CARRIER SCREENING 600

MedGEN
Complete Health Solution

[Diseases Tested]

This test contains variants amounts to 5971, as described in the following table:

1）Variants of group A

2-Methylbutyryl Glycinuria. Gene:*ACADSB*. **Exons**: NM_001609.3:2-11. **Variants(3)**: c.1159G>A, c.763C>T, c.443C>T
3-hydroxy-3-methylglutaryl-CoA lyase deficiency. Gene:*HMGCL*. **Exons**: NM_000191.2:1-9. **Variants(2)**: c.835G>A, c.122G>A
3-Hydroxyisobutryl-CoA Hydrolase Deficiency. Gene:*HIBCH*. **Exons**: NM_014362.3:1-14. **Variants(3)**: c.365A>G, c.220-9T>G, c.79-3C>G
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency. Gene:*MCCC1*. **Exons**: NM_020166.3:1-19. **Variants(4)**: c.1310T>C, c.2079delA, c.1594G>C, c.1380T>G
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency. Gene:*MCCC2*. **Exons**: NM_022132.4:1-17. **Variants(7)**: c.464G>A, c.295G>C, c.517_518insT, c.929C>G, c.1015G>A, c.803G>C, c.838G>T
3-Methylglutaconic Aciduria Type 1. Gene:*AUH*. **Exons**: NM_001698.2:1-10. **Variants(5)**: c.589C>T, c.559G>A, c.991A>T, c.895-1G>A, c.650G>A
3-Methylglutaconic Aciduria Type 3. Gene:*OPA3*. **Exons**: NM_025136.3:1-2. **Variants(1)**: c.143-1G>C
3-Methylglutaconic Aciduria Type 5. Gene:*DNAJC19*. **Exons**: NM_145261.3:1-6. **Variants(1)**: c.130-1G>C
ABCC8-Related Congenital Hyperinsulinism. Gene:*ABCC8*. **Exons**: NM_000352.3:1-39. **Variants(5)**: c.3989-9G>A, c.560T>A, c.2147G>T, c.4160_4162delTCT, c.4307G>A
ABCD Syndrome. Gene:*EDNRB*. **Exons**: NM_000115.3:2-8. **Variants(1)**: c.601C>T
Abetalipoproteinemia. Gene:*MTTP*. **Exons**: NM_000253.2:2-19. **Variants(3)**: c.1769G>T, c.1619G>A, c.2593G>T
ACAD9 deficiency. Gene:*ACAD9*. **Exons**: NM_014049.4:1-18. **Variants(4)**: c.1594C>T, c.130T>A, c.1249C>T, c.797G>A
ACE-Related Renal Tubular Dysgenesis. Gene:*ACE*. **Exons**: NM_000789.3:2-25. **Variants(4)**: c.1486C>T, c.1319_1322delTGGA, c.798C>G, c.2371C>T
Achondrogenesis Type 1B. Gene:*SLC26A2*. **Exons**: NM_000112.3:2-3. **Variants(4)**: c.1020_1022delTGT, c.1273A>G, c.532C>T, c.2033G>T
Acrocallosal Syndrome. Gene:*KIF7*. **Exons**: NM_198525.2:2-4,6-19. **Variants(3)**: c.687delG, c.3001C>T, c.460C>T
Acyl-CoA Dehydrogenase Deficiency,Medium-Chain. Gene:*ACADM*. **Exons**: NM_000016.4:1-12. **Variants(2)**: c.199T>C, c.985A>G
Acyl-CoA Dehydrogenase Deficiency,Very Long-Chain. Gene:*ACADVL*. **Exons**: NM_000018.3:1-20. **Variants(3)**: c.779C>T, c.848T>C, c.1322G>A
Adenosine Deaminase Deficiency. Gene:*ADA*. **Exons**: NM_000022.2:2-12. **Variants(4)**: c.632G>A, c.226C>T, c.986C>T, c.320T>C
AGTR1-Related Renal Tubular Dysgenesis. Gene:*AGTR1*. **Exons**: NM_031850.3:3-4. **Variants(1)**: c.215dupT
AGT-Related Renal Tubular Dysgenesis. Gene:*AGT*. **Exons**: NM_000029.3:2-5. **Variants(2)**: c.604C>T, c.1290delT
Aicardi-goutières Syndrome 1. Gene:*TREX1*. **Exons**: NM_033629.3:2. **Variants(2)**: c.490C>T, c.341G>A
AICA-ribosiduria due to ATIC deficiency. Gene:*ATIC*. **Exons**: NM_004044.6:2-16. **Variants(1)**: c.1277A>G
Alkaptonuria. Gene:*HGD*. **Exons**: NM_000187.3:1-14. **Variants(11)**: c.688C>T, c.457_458insG, c.1111_1112insC, c.342+1G>A, c.808G>A, c.899T>G, c.175delA, c.1102A>G, c.360T>G, c.16-1G>A, c.481G>A
Alpha-Mannosidosis. Gene:*MAN2B1*. **Exons**: NM_000528.3:1-24. **Variants(7)**: c.215A>T, c.1067C>G, c.1830+1G>C, c.1915C>T, c.2278C>T, c.2426T>C, c.2248C>T
Alpha-Methylacyl-CoA Racemase Deficiency. Gene:*AMACR*. **Exons**: NM_014324.5:1-5. **Variants(1)**: c.154T>C
Alpha-thalassemia.Gene:*HBA1/HBA2*. **Variants(5)**: aa/-a3.7, aa/-a4.2, aa/-aSEA, aa/-aFIL, aa/-aTHAI
Alstrom Syndrome. Gene:*ALMS1*. **Exons**: NM_015120.4:2-23. **Variants(6)**: c.11449C>T, c.11316_11319delAGAG, c.8164C>T, c.8383C>T, c.10775delC, c.10483C>T
AMT-Related Glycine Encephalopathy. Gene:*AMT*. **Exons**: NM_000481.3:1-9. **Variants(3)**: c.125A>G, c.139G>A, c.959G>A
Androgen Insensitivity Syndrome. Gene:*AR*. **Exons**: NM_000044.3:2-8. **Variants(10)**: c.2650A>T, c.1769-11T>A, c.521T>G, c.2395C>G, c.340C>T, c.4G>A, c.1771A>T, c.2391G>A, c.2567G>A, c.2157G>A
Arginase Deficiency. Gene:*ARG1*. **Exons**: NM_000045.3:1-8. **Variants(7)**: c.365G>A, c.871C>T, c.703G>C, c.413G>T, c.32T>C, c.869C>G, c.61C>T
Argininosuccinic Aciduria. Gene:*ASL*. **Exons**: NM_000048.3:2-17. **Variants(8)**: c.35G>A, c.1060C>T, c.857A>G, c.1135C>T, c.532G>A, c.346C>T, c.1153C>T, c.283C>T
Aromatic L-Amino Acid Decarboxylase Deficiency. Gene:*DDC*. **Exons**: NM_000790.3:2-14. **Variants(2)**: c.749C>T, c.304G>A
Arthrogryposis-Renal Dysfunction-Cholestasis 1. Gene:*VPS33B*. **Exons**: NM_018668.3:1-23. **Variants(3)**: c.1594C>T, c.89T>C, c.1312C>T
Aspartylglucosaminuria. Gene:*AGA*. **Exons**: NM_000027.3:1-9. **Variants(5)**: c.302C>T, c.488G>C, c.214T>C, c.904G>A, c.800dupT
Ataxia With Oculomotor Apraxia Type 1. Gene:*APTX*. **Exons**: NM_175073.2:3-9. **Variants(7)**: c.837G>A, c.875-1G>A, c.788T>G, c.602A>G, c.167delT, c.320delC, c.617C>T
Ataxia With Oculomotor Apraxia Type 2. Gene:*SETX*. **Exons**: NM_015046.5:3-26. **Variants(7)**: c.6638C>T, c.3880C>T, c.5927T>G, c.1027G>T, c.994C>T, c.2602C>T, c.4087C>T
Ataxia With Vitamin E Deficiency. Gene:*TTPA*. **Exons**: NM_000370.3:2-5. **Variants(5)**: c.485delG, c.513_514insTT, c.400C>T,

