLCNKB:Bartter syndrome Type3 {(NM_000085) "c.1830G>A" | (NM_000085.4) "c.1313G>A"}
182 - 182	CLN8:Ceroid lipofuscinosis neuronal 8 {(NM_018941.3) "c.766C>G"}
183 - 185	CLRN1:Usher syndrome, type 3A {(NM_174878.2) "c.144T>G", "c.349_358del", "c.528T>G"}
186 - 187	CNGA3:Achromatopsia 2 {(NM_001298.2) "c.1585G>A", "c.940_942del"}
188 - 189	CNGB1:Retinitis pigmentosa 45 {(NM_001286130) "c.2957A>T" | (NM_001297.5) "c.2284C>T"}
190 - 190	COL11A1:Fibrochondrogenesis 1 {(NM_001854) "c.4367G>T"}
191 - 191	COL11A2:OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA, WEISSENBACHER-ZWEYMULLER SYNDROME; WZS

	{(NM_080680.2) "c.3991C>T"}
192 - 192	COL6A2:Ullrich Congenital Muscular Dystrophy {(NM_001849.4) "c.1402C>T"}
193 - 195	COLQ:Myasthenic syndrome, congenital, 5 {(NM_005677) "c.788InsC" | (NM_005677.4) "c.1228C>T", "c.718G>T"}
196 - 199	CPS1:Carbamoyl phosphate synthetase I deficiency (CPS1d) {(NM_001122633) "c.3392C>T" | (NM_001875.5) "c.3265C>T", "c.3558+1G>C", "c.4101+2T>C"}
200 - 200	CPT1A:CPT1A (Carnitine Palmitoyltransferase I) {(NM_001031847) "c.1361A>G"}
201 - 201	CPT2:CPT2 CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, {(NM_000098) "c.1239_1240delGA"}
202 - 202	CRADD:Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly {(NM_003805) "c.52_59delGCAGAGGT"}
203 - 206	CRB1:Leber congenital amaurosis 8 {(NM_201253) "c.498_506del" | (NM_201253.3) "c.1148G>A", "c.1576C>T", "c.4121_4130delCAACTCAGGG"}
207 - 208	CRB2:Ventriculomegaly With Cystic Kidney Disease {(NM_173689.7) "c.1928A>C", "c.2400C>G"}
209 - 209	CRPPA (ISPD):Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7 {(NM_001101426.4) "c.165dupG"}
210 - 210	CTNS:Cystinosis {(NM_004937.3) "c.1015G>A"}
211 - 211	CTSK:Pycnodysostosis {(NM_000396.4) "c.990A>G"}
212 - 212	CYP11B1:Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency {(NM_000497.4) "c.1343G>A"}
213 - 214	CYP1B1:Glaucoma primary congenital GLC3A {(NM_000104) "c.1405C>T" | (NM_000104.3) "c.182G>A"}
215 - 217	CYP27A1:Cerebrotendinous xanthomatosis {(NM_000784.4) "c.355delC", "c.819delT", "c.845-1G>A"}
218 - 219	DBT:Maple syrup urine disease, type II {(NM_001918) "c.581C>G" | (NM_001918.5) "c.1064T>C"}
220 - 221	DCLRE1C:Severe combined immunodeficiency, Athabascan type {(NM_001033855) "c.1299_1306dupAGGATGCT", "c.310G>A"}
222 - 222	DDHD2:Spastic paraplegia 54 {(NM_015214.3) "c.94_101dupGATGCTGG"}
223 - 223	DDRGK1:Spondyloepimetaphyseal dysplasia, Shohat type {(NM_023935) "c.408+1G>A"}
224 - 224	DDX11:Warsaw breakage syndrome {(NM_030653.4) "c.1763-1G>C"}
225 - 225	DEGS1:Leukodystrophy, hypomyelinating, 18 {(NM_003676.4) "c.99A>G"}
226 - 226	Deletion_includes_PREPL_and_SLC3A1:2p21 DELETION SYNDROME, (HYPOTONIA-CYSTINURIA SYNDROME) {"2p21del"}
227 - 227	DGUOK:Mitochondrial DNA depletion syndrome 3 {(NM_080916.3) "c.255delA"}

228 - 228	DHCR24:DESMOSTEROLOSIS {(NM_014762.4) "c.307C>T"}
229 - 229	DHCR7:Smith Lemli Opitz syndrome {(NM_001360.3) "c.725G>A"}
230 - 232	DHCR7:Smith Lemli Opitz syndrome {(NM_001360.3) "c.452G>A", "c.755A>G", "c.964-1G>C"}