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c.552G>A, c.744delA

Ataxia, Posterior Column, With Retinitis Pigmentosa. Gene:*FLVCR1*. **Exons**: NM_014053.3:1-10. **Variants(4)**: c.1477G>C, c.721G>A, c.361A>G, c.574T>C

Ataxia-telangiectasia. Gene:*ATM*. **Exons**: NM_000051.3:2-63. **Variants(11)**: c.7517_7520delGAGA, c.7967T>C, c.8030A>G, c.9139C>T, c.8480T>G, c.3245_3247delATCinsTGAT, c.103C>T, c.7875_7876delTGinsGC, c.5932G>T, c.5908C>T, c.3576G>A

Atelosteogenesis Type 2. Gene:*SLC26A2*. **Exons**: NM_000112.3:2-3. **Variants(4)**: c.1535C>A, c.835C>T, c.764G>A, c.2144C>T

ATP7A-Related Copper Transport Disorders. Gene:*ATP7A*. **Exons**: NM_000052.5:2-23. **Variants(7)**: c.3911A>G, c.601C>T, c.1910C>T, c.2938C>T, c.4156C>T, c.2981C>T, c.3056G>A

Autoimmune Polyendocrine Syndrome Type 1. Gene:*AIRE*. **Exons**: NM_000383.3:1-11,13-14. **Variants(8)**: c.239T>G, c.1103_1104insC, c.247A>G, c.415C>T, c.1513delG, c.769C>T, c.254A>G, c.789delC

Autosomal Recessive Deafness 12. Gene:*CDH23*. **Exons**: NM_022124.5:2-70. **Variants(5)**: c.4021G>A, c.719C>T, c.902G>A, c.6442G>A, c.5663T>C

Autosomal Recessive Deafness 1A. Gene:*GJB2*. **Exons**: NM_004004.5:2. **Variants(58)**: c.340G>T, c.195C>G, c.134G>A, c.71G>A, c.533T>C, c.231G>A, c.56G>C, c.139G>T, c.416G>A, c.647_650delGATA, c.427C>T, c.35_36insC, c.268C>G, c.59T>C, c.358_360delGAG, c.269T>C, c.476A>T, c.617A>G, c.299_300delAT, c.334_335delAA, c.641T>C, c.408C>A, c.370C>T, c.638T>A, c.290_291insT, c.229T>C, c.398G>A, c.230G>A, c.614T>C, c.132G>A, c.283G>A, c.550C>T, c.523C>A, c.167delT, c.633T>A, c.44A>C, c.504_505insCCTT, c.572delT, c.94C>T, c.246C>G, c.508_509insT, c.169C>T, c.250G>C, c.119C>A, c.508_511dupAACG, c.428G>A, c.176_191delGCTGCAAGAACGTGTG, c.363delC, c.279G>A, c.516G>A, c.148G>A, c.439G>A, c.235delC, c.270dupA, c.518C>G, c.598G>A, c.1A>G, c.35delG.

Autosomal Recessive Deafness 2. Gene:*MYO7A*. **Exons**: NM_000260.3:2-49. **Variants(5)**: c.1797G>A, c.133-2A>G, c.1184G>A, c.731G>C, c.3596dupT

Autosomal Recessive Deafness 21. Gene:*TECTA*. **Exons**: NM_005422.2:1-23. **Variants(3)**: c.6037delG, c.2941+1G>A, c.651_652insC

Autosomal Recessive Deafness 22. Gene:*OTOA*. **Exons**: NM_144672.3:1-19. **Variants(1)**: c.1320+2T>C

Autosomal Recessive Deafness 23. Gene:*PCDH15*. **Exons**: NM_033056.3:2-33. **Variants(3)**: c.785G>A, c.400C>G, c.1583T>A

Autosomal Recessive Deafness 24. Gene:*RDX*. **Exons**: NM_002906.3:2-14. **Variants(3)**: c.1405dupG, c.1732G>A, c.463C>T

Autosomal Recessive Deafness 25. Gene:*GRXCR1*. **Exons**: NM_001080476.2:1-4. **Variants(4)**: c.628-9C>A, c.412C>T, c.627+19A>T, c.229C>T

Autosomal Recessive Deafness 28. Gene:*TRIOBP*. **Exons**: NM_001039141.2:3-6,8-17,19,21-23. **Variants(6)**: c.3349C>T, c.1039C>T, c.2362C>T, c.1741C>T, c.889C>T, c.3202C>T

Autosomal Recessive Deafness 29. Gene:*CLDN14*. **Exons**: NM_144492.2:3. **Variants(3)**: c.254T>A, c.398delT, c.301G>A

Autosomal Recessive Deafness 3. Gene:*MYO15A*. **Exons**: NM_016239.3:3-66. **Variants(9)**: c.10573delA, c.8789-1G>C, c.5492G>T, c.3313G>T, c.3336delG, c.9958_9961delGACT, c.3756+1G>T, c.8148G>T, c.3685C>T

Autosomal Recessive Deafness 30. Gene:*MYO3A*. **Exons**: NM_017433.4:3-35. **Variants(3)**: c.1777-12G>A, c.732-2A>G, c.3129T>G

Autosomal Recessive Deafness 35. Gene:*ESRRB*. **Exons**: NM_004452.3:4-11. **Variants(2)**: c.1018_1024dupGAGTTTG, c.1024G>T

Autosomal Recessive Deafness 37. Gene:*MYO6*. **Exons**: NM_004999.3:2-35. **Variants(3)**: c.647A>T, c.3496C>T, c.36dupT

Autosomal Recessive Deafness 39. Gene:*HGF*. **Exons**: NM_000601.4:1-18. **Variants(1)**: c.495G>A

Autosomal Recessive Deafness 4, with Enlarged Vestibular Aqueduct. Gene:*SLC26A4*. **Exons**: NM_000441.1:3-21. **Variants(94)**: c.1803G>A, c.2219G>T, c.1334T>G, c.1341+1delG, c.322delC, c.919-2A>G, c.2228T>A, c.1595G>T, c.916dupG, c.170C>A, c.1863delT, c.2168A>G, c.941C>A, c.1001+1G>A, c.281C>T, c.783_784insT, c.754T>C, c.2089+1G>A, c.440T>C, c.1160C>T, c.415+7A>G, c.1226G>A, c.1151A>G, c.1614+1G>A, c.2027T>A, c.1337A>G, c.1363A>T, c.1172G>A, c.2343A>G, c.1707+5G>A, c.589G>A, c.1246A>C, c.1409G>A, c.2173G>C, c.753_756delCTCT, c.1174A>T, c.365_366insT, c.304G>A, c.1198delT, c.1343C>T, c.707T>C, c.1588T>C, c.1229C>T, c.1615-1G>A, c.1975G>C, c.397T>A, c.626G>T, c.2080T>C, c.233A>G, c.1919G>A, c.395C>T, c.1105A>G, c.1541A>G, c.1589A>C, c.1149+3A>G, c.1079C>T, c.2127delT, c.2086C>T, c.1147delC, c.946G>T, c.-3-2A>G, c.230A>T, c.1392delG, c.1264-1G>C, c.165-1G>A, c.412G>T, c.716T>A, c.1667A>G, c.601-1G>A, c.918+1G>A, c.279T>A, c.1439T>A, c.2162C>T, c.1694G>A, c.1334_1335insAGTC, c.1371C>A, c.235C>T, c.2170G>A, c.407_411delTCTCA, c.2015G>A, c.665G>T, c.1985G>A, c.249G>A, c.269C>T, c.1536_1537delAG, c.890delC, c.2186T>C, c.279delT, c.129dupC, c.149T>G, c.68C>A, c.84C>A, c.85G>C, c.87G>C

Autosomal Recessive Deafness 49. Gene:*MARVELD2*. **Exons**: NM_001038603.2:2-7. **Variants(5)**: c.1331+1G>A, c.1183-1G>A, c.1498C>T, c.1331+2T>C, c.1331+2_1331+5delTGAG

Autosomal Recessive Deafness 59. Gene:*DFNB59*. **Exons**: NM_001042702.3:2-7. **Variants(7)**: c.988delG, c.726delT, c.161C>T, c.547C>T, c.499C>T, c.122delA, c.113dupT

Autosomal Recessive Deafness 6. Gene:*TMIE*. **Exons**: NM_147196.2:2-4. **Variants(3)**: c.170G>A, c.241C>T, c.250C>T

Autosomal Recessive Deafness 61. Gene:*SLC26A5*. **Exons**: NM_198999.2:3-20. **Variants(3)**: c.390A>C, c.209G>A, c.-53-2A>G

Autosomal Recessive Deafness 63. Gene:*LRTOMT*. **Exons**: NM_001145308.4:3-7. **Variants(5)**: c.242G>A, c.313T>C, c.333C>G, c.328G>A, c.358+4A>C

Autosomal Recessive Deafness 67. Gene:*LHFPL5*. **Exons**: NM_182548.3:1-3. **Variants(4)**: c.250delC, c.380A>G, c.649delG, c.494C>T

Autosomal Recessive Deafness 7/11. Gene:*TMC1*. **Exons**: NM_138691.2:5-24. **Variants(4)**: c.1543T>C, c.1960A>G, c.1165C>T, c.100C>T

Autosomal Recessive Deafness 77. Gene:*LOXHD1*. **Exons**: NM_144612.6:1-40. **Variants(2)**: c.4714C>T, c.2008C>T

Autosomal Recessive Deafness 79. Gene:*TPRN*. **Exons**: NM_001128228.2:2-4. **Variants(2)**: c.1427delC, c.1239G>A

Autosomal Recessive Deafness 8/10. Gene:*TMPRSS3*. **Exons**: NM_024022.2:2-13. **Variants(4)**: c.647G>T, c.753G>C, c.208delC, c.1211C>T

Autosomal Recessive Deafness 9. Gene:*OTOF*. **Exons**: NM_194248.2:1-46. **Variants(8)**: c.1544T>C, c.3032T>C, c.1778delT, c.766-2A>G, c.2485C>T, c.2348delG, c.4491T>A, c.5816G>A

Autosomal Recessive Distal Spinal Muscular Atrophy 1. Gene:*IGHMBP2*. **Exons**: NM_002180.2:2-15. **Variants(7)**: c.675delT, c.638A>G, c.1738G>A, c.121C>T, c.2362C>T, c.707T>G, c.2611+1G>T

Autosomal Recessive Distal Spinal Muscular Atrophy 4. Gene:*PLEKHG5*. **Exons**: NM_020631.4:2-21. **Variants(1)**: c.1940T>C

Autosomal Recessive Hypophosphatemic Rickets 1. Gene:*DMP1*. **Exons**: NM_004407.3:2-6. **Variants(3)**: c.362delC, c.1A>G, c.55-1G>C

Autosomal Recessive Hypophosphatemic Rickets 2. Gene:*ENPP1*. **Exons**: NM_006208.2:2-25. **Variants(2)**: c.2702A>C, c.797G>T

Autosomal Recessive Neutropenia, Severe Congenital 4. Gene:*G6PC3*. **Exons**: NM_138387.3:1-6. **Variants(7)**: c.778G>C, c.758G>A, c.346A>G, c.141C>G, c.935_936insT, c.784G>C, c.554T>C

Autosomal Recessive Osteopetrosis 1. Gene:*TCIRG1*. **Exons**: NM_006019.3:2-20. **Variants(4)**: c.2236+1G>A, c.1331G>T, c.1213G>A, c.1392C>A

Autosomal Recessive Osteopetrosis 3. Gene:*CA2*. **Exons**: NM_000067.2:2-7. **Variants(3)**: c.232+1G>A, c.120T>G, c.319C>T

Autosomal Recessive Osteopetrosis 4. Gene:*CLCN7*. **Exons**: NM_001287.5:2-25. **Variants(3)**: c.2285G>A, c.1663C>T, c.2297T>C

Autosomal Recessive Osteopetrosis 5. Gene:*OSTM1*. **Exons**: NM_014028.3:1-6. **Variants(1)**: c.415_416delAG

Autosomal Recessive Polycystic Kidney Disease. Gene:*PKHD1*. **Exons**: NM_138694.3:2-67. **Variants(137)**: c.8579_8583delACAGT, c.711_714delAATG, c.1626_1629delACTT, c.9464A>G, c.10412T>G, c.10219C>T, c.528-2A>G, c.5323C>T, c.10136delC, c.8068T>C, c.10765C>T, c.5378_5380+1delATGG, c.737T>C, c.2269A>C, c.8588A>G, c.9707delC, c.4330C>T, c.10628_10635delTGTATGTT, c.6333-2A>G, c.10658T>C, c.1057C>T, c.2810G>A, c.3467C>T, c.5895dupA, c.8312T>A, c.2936C>T, c.1880T>A, c.2542T>A, c.982C>T, c.1095G>A, c.9348delG, c.5498C>T, c.6992T>A, c.9689delA, c.5993T>C, c.10728G>A, c.9765G>A, c.3306delT, c.2532C>A, c.7477C>T, c.3848C>A, c.664A>G, c.977G>T, c.7177C>T, c.9319C>T, c.5485C>T, c.10856delA, c.3958_3959delGG, c.4256_4257delGG, c.10637delT, c.5375delT, c.474G>A, c.3367G>A, c.6097A>G, c.6029delA, c.9861C>A, c.2279G>A, c.10240delA, c.6907A>T, c.107C>T, c.11074C>T, c.10364delC, c.11506+1G>A, c.9683C>A, c.3118C>T, c.2341C>T, c.11612G>A, c.8302+2T>C, c.1486C>T, c.383delC, c.5750A>C, c.10972_10973delAT, c.6209G>A, c.2520_2526delATACACT, c.7264T>G, c.8829_8830insG, c.5748_5749delTC, c.10031T>G, c.8011C>T, c.10077delG, c.1411_1412delGT, c.11524C>T, c.10637_10638insG, c.9718C>T, c.10444C>T, c.7916C>A, c.2264C>T, c.10174C>T, c.9719G>A, c.9239G>A, c.8909_8912delTTGT, c.8642+1G>A, c.7912-1G>C, c.8870T>C, c.3761_3762delCCinsG, c.2216C>T, c.7351-2A>T, c.53-3C>A, c.3229-2A>C, c.1418T>G, c.6929delG, c.2141-1G>T, c.881-1G>A, c.4870C>T, c.977-1G>A, c.4457C>T, c.5075G>A, c.9530T>C, c.353delG, c.2854G>A, c.5646delT, c.1690C>T, c.370C>T, c.5513A>G, c.2812_2813delTA, c.6296_6297delTG, c.8518C>T, c.1233+1G>A, c.8935C>T, c.5381-2A>C, c.3747T>G, c.9347delT, c.4220T>G, c.6383delT, c.976+2T>A, c.1937G>A, c.8114delG, c.5825A>G, c.9370C>T, c.4751G>T, c.1774C>T, c.5237-1G>A, c.1830T>A, c.2179_2180insT, c.3364G>A, c.10709C>G, c.8824C>T

Autosomal Recessive Segawa Syndrome. Gene:*TH*. **Exons**: NM_000360.3:1-13. **Variants(6)**: c.605G>A, c.614T>C, c.1388C>T, c.917G>A, c.1141C>A, c.733A>C

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. Gene:*SACS*. **Exons**: NM_014363.4:2,4-10. **Variants(4)**: c.8844delT, c.7504C>T, c.12160C>T, c.10907G>A

Autosomal Recessive Spastic paraplegia 11. Gene:*SPG11*. **Exons**: NM_025137.3:1-40. **Variants(8)**: c.6100C>T, c.733_734delAT, c.7152-1G>C, c.5623C>T, c.2472_2473insT, c.529_533delATATT, c.442+1G>C, c.118C>T

Autosomal Recessive Spastic paraplegia 15. Gene:*ZFYVE26*. **Exons**: NM_015346.3:2-42. **Variants(4)**: c.5485-1G>A, c.5422C>T, c.4312C>T, c.1477C>T

Autosomal Recessive Spastic paraplegia 20. Gene:*SPG20*. **Exons**: NM_015087.4:2-9. **Variants(2)**: c.364_365delAT, c.1110delA

Autosomal Recessive Spastic paraplegia 7. Gene:*SPG7*. **Exons**: NM_003119.2:2-17. **Variants(10)**: c.1617delC, c.2216_2217insA, c.1045G>A, c.1529C>T, c.1749G>C, c.1742_1744delTGG, c.850_851delTTinsC, c.233T>A, c.784_785delGC, c.2075G>C