233 - 234	DHCR7:Smith Lemli Opitz syndrome {(NM_001360.3) "c.1210C>T", "c.506C>T"}
235 - 235	DHDDS:Retinitis pigmentosa 59 {(NM_024887.3) "c.124A>G"}
236 - 236	DHDPSL (HOGA1):Hyperoxaluria III {(NM_138413) **"c.944_946delAGG"}
237 - 239	DLD:DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY {(NM_000108) "c.658G>T" | (NM_000108.5) "c.1123G>A", "c.1436A>T"}
240 - 241	DMD:Duchenne Muscular Dystrophy {(NM_004009) "Whole exon DEL", "Whole exon DUP"}
242 - 243	DNAH11:Primary Ciliary Dyskinesia 7 {(NM_001277115.1) "c.7267-1G>T" | (NM_001277115.2) "c.11929G>T"}
244 - 245	DNAH5:Primary Ciliary Dyskinesia 3 {(NM_001369) "c.13432_13435delCACT" | (NM_001369.2) "c.8011-2A>G"}
246 - 246	DOCK8:HYPER-IgE RECURRENT INFECTION SYNDROME 2 {(NM_203447) "c.5132C>A"}
247 - 247	DOLK:Congenital disorder of glycosylation type Im {(NM_014908.3) "c.912G>T"}
248 - 250	DYSF:Limb Girdle Muscular Dystrophy 2 {(NM_003494.4) "c.2779delG", "c.4741C>T", "c.4872_4876delGCCCGinsCCCC"}
251 - 251	ECEL1:Distal arthrogryposis type 5 {(NM_004826) "c.110_155del"}
252 - 252	ELP1 (IKBKAP):Familial dysautonomia {(NM_003640.5) "c.2087G>C"}
253 - 253	ELP1 (IKBKAP):Familial dysautonomia {(NM_003640.5) "c.2204+6T>C"}
254 - 254	EOGT:ADAMS-OLIVER SYNDROME 4 {(NM_001278689.2) "c.1074delA"}
255 - 255	EPCAM:Congenital Tufting Enteropathy (DIAR5) {(NM_002354.3) "exon 1-9 Del"}
256 - 256	ERBB3:LETHAL CONGENITAL CONTRACTURAL SYNDROME 2 {(NM_001982.3) "c.1184-9A>G"}
257 - 257	ERCC2:Xeroderma pigmentosum group D {(NM_000400.4) "c.2048G>A"}
258 - 258	ERCC3:Xeroderma pigmentosum, group B {(NM_000122) "c.325C>T"}
259 - 259	ERCC5:Xeroderma pigmentosum, group G {(NM_000123.3) "c.205C>T"}
260 - 260	ERCC6:Cockayne syndrome type B {(NM_000124.4) "c.1034insT"}
261 - 263	ERCC8:Cockayne syndrome, type A {(NM_000082.3) "c.37G>T", "c.843+1G>C", "c.966C>A"}
264 - 264	ETFDH:Glutaric aciduria type IIC {(NM_004453.4) "c.1084G>A"}

265 - 269	EYS:Retinitis pigmentosa 25 {(NM_001142800.1) "DEL:chr6-65366279-65743530" | (NM_001142800.2) "c.403_423delinsCTTTT", "c.8155_8156delCA", "c.9286_9295del10", "c.9405T>A"}
270 - 270	F7:Factor 7 deficiency {(NM_000131.4) "c.1256C>T"}
271 - 274	FAH:Tyrosinemia type I {(NM_000137.2) "c.1062+5G>A", "c.554-1G>T", "c.707-1G>C", "c.782C>T"}
275 - 276	FAM161A:Retinitis pigmentosa 28 {(NM_001201543.1) "c.1355_1356delCA" | (NM_001201543.2) "c.1567C>T"}
277 - 283	FANCA:Fanconi anemia, complementation group A {(NM_000135.4) "c.189+1G>A", "c.2172_2173insG", "c.3382C>T", "c.3788_3790del", "c.4261-2A>C", "c.4275delT", "c.891_893+1del"}
284 - 288	FANCC:Fanconi anemia, complementation group C {(NM_000136.3) "c.1642C>T", "c.37C>T", "c.456+4A>T", "c.553C>T", "c.67del"}
289 - 289	FERMT1:Kindler syndrome {(NM_017671.4) "g.711-1241del"}
290 - 290	FKRP:Muscular dystrophy-dystroglycanopathy , type B {(NM_024301.5) "c.160C>T"}
291 - 291	FKTN:Walker Warburg Syndrome {(NM_001079802.2) "c.1167dupA"}
292 - 292	FRMD4A:BIRK-FLUSSER SYNDROME {(NM_018027) "c.2134_2146dupCTGGAGTCCCAGG"}
293 - 298	G6PC:Glycogen storage disease Ia - GDS1a {(NM_000151.4) "c.1039C>T", "c.247C>T", "c.248G>A", "c.508C>T", "c.562G>A", "c.562G>C"}