Bardet-Biedl syndrome 1. Gene:*BBS1*. **Exons**: NM_024649.4:1-17. **Variants(1)**: c.1169T>G

Bardet-Biedl syndrome 10. Gene:*BBS10*. **Exons**: NM_024685.3:1-2. **Variants(1)**: c.217_218insT

Bartter Syndrome Type 1. Gene:*SLC12A1*. **Exons**: NM_000338.2:2-27. **Variants(3)**: c.1942G>A, c.814G>T, c.1875G>A

Bartter Syndrome Type 2. Gene:*KCNJ1*. **Exons**: NM_000220.3:1-2. **Variants(8)**: c.592G>A, c.641C>T, c.372T>A, c.500G>A, c.237C>G, c.657C>G, c.80G>A, c.322G>C

Bartter Syndrome Type 4A. Gene:*BSND*. **Exons**: NM_057176.2:1-4. **Variants(8)**: c.139G>A, c.28G>A, c.3G>A, c.35T>C, c.1A>T, c.23G>T, c.10G>T, c.22C>T

Bestrophinopathy. Gene:*BEST1*. **Exons**: NM_004183.3:2-11. **Variants(4)**: c.422G>A, c.122T>C, c.949G>A, c.598C>T

Beta-Ketothiolase Deficiency. Gene:*ACAT1*. **Exons**: NM_000019.3:1-12. **Variants(11)**: c.433C>G, c.2T>A, c.935T>C, c.1083dupA, c.997G>C, c.814C>T, c.1136G>T, c.547G>A, c.1035_1037delAGA, c.1138G>A, c.278A>G

Beta-thalassemia. Gene:*HBB*. **Exons**: NM_000518.4:1-3. **Variants(110)**: c.216_217insT, c.79G>A, c.170G>A, c.316-3C>A, c.20A>T, c.364G>A, c.316-106C>G, c.4delG, c.111_118delTTGGACCC, c.1A>G, c.27dupG, c.52A>T, c.19G>T, c.92+5G>C, c.226delC, c.59A>G, c.92+5G>T, c.212C>G, c.215_216insA, c.114_120delGACCCAG, c.93-1G>C, c.315+1G>A, c.176_177insC, c.113G>A, c.92+1G>T, c.316-14T>G, c.110delC, c.22_24delGAG, c.92+2T>A, c.2T>A, c.3G>T, c.230delC, c.108C>A, c.74delGinsCAC, c.82G>T, c.165delT, c.94delC, c.316-2A>G, c.93-3T>G, c.316-146T>G, c.349_350insTGAT, c.17_18delCT, c.332T>C, c.68_74delAAGTTGG, c.316-2A>C, c.2T>G, c.112delT, c.316-197C>T, c.-50-u30T>A, c.25_26delAA, c.47G>A, c.92G>C, c.189_195delTCATGGC, c.2T>C, c.315+1G>T, c.93-21G>A, c.36delT, c.-50-u31A>G, c.77_79delGTG, c.19G>A, c.380T>G, c.85_86insC, c.322_323insC, c.48G>A, c.364G>C, c.-50-u29A>G, c.425_433delTGGCCCACA, c.138delT, c.316-7C>G, c.75T>A, c.315+2delT, c.-43C>T, c.20delA, c.54_59delGGTGAA, c.92+1G>A, c.316-1G>T, c.287_288insA, c.78dupT, c.114G>A, c.118C>T, c.92+2T>C, c.3G>C, c.*110_*111delTA, c.92+6T>C, c.28_29insTA, c.315+1G>C, c.93-2A>G, c.-50-u28A>G, c.-50-u87C>T, c.-29G>A, c.-50-u87C>G, c.380T>A, c.93-1G>A, c.*110T>C, c.203_204delTG, c.135delC, c.126_129delCTTT, c.79G>T, c.67G>T, c.143_146dupATCT, c.184A>T, c.-50A>C, c.235delC, c.3G>A, c.45dupG, c.93-2A>C, c.92+2T>G, c.92+5G>A, c.178A>T, c.217dupA

Bietti Crystalline Dystrophy. Gene:*CYP4V2*. **Exons**: NM_207352.3:2-11. **Variants(2)**: c.327+1G>A, c.1091-2A>G

Biotinidase Deficiency. Gene:*BTD*. **Exons**: NM_000060.2:1-4. **Variants(8)**: c.1612C>T, c.235C>T, c.100G>A, c.1595C>T, c.511G>A, c.755A>G, c.98_104delGCGGCTGinsTCC, c.1368A>C

Bjornstad Syndrome. Gene:*BCS1L*. **Exons**: NM_004328.4:3-9. **Variants(3)**: c.548G>A, c.103G>C, c.550C>T

Bloom syndrome. Gene:*BLM*. **Exons**: NM_000057.2:2-22. **Variants(1)**: c.2207_2212delATCTGAinsTAGATTC

Bothnia Type Retinitis Pigmentosa. Gene:*RLBP1*. **Exons**: NM_000326.4:3-9. **Variants(1)**: c.700C>T

Brittle Cornea Syndrome. Gene:*ZNF469*. **Exons**: NM_001127464.1:1-2. **Variants(1)**: c.4174G>T

C3 deficiency. Gene:*C3*. **Exons**: NM_000064.2:1-41. **Variants(5)**: c.1004-2A>T, c.2354+1G>A, c.3116dupT, c.1655G>A, c.1119+1G>T

Canavan Disease. Gene:*ASPA*. **Exons**: NM_000049.2:1-6. **Variants(5)**: c.654C>A, c.433-2A>G, c.854A>C, c.693C>A, c.914C>A

CARASIL Syndrome. Gene:*HTRA1*. **Exons**: NM_002775.4:2-9. **Variants(4)**: c.1108C>T, c.904C>T, c.889G>A, c.754G>A

Carbamoylphosphate Synthetase I Deficiency. Gene:*CPS1*. **Exons**: NM_001875.4:1-38. **Variants(5)**: c.130C>T, c.1631C>T, c.2945G>A, c.2359C>T, c.1010A>G

Carnitine Palmitoyltransferase I Deficiency. Gene:*CPT1A*. **Exons**: NM_001876.3:2-19. **Variants(6)**: c.1493A>G, c.1361A>G, c.1079A>G, c.298C>T, c.2126G>A, c.1241C>T

Carnitine Palmitoyltransferase II Deficiency. Gene:*CPT2*. **Exons**: NM_000098.2:1-5. **Variants(11)**: c.1657G>A, c.1891C>T, c.452G>A, c.520G>A, c.359A>G, c.1507C>T, c.1148T>A, c.680C>T, c.338C>T, c.1883A>C, c.370C>T

Carpenter Syndrome. Gene:*RAB23*. **Exons**: NM_183227.1:2-7. **Variants(1)**: c.434T>A

Cartilage-hair hypoplasia. Gene:*RMRP*. **Exons**: NR_003051.3:1. **Variants(1)**: n.71A>G

CC2D2A-Related COACH Syndrome. Gene:*CC2D2A*. **Exons**: NM_001080522.2:3-38. **Variants(1)**: c.3145C>T

Central Core Disease. Gene:*RYR1*. **Exons**: NM_000540.2:1-90,92-106. **Variants(2)**: c.14545G>A, c.10579C>T

Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma Syndrome. Gene:*SNAP29*. **Exons**: NM_004782.3:1-5. **Variants(1)**: c.487dupA

Cerebrooculofacioskeletal Syndrome 1. Gene:*ERCC6*. **Exons**: NM_000124.2:2-21. **Variants(2)**: c.2047C>T, c.3862C>T

Cerebrotendinous xanthomatosis. Gene:*CYP27A1*. **Exons**: NM_000784.3:1-9. **Variants(6)**: c.1435C>G, c.1016C>T, c.1421G>A, c.1420C>T, c.434G>A, c.1214G>A

Charcot-Marie-Tooth disease Type 1F/Type 2E. Gene:*NEFL*. **Exons**: NM_006158.3:1-4. **Variants(2)**: c.628G>T, c.418G>T

Charcot-Marie-Tooth disease Type 2B1. Gene:*LMNA*. **Exons**: NM_170707.3:1-12. **Variants(1)**: c.892C>T

Charcot-Marie-Tooth disease Type 2B2. Gene:*MED25*. **Exons**: NM_030973.3:1-18. **Variants(1)**: c.1004C>T

Charcot-Marie-Tooth disease Type 4A. Gene:*GDAP1*. **Exons**: NM_018972.2:1-6. **Variants(5)**: c.581C>G, c.92G>A, c.715C>T, c.844C>T, c.487C>T

Charcot-Marie-Tooth disease Type 4B1. Gene:*MTMR2*. **Exons**: NM_016156.5:1-15. **Variants(3)**: c.1276C>T, c.826G>T, c.1444C>T

Charcot-Marie-Tooth disease Type 4B2. Gene:*SBF2*. **Exons**: NM_030962.3:2-40. **Variants(3)**: c.1459C>T, c.3586C>T, c.2875C>T

Charcot-Marie-Tooth disease Type 4C. Gene:*SH3TC2*. **Exons**: NM_024577.3:1-17. **Variants(20)**: c.2710C>T, c.530-2A>G, c.3325C>T, c.3341delC, c.1982T>C, c.2829T>G, c.1178-1G>A, c.1747_1748delAG, c.3601C>T, c.217_227delGCTGCTCGGAGinsCCAGTAA, c.505T>C, c.2860C>T, c.2491_2492delAG, c.920G>A, c.1969G>A, c.3326G>C, c.1586G>A, c.2191delG, c.28delG, c.1972C>T

Charcot-Marie-Tooth disease Type 4D. Gene:*NDRG1*. **Exons**: NM_006096.3:2-16. **Variants(2)**: c.442C>T, c.538-1G>A

Charcot-Marie-Tooth disease Type 4F. Gene:*PRX*. **Exons**: NM_181882.2:4-7. **Variants(5)**: c.2098delG, c.2145T>A, c.3208C>T, c.1951G>A, c.586C>T

Charcot-Marie-Tooth disease Type 4H. Gene:*FGD4*. **Exons**: NM_139241.2:3-17. **Variants(7)**: c.823C>T, c.893T>G, c.670C>T, c.1628_1629delAG, c.1756G>T, c.1325G>A, c.893T>C

Charcot-Marie-Tooth disease Type 4J. Gene:*FIG4*. **Exons**: NM_014845.5:1-23. **Variants(2)**: c.547C>T, c.122T>C

Chorea-Acanthocytosis. Gene:*VPS13A*. **Exons**: NM_033305.2:1-72. **Variants(2)**: c.269T>A, c.622C>T

Citrullinemia Type I. Gene:*ASS1*. **Exons**: NM_000050.4:3-16. **Variants(13)**: c.910C>T, c.1085G>T, c.421-2A>G, c.970G>A, c.40G>A, c.794G>A, c.1168G>A, c.257G>A, c.835C>T, c.539G>A, c.470G>A, c.970+5G>A, c.1087C>T

Citrullinemia Type II. Gene:*SLC25A13*. **Exons**: NM_014251.2:1-18. **Variants(12)**: c.674C>A, c.1078C>T, c.852_855delTATG, c.1799dupA, c.1311+1G>A, c.615+1G>C, c.1801G>T, c.615+5G>A, c.1592G>A, c.1177+1G>A, c.1813C>T, c.1801G>A

Cockayne Syndrome Type A. Gene:*ERCC8*. **Exons**: NM_000082.3:1-12. **Variants(3)**: c.37G>T, c.966C>A, c.479C>T

Cockayne Syndrome Type B. Gene:*ERCC6*. **Exons**: NM_000124.2:2-21. **Variants(4)**: c.1550G>A, c.3592_3593insGA, c.2203C>T, c.1357C>T

Cohen syndrome. Gene:*VPS13B*. **Exons**: NM_017890.4:2-62. **Variants(1)**: c.3348_3349delCT

COL17A1-Related Junctional Epidermolysis Bullosa. Gene:*COL17A1*. **Exons**: NM_000494.3:2-56. **Variants(15)**: c.1706delC, c.3676C>T, c.433C>T, c.1898G>A, c.2336-2A>G, c.2944_2947+1delGAAGG, c.2383C>T, c.520_521delAG, c.2564T>G, c.2965delA, c.3908G>A, c.4003_4004delGG, c.4150_4151insG, c.2336-1G>T, c.3067C>T

COL4A3-Related Autosomal Recessive Alport Syndrome. Gene:*COL4A3*. **Exons**: NM_000091.4:2-52. **Variants(3)**: c.4441C>T, c.4420_4424delCTTTT, c.4571C>G

COL4A4-Related Autosomal Recessive Alport Syndrome. Gene:*COL4A4*. **Exons**: NM_000092.4:2-48. **Variants(5)**: c.3601G>A, c.4129C>T, c.3713C>A, c.4923C>A, c.4715C>T

Combined Oxidative Phosphorylation Deficiency 1. Gene:*GFM1*. **Exons**: NM_024996.5:1-18. **Variants(3)**: c.1487T>G, c.748C>T, c.139C>T

Combined Oxidative Phosphorylation Deficiency 2. Gene:*MRPS16*. **Exons**: NM_016065.3:1-3. **Variants(1)**: c.331C>T

Combined Oxidative Phosphorylation Deficiency 3. Gene:*TSFM*. **Exons** : NM_001172696.1:1-4,6-7. **Variants(1)**: c.997C>T

Combined Oxidative Phosphorylation Deficiency 5. Gene:*MRPS22*. **Exons**: NM_020191.2:1-8. **Variants(2)**: c.509G>A, c.644T>C

Combined pituitary hormone deficiency 1. Gene:*POU1F1*. **Exons**: NM_000306.2:1-6. **Variants(10)**: c.472G>C, c.433A>T, c.515G>A, c.748G>T, c.514C>T, c.688G>A, c.577T>C, c.404T>G, c.428G>A, c.715C>T

Combined pituitary hormone deficiency 2. Gene:*PROP1*. **Exons**: NM_006261.4:1-3. **Variants(17)**: c.349T>A, c.150delA, c.263T>C, c.310delC, c.358C>T, c.343-11C>G, c.157delA, c.247C>T, c.218G>A, c.301_302delAG, c.469_470insT, c.109+1G>T, c.112_124delTCGAGTGCTCCAC, c.2T>C, c.373C>T, c.295C>T, c.217C>T

Combined pituitary hormone deficiency 3. Gene:*LHX3*. **Exons**: NM_014564.3:2-3,5-6. **Variants(3)**: c.347A>G, c.687G>A, c.644C>T

Combined SAP Deficiency. Gene:*PSAP*. **Exons**: NM_002778.2:1-14. **Variants(1)**: c.1A>T

Cone Dystrophy 4. Gene:*PDE6C*. **Exons**: NM_006204.3:1-22. **Variants(12)**: c.85C>T, c.2457T>A, c.633G>C, c.256_257insAG, c.1483-2A>G, c.826C>T, c.2368G>A, c.481-12T>A, c.967T>A, c.1805A>T, c.1682dupA, c.1363A>G

Cone-rod Dystrophy 3. Gene:*ABCA4*. **Exons**: NM_000350.2:1-50. **Variants(6)**: c.2616_2617delCT, c.5714+5G>A, c.5285C>A, c.3540_3555delGTCTAAGGGTTTCTCC, c.2888delG, c.5461-10T>C

Congenital Amegakaryocytic Thrombocytopenia. Gene:*MPL*. **Exons**: NM_005373.2:1-12. **Variants(5)**: c.823C>A, c.1473G>A, c.769C>T, c.305G>C, c.556C>T

Congenital Bile Acid Synthesis Defect 3. Gene:*CYP7B1*. **Exons**: NM_004820.3:2-6. **Variants(1)**: c.1162C>T

Congenital Bile Acid Synthesis Defect 4. Gene:*AMACR*. **Exons**: NM_014324.5:1-5. **Variants(1)**: c.320T>C

Congenital Diarrhea 4, Malabsorptive. Gene:*NEUROG3*. **Exons**: NM_020999.3:2. **Variants(2)**: c.278G>T, c.319C>A