299 - 302	GAA:Pompe (glycogen storage disease II - GSD2) {(NM_000152.5) "c.1064T>C", "c.1210G>A", "c.1942G>A", "c.340_341insT"}
303 - 304	GALC:Krabbe {(NM_000153.4) "c.1630G>A", "c.1796T>G"}
305 - 310	GALT:Galactosemia {(NM_000155.3) "c.253-2A>G", "c.563A>G", "c.855G>T" | (NM_000155.4) "5.5-KB_DEL chr9:g.34645658-34651233del" | (NM_001378497.1) "c.667C>T" | (NM_001497) "c.590A>G"}
311 - 311	GATC:Hypertophic Cardiomyopathy {(NM_176818) "c.233T>G"}
312 - 318	GBA:Gaucher Disease Type 1 {(NM_000157.4) "c.115+1G>A", "c.1226A>G", "c.1297G>T", "c.1342G>C", "c.1448T>C", "c.84dupG" | (NM_001005741.2) "c.1604G>A"}
319 - 321	GCDH:GLUTARIC ACIDEMIA I {(NM_000159.4) "c.1173delG", "c.301G>A", "c.505+1G>A"}
322 - 322	GH1:ISOLATED GROWTH HORMONE DEFICIENCY, TYPE IB {(NM_000515.5) "c.456+5G>C"}
323 - 331	GJB2:Deafness Non Syndromic connexin 26 {(NM_004004.6) "c.-23+1G>A", "c.109G>A", "c.167delT", "c.229T>C", "c.269T>C", "c.35delG", "c.51del12insA", "c.614T>C", "c.71G>A"}
332 - 332	GJB6:Deafness Non Syndromic connexin 30 {(NM_001110219) "342Kb del(GJB6-D13S1830)"}
333 - 335	GLDC:Glycine Encephalopathy 1 {(NM_000170.2) "c.2607C>A",

	"c.2T>C" | (NM_000170.3) "c.2531T>G"}
336 - 336	GMPPA:Alacrima, achalasia, and impaired intellectual development syndrome {(NM_013335) "c.1000A>C"}
337 - 338	GNE:Nonaka myopathy (Hereditary inclusion body myopathy - HIBM) {(NM_001190383) "c.529C>A" | (NM_005476.7) "c.2135T>C"}
339 - 339	GNPTG:Mucolipidosis III {(NM_032520.5) "c.500insC"}
340 - 341	GUCY2D:LEBER CONGENITAL AMAUROSIS 1; LCA1 {(NM_000180.3) "c.2129C>T", "c.389delC"}
342 - 354	HEXA:Tay Sachs (TS) {(NM_000520.6) "c.1274_1277insTAT", "c.1351C>G", "c.1421+1G>C", "c.1528C>T", "c.459+2dupT", "c.496delC", "c.509G>A", "c.571-2A>G", "c.749G>A", "c.749G>T", "c.805G>A", "c.835T>C", "c.910_912delTTC"}
355 - 355	HEXB:Sandhoff disease {(NM_000521) "c.1082+5G>A"}
356 - 356	HMGCL:HMG CoA lyase deficiency {(NM_000191.3) "c.122G>A"}
357 - 357	HPD:Tyrosinemia, type III {(NM_002150.3) "c.415(-1)G>A"}
358 - 358	HSD17B4:D-bifunctional protein deficiency {(NM_000414.3) "c.944-1G>A"}
359 - 359	HSPD1:Pelizaeus-Merzbacher like disease (Leukodystrophy, hypomyelinating, 4) {(NM_199440.1) "c.86A>G"}
360 - 360	IDUA:Hurler syndrome {(NM_000203.5) "c.1096A>C"}
361 - 361	IFT140:Retinitis pigmentosa 80 {(NM_014714) "c.1646C>T"}
362 - 363	IGHMBP2:spinal muscular dystrophy related disease (Neuronopathy, distal hereditary motor, autosomal recessive 1) {(NM_002180.2) "c.114delA", "c.707T>G"}
364 - 364	IL10RA:INFLAMMATORY BOWEL DISEASE 28, AUTOSOMAL RECESSIVE; IBD28 {(NM_001558) "c.537G>A"}
365 - 366	IL7RA:Severe combined immunodeficiency 104 {(NM_002185.5) "c.120C>G", "c.222T>G"}
367 - 367	INSR:Leprechaunism- Donoho Syndrome {(NM_000208.4) "c.167T>C"}
368 - 368	INVS:NEPHRONOPHTHISIS 2; NPHP2 {(NM_014425.5) "c.2719C>T"}
369 - 369	ITGB4:CARMI SYNDROME (EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA) {(NM_000213.5) "c.3279_3793+180del"}
370 - 373	IVD:IVD:Isovaleric academia {(NM_002225.5) "c.148C>T", "c.286+2T>C", "c.456+2T>C", "c.932C>T"}