Congenital Disorders of Glycosylation Ia. Gene:*PMM2*. **Exons**: NM_000303.2:1-8. **Variants(21)**: c.422G>A, c.669C>G, c.563A>G, c.338C>T, c.722G>C, c.691G>A, c.647A>T, c.95T>G, c.620T>C, c.484C>T, c.193G>T, c.677C>G, c.349G>C, c.131T>C, c.26G>A, c.385G>A, c.710C>G, c.357C>A, c.395T>C, c.368G>A, c.317A>T

Congenital Disorders of Glycosylation Ib. Gene:*MPI*. **Exons**: NM_002435.1:1-8. **Variants(4)**: c.656G>A, c.305C>T, c.413T>C, c.884G>A

Congenital Disorders of Glycosylation Ic. Gene:*ALG6*. **Exons**: NM_013339.3:2-15. **Variants(3)**: c.897_899delAAT, c.1432T>C, c.998C>T

Congenital Disorders of Glycosylation Ie. Gene:*DPM1*. **Exons**: NM_003859.1:1-9. **Variants(2)**: c.628delC, c.274C>G

Congenital Disorders of Glycosylation IIa. Gene:*MGAT2*. **Exons**: NM_002408.3:1. **Variants(3)**: c.869C>T, c.785A>G, c.1017T>A

Congenital Disorders of Glycosylation IIc. Gene:*SLC35C1*. **Exons**: NM_018389.4:1-2. **Variants(2)**: c.923C>G, c.439C>T

Congenital Disorders of Glycosylation IId. Gene:*B4GALT1*. **Exons**: NM_001497.3:1-6. **Variants(1)**: c.1031dupC

Congenital Disorders of Glycosylation IIf. Gene:*SLC35A1*. **Exons**: NM_006416.4:2-8. **Variants(1)**: c.277_280delGTGCinsTG

Congenital Disorders of Glycosylation Ij. Gene:*DPAGT1*. **Exons**: NM_001382.3:1-9. **Variants(1)**: c.509A>G

Congenital Disorders of Glycosylation Ik. Gene:*ALG1*. **Exons**: NM_019109.4:1-9,11,13. **Variants(2)**: c.450C>G, c.434G>A

Congenital Disorders of Glycosylation It. Gene:*PGM1*. **Exons**: NM_002633.2:1-11. **Variants(2)**: c.361G>C, c.1507C>T

Congenital Erythropoietic Porphyria. Gene:*UROS*. **Exons**: NM_000375.2:2-10. **Variants(1)**: c.217T>C

Congenital Generalized Lipodystrophy Type 2. Gene:*BSCL2*. **Exons**: NM_032667.6:2-11. **Variants(5)**: c.412C>T, c.823C>T, c.671+5G>A, c.672-3C>G, c.634G>C

Congenital Hypomyelinating Neuropathy 1. Gene:*EGR2*. **Exons**: NM_000399.3:1-2. **Variants(1)**: c.803T>A

Congenital Hypothryoidism Nongoitrous 1. Gene:*TSHR*. **Exons**: NM_000369.2:1-10. **Variants(13)**: c.970C>T, c.1657G>A, c.1798T>C, c.122G>C, c.500T>A, c.1228G>A, c.928C>T, c.326G>A, c.484C>G, c.1637G>A, c.1170T>G, c.1575C>A, c.202C>T

Congenital Hypothryoidism Nongoitrous 4. Gene:*TSHB*. **Exons**: NM_000549.3:2-3. **Variants(3)**: c.205C>T, c.145G>A, c.94G>T

Congenital myasthenic Syndrome with tubular aggregates 2. Gene:*DPAGT1*. **Exons**: NM_001382.3:1-9. **Variants(3)**: c.349G>A, c.358C>A, c.791T>G

Congenital Plasminogen Deficiency. Gene:*PLG*. **Exons**: NM_000301.3:1-18. **Variants(6)**: c.112A>G, c.1848G>A, c.1120G>T, c.704G>A, c.693_695delGAA, c.1435G>T

Corneal Endothelial Dystrophy. Gene:*SLC4A11*. **Exons**: NM_032034.3:1-19. **Variants(14)**: c.1813C>T, c.2240_2240+1insTATGACAC, c.637T>C, c.473_480delGCTTCGCC, c.2233_2240dupTATGACAC, c.2605C>T, c.2528T>C, c.1463G>A, c.1466C>T, c.1391G>A, c.2264G>A, c.353_356delAGAA, c.2606G>A, c.2566A>G

Crigler-Najjar Syndrome. Gene:*UGT1A1*. **Exons**: NM_000463.2:1-5. **Variants(5)**: c.674T>G, c.1021C>T, c.524T>A, c.44T>G, c.991C>T

Crisponi Syndrome. Gene:*CRLF1*. **Exons**: NM_004750.4:2-6,8-9. **Variants(12)**: c.538C>T, c.829C>T, c.413C>T, c.303delC, c.527+5G>A, c.713_714insC, c.852G>T, c.708_709delCCinsT, c.935G>A, c.845_846delTG, c.226T>G, c.676dupA

Cystic Fibrosis. Gene:*CFTR*. **Exons**: NM_000492.3:1-27. **Variants(151)**: c.1397C>A, c.3752G>A, c.579+1G>T, c.3937C>T, c.2583delT, c.1397C>G, c.1911delG, c.1209+1G>A, c.948delT, c.1130_1131insA, c.2834C>T, c.1007T>A, c.2668C>T, c.273+1G>A, c.3276C>G, c.3744delA, c.2464G>T, c.223C>T, c.3154T>G, c.328G>C, c.1680-1G>A, c.1766+1G>A, c.1A>G, c.3846G>A, c.3528delC, c.366T>A, c.2988G>A, c.2657+5G>A, c.803delA, c.200C>T, c.1865G>A, c.3310G>T, c.178G>T, c.2175_2176insA, c.3454G>C, c.1327_1330dupGATA, c.1766+3A>G, c.262_263delTT, c.164+12T>C, c.2551C>T, c.1022_1023insTC, c.3302T>A, c.2052delA, c.3276C>A, c.2780T>C, c.3873+1G>A, c.3230T>C, c.273+3A>C, c.3536_3539delCCAA, c.1585-8G>A, c.2538G>A, c.325_327delTATinsG, c.4077_4080delTGTTinsAA, c.489+1G>T, c.3197G>A, c.3587C>G, c.3140-26A>G, c.3717+4A>G, c.1393-1G>A, c.274G>A, c.1466C>A, c.1075C>A, c.613C>T, c.1572C>A, c.349C>T, c.2249C>T, c.115C>T, c.1055G>A, c.3659delC, c.3764C>A, c.292C>T, c.1675G>A, c.2537G>A, c.254G>A, c.1203G>A, c.350G>A, c.1079C>A, c.2128A>T, c.1202G>A, c.1766+1G>T, c.3612G>A, c.1679G>A, c.658C>T, c.4251delA, c.2491G>T, c.2052_2053insA, c.2988+1G>A, c.1645A>C, c.1081delT, c.3712C>T, c.3484C>T, c.1624G>T, c.1923_1931delCTCAAAACTinsA, c.595C>T, c.442delA, c.579+3A>G, c.1519_1521delATC, c.2125C>T, c.1654C>T,

c.3196C>T, c.2051_2052delAAinsG, c.2490+1G>A, c.2453delT, c.617T>G, c.3909C>G, c.805_806delAT, c.1013C>T, c.3611G>A, c.3266G>A, c.1679G>C, c.2195T>G, c.580-1G>T, c.1585-1G>A, c.1477C>T, c.772A>G, c.1545_1546delTA, c.1475C>T, c.1766+5G>T, c.1647T>G, c.3773_3774insT, c.1558G>T, c.1156_1157insTA, c.2215delG, c.1364C>A, c.2875delG, c.2012delT, c.274-1G>A, c.1116+1G>A, c.1646G>A, c.988G>T, c.531delT, c.2989-1G>A, c.2290C>T, c.1438G>T, c.1040G>A, c.1721C>A, c.274G>T, c.1657C>T, c.1040G>C, c.722_743delGGAGAATGATGATGAAGTACAG, c.1652G>A, c.2039delC, c.3731G>A, c.3194T>C, c.1521_1523delCTT, c.2869_2870insG, c.3889_3890insT, c.579+5G>A, c.3472C>T, c.1753G>T, c.1000C>T