374 - 375	KCNJ1:Bartter syndrome, antenatal, type 2 {(NM_000220.6) "c.1076T>G" | (NM_000492.4) "c.658C>T"}
376 - 376	KRT14:EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1; EBSB1 {(NM_000526) "c.915G>A"}
377 - 377	KY:Myopathy, Myofibrillar, 7; MF7 {(NM_178554) "c.51_52insTATCGACATGTGCTGTATCTATCGACAT (28)"}
378 - 381	LAMA2:MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A; MDC1A {(NM_000426) "c.5259delA" | (NM_000426.3) "c.3718C>T", "c.8665G>A", "c.8689C>T"}

382 - 383	LAMA3:Epidermolysis bullosa type2B {(NM_000227.4) "c.1981C>T", "c.2975delA"}
384 - 385	LAMB3:Epidermolysis bullosa type1B {(NM_000228.3) "c.124C>T", "c.3247C>T"}
386 - 386	LIFR:Stuve Wiedemann {(NM_002310.5) "c.2472_2476delTATGT"}
387 - 388	LIPA:Wolman disease {(NM_001127605.2) "c.260G>T", "c.398delC"}
389 - 390	LONP1:CODAS syndrome {(NM_004793) "c.1939G>A" | (NM_004793.4) "c.2009C>T"}
391 - 391	LTBP3:Dental Anomalies And Short Stature Skeletal Dysplasia {(NM_021070) "c.1346-1G>A"}
392 - 392	MADD:Deeah syndrome, IMNEPD2 {(NM_003682.4) "c.2816+1G>A"}
393 - 393	MAN1B1:CDG type II (MR AR15) {(NM_016219.5) "c.1863G>A"}
394 - 394	MAN2B1:Mannosidosis,alpha (types I & II)-MANSA {(NM_000528.3) "c.454A>T"}
395 - 398	MCOLN1:Mucolipidosis type IV - ML4 {(NM_020533.3) "c.1207C>T", "c.406-2A>G", "c.874_788del6433", "c.964C>T"}
399 - 400	MECR:Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities {(NM_016011) "c.695G>A", "c.830+2dupT"}
401 - 401	MED17:Microcephaly, postnatal progressive, with seizures and brain atrophy ((ICCA) {(NM_004268.5) "c.1112T>C"}
402 - 402	MED25:Basel-Vanagaite-Smirin-Yosef Syndrome {(NM_030973.3) "c.116A>G"}
403 - 403	MICOS13:Methyl Glutaric Aciduria {(NM_205767) "c.44delG"}
404 - 405	MLC1:Megalencephalic leukoencephalopathy with subcortical cysts {(NM_015166.3) "c.176G>A", "c.274C>T"}
406 - 406	MMACHC:Methylmalonic aciduria and homocystinuria type cblC {(NM_015506.3) "c.271dupA"}
407 - 407	MMUT:Methylmalonic acidemia due to methylmalonyl-CoA mutase deficiency {(NM_000255.4) "c.1091A>C"}
408 - 408	MOCS1:Molybdenum cofactor deficiency A {(NM_001075098.3) "c.971G>A"}
409 - 409	MOCS2:Molybdenum cofactor deficiency B {(NM_004531.5) "c.226G>A"}
410 - 410	MPL:MPL:Thrombocytopenia, congenital amegakaryocytic {(NM_005373.3) "c.79+2T>A"}
411 - 411	MRE11:ATAXIA-TELANGIECTASIA-LIKE DISORDER 1; ATLD1 {(NM_005591.3) "c.290A>G"}
412 - 413	MTHFR:Homocystinuria due to MTHFR deficiency {(NM_005957.4) "c.16delA", "c.474A>T"}
414 - 414	MYBPC1:LETHAL CONGENITAL CONTRACTURE SYNDROME 4; LCCS4 (Arthrogryposis) {(NM_002465.4) "c.952C>T"}
415 - 418	MYO15A:Deafness, Autosomal Recessive 3 {(NM_000260.5) "c.9861C>T" | (NM_016239) "c.4198G>A" | (NM_016239.4) "c.8183G>A", "c.8467G>A"}

419 - 427	MYO7A:MYO7A:Usher syndrome, type 1B {(NM_000260.4) "c.1190C>A", "c.1969C>T", "c.1996C>T", "c.2187+1G>A", "c.2476G>A", "c.29T>C", "c.620A>G", "c.640G>A", "c.6487G>A"}
428 - 429	NARS2:Combined oxidative phosphorylation deficiency 24 {(NM_024678) "c.500A>G" | (NM_024678.6) "c.434T>G"}
430 - 430	NBEAL2:Gray platelet syndrome {(NM_015175.2) "c.2701C>T"}
431 - 431	NCF1:Chronic granulomatous disease due to deficiency of NCF-1 {(NM_000265.6) *"c.579G>A"}
432 - 432	NDUFA11:Mitochondrial complex I deficiency nuclear type 1 (Hypertrophic cardiomyopathy) {(NM_001193375.1) "c.97+5G>A"}
433 - 433	NDUFS2:Mitochondrial complex I deficiency nuclear type 6 {(NM_004550.4) "c.1237T>C"}
434 - 434	NDUFS6:Mitochondrial complex I deficiency nuclear type 9 {(NM_004553.4) "c.344G>A"}
435 - 436	NEB:Nemaline myopathy 2 {(NM_001271208.2) "c.9619-2A>G" | (NM_004543.4) "c.7431+1917_7536+372del"}
437 - 437	NGLY1:CONGENITAL DISORDER OF DEGLYCOSYLATION 1 {(NM_018297.4) "c.1294G>T"}
438 - 438	NHEJ1:Microphthalmia / Anophthalmia / Coloboma {(NM_024782.3) "c.588+18131A>G"}
439 - 440	NPC1:NIEMANN-PICK DISEASE, TYPE C1; NPC1 {(NM_000271.5) "c.1211G>A", "c.2972_2973delAG"}
441 - 441	NPHP1:Joubert syndrome/nephronophthisis 1 {(NM_000272) "del exons 2-7"}
442 - 445	NPHS1:Congenital nephrotic syndrome {(NM_004646.3) "c.1138C>T", "c.2104G>A", "c.2160dupC", "c.514_516delACC"}
446 - 448	NTRK1:Congenital insensivity to pain with anhidrosis {(NM_002529.3) "c.207_208delTG", "c.2084C>T" | (NM_002529.4) "c.1860_1861insT"}
449 - 449	NUP62:Infantile bilateral striatal necrosis (Strionigral degeneration) {(NM_016553.4) "c.1172A>C"}
450 - 450	OCA2:Albinism, oculocutaneous, type II {(NM_000275) "c.1327G>A"}
451 - 451	OPA3:Methylglutaconic aciduria, type III - Costeff {(NM_025136.4) "c.143-1G>C"}
452 - 453	OTOF:Deafness, Autosomal Recessive 9 {(NM_194248) "c.5193-1G>A", "c.5332G>T"}
454 - 465	PAH:PAH:phenylketonuria {(NM_000277.3) "c.1066-11G>A", "c.1222C>T", "c.143T>C", "c.165delT", "c.168+1G>A", "c.441+5G>T", "c.442-5C>G", "c.473G>A", "c.722G>A", "c.782G>A", "c.842C>T", "g.22736_29335delinsGGCACCTG (exon3del)"}
466 - 466	PAX7:MYOPATHY CONGENITAL PROGRESSIVE with SCOLIOSIS {(NM_001135254) "c.1403-2A>G"}
467 - 468	PCDH12:Diencephalic-mesencephalic junction dysplasia syndrome 1 {(NM_016580) "c.2515C>T", "c.995delT"}
469 - 469	PCDH15:Usher syndrome, type 1F {(NM_033056.3) "c.733C>T"}

470 - 470	PCNT:Microcephalic osteodysplastic (MOPDII) {(NM_006031.5) "c.3465-1G>A"}
471 - 471	PDE6G:Retinitis pigmentosa 57 {(NM_002602.4) "c.187+1G>T"}
472 - 473	PEPD:Prolidase Deficiency {(NM_000285.4) "c.1103T>G", "c.605C>T"}
474 - 475	PEX2:Peroxisome biogenesis disorder 5a {(NM_001079867.2) "c.355C>T", "c.550delC"}
476 - 476	PEX6:PEROXISOME BIOGENESIS DISORDER 1A (ZELLWEGER); PBD1A {(NM_000287.4) "c.1947delG"}
477 - 477	PGAP3:Hyperphosphatasia mental retardation syndrome 4 {(NM_033419.5) "c.845A>G"}
478 - 478	PGM1:CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1T ; CDG1T {(NM_002633) "c.112A>T"}
479 - 479	PIP5K1C:LETHAL CONGENITAL CONTRACTURE SYNDROME 3; LCCS3 {(NM_012398.2) "c.757G>A"}
480 - 480	PJVK:Deafness Non Syndromic 59 {(NM_001042702.4) "c.406C>T"}
481 - 488	PKHD1:Polycystic Kidney disease 4 PKD4 {(NM_138694.4) "c.10444C>T", "c.1350delC", "c.1486C>T", "c.2279G>A", "c.3761_3762delCCinsG", "c.4870C>T", "c.6122-12G>A", "c.8870T>C"}
489 - 490	PLA2G6:Infantile neuroaxonal dystrophy (INAD) {(NM_003560.4) "c.2070_2072delT", "c.2251G>A"}