D-2-Hydroxyglutaric Aciduria. Gene:*D2HGDH*. **Exons**: NM_152783.3:2-10. **Variants(3)**: c.491-2A>G, c.440T>G, c.1315A>G

D-Bifunctional Protein Deficiency. Gene:*HSD17B4*. **Exons**: NM_000414.3:1-24. **Variants(5)**: c.46G>A, c.423_424delGA, c.650A>G, c.1369A>T, c.317G>C

DCLRE1C-Related Omenn Syndrome. Gene:*DCLRE1C*. **Exons**: NM_001033855.1:1-7,9-14. **Variants(1)**: c.2T>C

Dejerine-Sottas disease, autosomal recessive. Gene:*PRX*. **Exons**: NM_181882.2:4-7. **Variants(3)**: c.247delC, c.2857C>T, c.1102C>T

Dent Disease. Gene:*OCRL*. **Exons**: NM_000276.3:2-24. **Variants(2)**: c.166_167delAT, c.2530C>T

Diastrophic Dysplasia. Gene:*SLC26A2*. **Exons**: NM_000112.3:2-3. **Variants(4)**: c.1724delA, c.-26+2T>C, c.1361A>C, c.1957T>A

Dihydropyrimidine Dehydrogenase Deficiency. Gene:*DPYD*. **Exons**: NM_000110.3:1-23. **Variants(1)**: c.1905+1G>A

Donnai-Barrow Syndrome. Gene:*LRP2*. **Exons**: NM_004525.2:2-79. **Variants(13)**: c.8519_8522delATTT, c.770-2A>G, c.1341+2T>G, c.9484_9485delGT, c.6160G>A, c.2640-1G>A, c.8452+1G>A, c.13139_13140insC, c.11469_11472delTTTG, c.10195C>T, c.7564T>C, c.9358_9359delAG, c.1093C>T

Donohue Syndrome. Gene:*INSR*. **Exons**: NM_000208.2:2-22. **Variants(4)**: c.1114C>T, c.1378A>G, c.698T>C, c.172G>A

Duchenne Muscular Dystrophy. Gene:*DMD*. **Exons**: NM_004006.2:1-79. **Variants(903)**: c.6223C>T, c.10018T>C, c.1332-9A>G, c.3121C>T, c.4558G>T, c.4711A>T, c.10019G>A, c.10033C>T, c.7339C>T, c.187-1G>C, c.8754delG, c.440C>A, c.8391-1G>C, c.3679C>T, c.5985T>G, c.1132C>T, c.2974C>T, c.7542+1G>A, c.7672C>T, c.3464_3471delGTTTGGAG, c.9663delA, c.9739C>T, c.3151C>T, c.903C>A, c.4841delG, c.10202T>G, c.8713C>T, c.2368C>T, c.7392delC, c.10606delC, c.1594C>T, c.9691C>T, c.2365G>T, c.5314A>T, c.6913-2A>G, c.5287C>T, c.3562A>T, c.357+1G>C, c.1633A>T, c.2776C>T, c.5401_5402delAT, c.409G>T, c.8686A>T, c.9649+2T>C, c.9001C>T, c.721C>T, c.1483-1G>C, c.1603-1G>A, c.748G>T, c.10554-2A>G, c.3259C>T, c.4898_4899delAG, c.7739delA, c.7873delC, c.3274A>T, c.10855C>T, c.9176delG, c.1978_1979delAA, c.1952G>A, c.10563delA, c.3365_3366delAG, c.2803+1G>A, c.1865C>G, c.9807+4delA, c.4057G>T, c.9974+2T>A, c.4084C>T, c.10141C>T, c.5563C>T, c.10094C>A, c.2866C>T, c.8938-9T>A, c.3427C>T, c.2991C>G, c.2638delC, c.1093C>T, c.745C>T, c.7542+2T>C, c.10135A>T, c.2475G>A, c.4087A>T, c.9600_9601delAA, c.3603+2T>G, c.4757G>A, c.9974+1G>T, c.488G>A, c.8176G>T, c.8774G>A, c.5800G>T, c.7247delT, c.6730C>T, c.313A>T, c.1324C>T, c.5209C>T, c.2128A>T, c.5266C>T, c.1527_1530delTCTC, c.2299G>T, c.9100C>T, c.3244G>T, c.4519-1G>C, c.8357G>A, c.9135delT, c.1873C>T, c.2407C>T, c.1663C>T, c.2281_2285delGAAAA, c.3430C>T, c.10088delC, c.4375C>T, c.2512C>T, c.4071+1G>A, c.1207G>T, c.2761delG, c.7229G>A, c.3125delA, c.6238C>T, c.2314G>T, c.7564C>T, c.2302C>T, c.8914C>T, c.6436A>T, c.689delT, c.9862G>T, c.6614+2T>C, c.6674T>G, c.5694_5697delAAAA, c.10477delC, c.4693C>T, c.3432+1G>T, c.10121delA, c.1615C>T, c.5845delC, c.5771_5772delAG, c.1555G>T, c.9649+5G>T, c.10219G>T, c.2956C>T, c.1235delT, c.6460C>T, c.9090delC, c.7054G>T, c.5637G>A, c.10651C>T, c.3470delA, c.1331+1G>T, c.7720C>T, c.4870C>T, c.2650C>T, c.1974delT, c.6200delC, c.2949+1G>T, c.7310-1G>A, c.676_678delAAG, c.9148C>T, c.6804_6807delACAA, c.3478_3479delGT, c.1474C>T, c.10969G>T, c.9380C>G, c.3196G>T, c.453T>A, c.9183G>A, c.5032C>T, c.4980delG, c.7755G>A, c.580C>T, c.6276C>G, c.3397G>T, c.4545_4549delGAAGT, c.6277A>T, c.568C>T, c.8608C>T, c.6592C>T, c.10086+5G>C, c.53delA, c.31+1G>T, c.7657C>T, c.9337C>T, c.2558T>A, c.883C>T, c.1061G>A, c.6790C>T, c.1087C>T, c.8944C>T, c.9563+1215A>G, c.583C>T, c.4151delA, c.4103delG, c.4852C>T, c.8807_8808delTC, c.6255G>A, c.2707G>T, c.4150G>T, c.433C>T, c.3033delC, c.5272_5273delTC, c.1682G>A, c.1388G>A, c.5653C>T, c.10279C>T, c.2236G>T, c.10498_10499delAG, c.2968C>T, c.10003G>C, c.9403C>T, c.8416C>T, c.4294C>T, c.10108C>T, c.1292G>A, c.516delC, c.1726delT, c.3940C>T, c.6614+1G>A, c.2869C>T, c.6393_6394delAA, c.5917C>T, c.3768delG, c.5766_5770delGAAAG, c.6373C>T, c.956C>G, c.8443C>T, c.1783G>T, c.7309+1G>A, c.1489C>T, c.4996C>T, c.5740-2A>G, c.503C>A, c.7582G>T, c.6439-1G>A, c.2804-2A>C, c.178C>T, c.4845+2T>C, c.7768_7771delGAAG, c.475_476delTT, c.3220G>T, c.10072G>T, c.6423C>A, c.9197C>A, c.3086G>A, c.4918delA, c.5530C>T, c.10203delA, c.3268C>T, c.5350G>T, c.9361+5G>C, c.10223+1G>C, c.6880A>T, c.8087delT, c.8214G>A, c.2227C>T, c.4147C>T, c.11G>A, c.7348delG, c.1990C>T, c.2737delG, c.4071+2T>A, c.9360C>A, c.433delC, c.998C>A, c.5461G>T, c.9072G>A, c.6292C>T, c.4414C>T, c.2791G>T, c.673A>T, c.9364delG, c.9461T>A, c.3147delA, c.3336delG, c.724C>T, c.3603+1G>T, c.9928C>T, c.6943G>T, c.8269G>T, c.9619_9626delTGTAAAGC, c.7401_7402delGGinsAT, c.1812delC, c.4108C>T, c.253C>T, c.2293-1G>T, c.9427C>T, c.6364G>T, c.58delA, c.572C>G, c.5154+2T>G, c.9986delT, c.965T>A, c.8074C>T, c.9959delC, c.10368delT, c.7105G>T, c.193G>T, c.10225_10229delCCCGT, c.9333delA, c.1619G>A, c.1149+2T>C, c.10722delC, c.8038C>T, c.2359delC, c.777delA, c.6955C>T, c.565C>T, c.10903C>T, c.9938G>T, c.9204_9207delCAAA, c.2701G>T, c.1683G>A, c.4518+5G>A, c.9649+1G>A, c.3469G>T, c.94-1G>A, c.772_773delCC, c.1533_1536delTCAC, c.8098A>T, c.1609G>T, c.8027+1G>A, c.7669delA, c.9563+1G>A, c.10171C>T, c.9109C>T, c.10126delC, c.2669T>G, c.530+1G>T, c.6391C>T, c.8728G>T, c.2276T>G, c.3188G>A, c.3337C>T, c.2622+1G>A, c.6720delT, c.6283C>T, c.2873C>G, c.3864delA, c.8287delC, c.2933_2934delGA, c.6544C>T, c.8064_8065delTA, c.9346C>T, c.832-15A>G, c.2017C>T, c.9748G>T, c.272T>A, c.5044G>T, c.5851C>T, c.6429G>A, c.5899C>T, c.1519delG, c.372delG, c.3222delA, c.9445C>T, c.354G>A, c.2665C>T, c.907C>T, c.5487_5488delAA, c.9568C>T, c.9471_9474delTTAT, c.8420G>A, c.1699G>T, c.2270C>G, c.2521C>T, c.3061G>T, c.4117C>T, c.5154+1G>A, c.5542A>T, c.2381-2A>G, c.4213C>T, c.1438G>T, c.3432+2240A>G, c.615T>A, c.5551C>T, c.649+2T>C, c.3347_3350delAGAA, c.5922+2T>C, c.5641C>T, c.2767delT, c.133C>T, c.8009G>A, c.6868A>T, c.9225-1G>A, c.4729C>T, c.3580C>T, c.10086+1G>T, c.199G>T, c.5554C>T, Exon 1-79 del, Exon 10-11 del, Exon 10-13 del, Exon 10-17 del, Exon 10-18 del, Exon 10-23 del, Exon 10-30 del, Exon 10-33 del, Exon 10-42 del, Exon 10-43 del, Exon 10-44 del, Exon 10-46 del, Exon 10-48 del, Exon 10-53 del, Exon 10-62 del, Exon 11-13 del, Exon 11-41 del, Exon 11-48 del, Exon 1-18 del, Exon 1-2 del, Exon 1-20 del, Exon 12-13 del, Exon 12-16 del, Exon 12-17 del, Exon 12-18 del, Exon 12-19 del, Exon 1-22 del, Exon 12-20 del, Exon 12-25 del, Exon 12-30 del, Exon 12-41 del, Exon 12-43 del, Exon 12-45 del, Exon 12-50 del, Exon 1-30 del, Exon 13-18 del, Exon 13-19 del, Exon 13-29 del, Exon 13-41 del, Exon 13-43 del, Exon 13-44 del, Exon 13-48 del, Exon 14-17 del, Exon 14-18 del, Exon 14-43 del, Exon 14-60 del, Exon 1-5 del, Exon 1-6 del, Exon 16-17 del, Exon 16-19 del, Exon 16-27 del, Exon 16-44 del, Exon 1-7 del, Exon 17-19 del, Exon 17-21 del, Exon 1-73 del, Exon 17-44 del, Exon 17-48 del, Exon 17-51 del, Exon 1-79 del, Exon 18-20 del, Exon 18-22 del, Exon 18-26 del, Exon 18-29 del, Exon 18-32 del, Exon 18-37 del, Exon 18-38 del, Exon 18-39 del, Exon 18-41 del, Exon 18-44 del, Exon 19-20 del, Exon 19-21 del, Exon 19-44 del, Exon 20-21 del, Exon 20-22 del, Exon 20-23 del, Exon 20-26 del, Exon 20-29 del, Exon 20-37 del, Exon 20-41 del, Exon 20-43 del, Exon 20-45 del, Exon 20-47 del, Exon 20-50 del, Exon 2-12 del, Exon 2-13 del, Exon 21-42 del, Exon 21-43 del, Exon 2-17 del, Exon 2-18 del, Exon 2-19 del, Exon 2-20 del, Exon 22-25 del, Exon 22-30 del, Exon 22-31 del, Exon 22-33 del, Exon 22-