491 - 491	PLAA:Autosomal recessive progressive Leukoencephalopathy {(NM_001031689.3) "c.2254C>T"}
492 - 492	PMM2:Congenital Disorder of Glycosylation type1A {(NM_000303.3) "c.422G>A"}
493 - 493	POC1A:POC1A deficiency-SHORT STATURE, ONYCHODYSPLASIA, FACIAL DYSMORPHISM, AND HYPOTRICHOSIS; SOFT {(NM_015426.5) "c.512T>C"}
494 - 494	POMGNT2:Muscular dystrophy-dystroglycanopathy LGMD {(NM_032806.6) "c.1232_1233delAG"}
495 - 495	POR:DISORDERED STEROIDOGENESIS DUE TO CYTOCHROME P450 OXIDOREDUCTASE DEFICIENCY {(NM_000941.3) "c.1615G>A"}
496 - 496	PPP1R13L:Cardio-cutaneous syndrome {(NM_006663.4) "c.2241C>G"}
497 - 497	PRCD:Retinitis pigmentosa 36 {(NM_001077620) "c.64C>T"}
498 - 498	PRF1:Familial Hemophagocytic Lymphohistiocytosis 2-FHL2 {(NM_005041) "c.1122G>A"}
499 - 499	PUS1:Mitochondrial myopathy and sideroblastic anemia 1 {(NM_025215.6) "c.430C>T"}
500 - 501	RAG1:Severe combined immune deficiency (SCID) B cell-negative {(NM_000448) "c.555delG" | (NM_000448.2) "c.1361T>A"}
502 - 503	RAG2:Severe combined immune deficiency (SCID) B cell-negative {(NM_000536.3) "c.379A>T", "c.685C>T"}
504 - 505	RAPSN:Myasthenic syndrome, congenital, associated with

	acetylcholine receptor deficiency {(NM_005055.5) "c.-210A>G", "c.264C>A"}
506 - 506	RARS2:Pontocerebellar hypoplasia, type 6 {(NM_020320.5) "c.110+5A>G"}
507 - 507	RBL2:Infantile hypotonia, severe developmental delay and microcephaly- Brunet-Wagner neurodevelopmental syndrome {(NM_005611.3) "c.926dup"}
508 - 511	RDH12:Leber congenital amaurosis 13 {(NM_152443) "c.821T>C" | (NM_152443.3) "c.146C>T", "c.377C>T", "c.740T>C"}
512 - 512	RFX5:BARE LYMPHOCYTE SYNDROME, TYPE II/SEVERE COMBINED IMMUNODEFICIENCY, HLA CLASS II-NEGATIVE (SCID, HLA CLASS II-NEGATIVE) {(NM_000449) "c.715C>T"}
513 - 513	RNASEH2B:Aicardi-Goutieres syndrome 2 {(NM_024570.4) "c.529G>A"}
514 - 514	ROGDI:Kohlschutter-Tonz Syndrome {(NM_024589.2) "c.469C>T"}
515 - 516	RPE65:Leber congenital amaurosis 2 (LCA2) {(NM_000329.3) "c.227A>C", "c.95-2A>T"}
517 - 517	RYR1:Multiminicore Disease {(NM_000540.2) "c.9623C>T"}
518 - 519	SAMD9:Tumoral calcinosis, familial, normophosphatemic {(NM_017654.4) "c.1030C>T", "c.4483A>G"}
520 - 520	SARS2:HUPRA {(NM_017827.3) "c.1169A>G"}
521 - 521	SCAPER:INTELLECTUAL DEVELOPMENTAL DISORDER AND RETINITIS PIGMENTOSA; IDDRP {(NM_020843) "c.2806delC"}
522 - 522	SCN9A:INDIFFERENCE TO PAIN, CONGENITAL, AUTOSOMAL RECESSIVE; CIP {(NM_002977.3) "c.2687G>A"}
523 - 523	SDHA:CARDIOMYOPATHY, DILATED, 1GG; CMD1GG {(NM_004168.4) "c.1664G>A"}
524 - 524	SELENOI:Spastic paraplegia 81 EPT1 deficiency {(NM_033505) "c.732-2A>G"}
525 - 526	SEPSECS:Progressive Cerebello Cerebro Atrophy Pontocerebellar hypoplasia type 2D (PCCA) {(NM_016955.4) "c.1001A>G", "c.715G>A"}
527 - 527	SGCA:Muscular dystrophy, limb-girdle, autosomal recessive 3 {(NM_000023) "c.600G>A"}
528 - 528	SGCG:Limb girdle muscular dystrophy (LGMD2C) {(NM_000231.2) "c.525delT"}
529 - 533	SGSH:MPS3A (sanfilippo syndrome A) {(NM_000199) "c.1231C>T", "c.267C>A" | (NM_000199.5) "c.1093C>T", "c.1298G>A", "c.416C>T"}
534 - 534	SGSM3:Intellctual disability and short stature {(NM_015705.6) "c.981dup"}
535 - 535	SLC12A1:Barter Syndrome, Type 1, Antenatal {(NM_000338.3) "c.2759G>A"}
536 - 536	SLC17A5:INFANTILE SIALIC ACID STORAGE DISEASE; ISSD {(NM_012434.5) "c.983G>A"}

537 - 537	SLC18A3:Myasthenic syndrome 21 {(NM_003055) "c.1078G>C"}
538 - 539	SLC1A4:Spastic tetraplegia, thin corpus callosum, and progressive microcephaly {(NM_003038.5) "c.766G>A", "c.944_945del"}
540 - 540	SLC25A20:Carnitine-acylcarnitine translocase deficiency {(NM_000387.6) "c.713A>G"}
541 - 542	SLC26A4:Pendred syndrome {(NM_000441.2) "c.1198delT", "c.349C>T"}
543 - 544	SLC29A3:Histiocytois Lymphadenopathy Plus Syndrome (H Syndrome) {(NM_018344.6) "c.1279G>A", "c.1309G>A"}
545 - 545	SLC2A2:Fanconi-Bickel syndrome {(NM_000340.2) "c.734A>C"}
546 - 546	SLC30A9:BIRK-LANDAU-PEREZ SYNDROME; BILAPES (Cerebro-Renal Syndrome) {(NM_006345.4) "c.1049_1051delCAG"}
547 - 548	SLC37A4:GLYCOGEN STORAGE DISEASE Ib; GSD1B {(NM_001164277.1) "c.1042_1043delCT", "c.83G>A"}
549 - 550	SLC38A8:Foveal hypoplasia 2 {(NM_001080442) "c.848A>C" | (NM_001080442.3) "c.95T>G"}
551 - 551	SLC4A4:RENAL TUBULAR ACIDOSIS (RTA), PROXIMAL, WITH OCULAR ABNORMALITIES AND MENTAL RETARDATION {(NM_003759.3) "c.2321G>A"}
552 - 552	SMN1:Spinal Muscular Atrophy type 1 {(NM_022874.2) "Exon 7 DEL"}
553 - 557	SMPD1:Niemann-Pick disease type B, SMPD1-related {(NM_000543.5) "c.1493G>T", "c.1828_1830delCGC", "c.573delT", "c.911T>C", "c.996delC"}
558 - 558	SNAP29:Cerebral dysgenesis, neuropathy, ichthyosis and keratodermia (CEDNIK) {(NM_004782) "c.223delG"}
559 - 559	SPEG:Myopathy, Centronuclear, 5; CNM5 {(NM_005876.4) "c.5038G>A"}
560 - 561	SPG11:Spastic paraplegia with mental impairment {(NM_025137) "c.5986dupT" | (NM_025137.4) "c.4339C>T"}
562 - 565	SPINK5:Netherton Syndrome NETH {(NM_006846.4) "c.2240+5G>A", "c.2557C>T", "c.649C>T", "c.691delC"}
566 - 567	SUMF1:MULTIPLE SULFATASE DEFICIENCY; MSD {(NM_182760.3) "c.1043C>T", "c.463T>C"}
568 - 568	SZT2:Epileptic encephalopathy, early infantile, 18 {(NM_001365999.1) "c.73C>T"}
569 - 570	TAF2:Mental Retardation, Autosomal Recessive 40; MRT40 {(NM_003184.4) "c.1247C>A", "c.557C>G"}
571 - 571	TBCB (CKAPI):Neurodevelopmental delay, autism, cognitive impairment and spastic gait {(NM_001281) "c.589T>A"}
572 - 572	TBCD:Infantile neurodegenerative disorder - Early onset progressive encephalopathy (PEBAT) {(NM_005993.4) "c.1423G>A"}
573 - 574	TBCE:Hypoparathyroidism retardation dysmorphism (HRD) {(NM_003193.5) "c.154_165delAGCCACGAAGGG", "c.355_356del"}

575 - 575	TBX19:Congenital thyroid hormone and glucocorticoid deficiency (ACTH DEFICIENCY ISOLATED; IAD) {(NM_005149.3) "c.574_577delATAG"}
576 - 577	TCIRG1:OSTEOPETROSIS, AUTOSOMAL RECESSIVE 1; OPTB1 {(NM_006019.4) "c.117+4A>T", "c.674delG"}
578 - 578	TCTN2:Meckel Gruber Syndrome Type8 {(NM_024809.5) "c.1506-2A>G"}
579 - 580	TECPR2:Spastic paraplegia 49, Autosomal Recessive {(NM_001172631.2) "c.1319del", "c.3416delT"}
581 - 582	THG1L:Spinocerebellar ataxia, autosomal recessive 28 {(NM_017872) "c.153C>G" | (NM_017872.5) "c.164T>C"}
583 - 583	TIMM50:3- Methylglutaconic aciduria type 9 {(NM_001001563.5) "c.734T>C"}
584 - 586	TK2:Mitochondrial DNA depletion syndrome 2 (MTDPS2) {(NM_004614.5) "c.360_361delGCinsAA", "c.361C>A", "c.635T>A"}
587 - 587	TKT:Transketolase deficiency, Short stature, developmental delay, and congenital heart defects {(NM_001135055.2) "c.769_770insCTACCTCCTTATCTTCTG"}
588 - 592	TMC1:DEAFNESS, AUTOSOMAL RECESSIVE 11; DFNB11 {(NM_138691.2) "c.100C>T", "c.1165C>T", "c.1210T>C", "c.1810C>T", "c.1939T>C"}
593 - 594	TMEM216:Joubert syndrome 2 {(NM_001173990.3) "c.218G>T", "c.230G>C"}
595 - 595	TMEM38B:OSTEOGENESIS IMPERFECTA, TYPE XIV; OI14 {(NM_018112) "c.454+279_543-5092delinsAATTAAGGTATA"}
596 - 598	TMEM67:Meckel Syndrome 3 (MKS3) {(NM_153704) "c.1065+1delG" | (NM_153704.5) "c.1975C>T", "c.725A>G"}
599 - 599	TNNT1:Nemaline myopathy 5 {(NM_001291774) "c.606_607insTAGTG"}
600 - 600	TRPM6:HYPOMAGNESEMIA 1 (HOMG1) {(NM_017662.5) "c.2009+1G>A"}
601 - 602	TSEN54:Pontocerebellar hypoplasia type 5, Pontocerebellar hypoplasia type 4 {(NM_207346) "c.371G>T" | (NM_207346.3) "c.919G>T"}
603 - 605	TSPEAR:Ectodermal dysplasia 14 {(NM_001272037) "c.1788_1790del" | (NM_144991.2) "c.1852T>A" | (NM_144991.3) "c.1877T>C"}
606 - 606	TTN:Lethal Congenital Arthrogryposis {(NM_001267550.2) "c.36122delC"}
607 - 625	TYR:Albinism, oculocutaneous, type IA {(NM_000372.5) "c.1037-1G>A(IVS2-1G>A)", "c.1037-7T>A", "c.1059del", "c.1118C>A", "c.1204C>T", "c.1217C>T", "c.1299C>G", "c.1342G>A", "c.1357C>T", "c.140G>A", "c.149C>G", "c.1A>G", "c.649delC", "c.650G>A", "c.74dupT", "c.757G>A", "c.832C>T", "c.880G>A", "c.896G>A"}
626 - 626	UNC80:HYPOTONIA, INFANTILE, WITH PSYCHOMOTOR RETARDATION AND CHARACTERISTIC FACIES 2; IHPRF2

	{(NM_032504.1) "c.151C>T"}
627 - 627	UPB1:Beta-ureidopropionase deficiency {(NM_016327.3) "c.917-1G>A"}
628 - 628	UQCRQ:MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 4; MC3DN4 {(NM_014402.5) "c.134C>T"}
629 - 630	USH1C:USHER SYNDROME, TYPE IC {(NM_153676.2) "c.1220delG", "c.238InsC"}
631 - 640	USH2A:Usher syndrome, type 2A {(NM_206933.4) "c.1000C>T", "c.12067-2A>G", "c.12575G>A", "c.2209C>T", "c.236_239insGTAC", "c.3368A>G", "c.4544C>T", "c.5519G>T", "c.5776+1G>A", "c.8719A>C"}
641 - 641	USMG5:Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6 {(NM_032747) "c.87+1G>C"}
642 - 642	VDR:Rickets 1,25 dehydroxy vitamin D3 resistant {(NM_001017535.1) "c.885C>A"}
643 - 643	VPS37A:Complex hereditary spastic paraparesis {(NM_152415.3) "c.1146A>T"}
644 - 645	VPS53:Progressive Cerebello Cerebro Atrophy Pontocerebellar hypoplasia type 2E (PCCA2) {(NM_001128159.3) "c.1556+5G>A", "c.2084A>G"}
646 - 646	WISP3 (CCN6):ARTHROPATHY, PROGRESSIVE PSEUDORHEUMATOID, OF CHILDHOOD; PPAC {(NM_198239.2) "c.536_537delGT"}
647 - 647	WWOX:Developmental and epileptic encephalopathy 28 {(NM_016373.4) "c.517-2A>G"}
648 - 648	XRCC2:Fancini Anemia type U {(NM_005431.2) "c.643C>T"}
649 - 649	YARS1:Neurologic, Endocrine And Pancreatic Disease, Multisystem, Infantile-Onset {(NM_003680.4) "c.1099C>T"}
650 - 650	ZNF341:Hyper IgE syndrome {(NM_001282933.2) "c.904C>T"}