37 del, Exon 22-41 del, Exon 22-47 del, Exon 2-29 del, Exon 2-30 del, Exon 24-26 del, Exon 24-27 del, Exon 2-44 del, Exon 2-6 del, Exon 26-43 del, Exon 26-44 del, Exon 2-7 del, Exon 28-49 del, Exon 30-42 del, Exon 3-11 del, Exon 3-12 del, Exon 3-13 del, Exon 31-40 del, Exon 31-43 del, Exon 3-15 del, Exon 31-57 del, Exon 3-16 del, Exon 3-17 del, Exon 3-19 del, Exon 3-20 del, Exon 3-21 del, Exon 3-24 del, Exon 3-25 del, Exon 3-26 del, Exon 3-27 del, Exon 3-28 del, Exon 3-29 del, Exon 3-30 del, Exon 3-34 del, Exon 33-43 del, Exon 33-45 del, Exon 3-37 del, Exon 3-4 del, Exon 3-41 del, Exon 3-42 del, Exon 3-43 del, Exon 3-44 del, Exon 35-42 del, Exon 35-43 del, Exon 35-44 del, Exon 35-45 del, Exon 3-6 del, Exon 3-7 del, Exon 37-43 del, Exon 3-8 del, Exon 38-43 del, Exon 39-43 del, Exon 4-12 del, Exon 4-13 del, Exon 4-17 del, Exon 4-18 del, Exon 4-19 del, Exon 4-22 del, Exon 42-43 del, Exon 42-45 del, Exon 42-50 del, Exon 42-53 del, Exon 4-30 del, Exon 43-50 del, Exon 43-51 del, Exon 43-52 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Dyskeratosis Congenita, Autosomal Recessive 1. Gene:*NOP10*. **Exons**: NM_018648.3:1-2. **Variants(1)**: c.100C>T

Dyskeratosis Congenita, Autosomal Recessive 2. Gene:*NHP2*. **Exons**: NM_017838.3:1-4. **Variants(2)**: c.460T>A, c.415T>C

Dyskeratosis Congenita, Autosomal Recessive 4. Gene:*TERT*. **Exons**: NM_198253.2:2-7,9-16. **Variants(3)**: c.2110C>T, c.2431C>T, c.2701C>T

Dyskeratosis Congenita, X-linked. Gene:*DKC1*. **Exons**: NM_001363.3:1-15. **Variants(7)**: c.106T>G, c.115A>G, c.91C>G, c.146C>T, c.113T>C, c.214_215delCTinsTA, c.196A>G

Dystonia 16. Gene:*PRKRA*. **Exons**: NM_003690.4:2-8. **Variants(2)**: c.267_268delTA, c.665C>T

Early Infantile Epileptic Encephalopathy 3. Gene:*SLC25A22*. **Exons**: NM_024698.5:2-10. **Variants(2)**: c.706G>T, c.617C>T

EEM Syndrome(Ectodermal Dysplasia, Ectrodactyly, and Macular Dystrophy). Gene:*CDH3*. **Exons**: NM_001793.4:1-16. **Variants(3)**: c.965A>T, c.1508G>A, c.830delG

Ehlers-Danlos Syndrome Type VI. Gene:*PLOD1*. **Exons**: NM_000302.3:1-19. **Variants(5)**: c.2032G>A, c.955C>T, c.1533C>G, c.1836G>C, c.2008C>T

Ehlers-Danlos Syndrome Type VIIC. Gene:*ADAMTS2*. **Exons**: NM_014244.4:2-22. **Variants(2)**: c.2384G>A, c.673C>T

Ehlers-Danlos syndrome, cardiac valvular form. Gene:*COL1A2*. **Exons**: NM_000089.3:1,3-52. **Variants(5)**: c.1404+1G>A, c.1404+1G>C, c.293_294insC, c.3601G>T, c.540+5G>A

Enhanced S-cone Syndrome. Gene:*NR2E3*. **Exons**: NM_014249.2:1-8. **Variants(4)**: c.119-2A>C, c.932G>A, c.226C>T, c.227G>A

Epidermolysis Bullosa Dystrophica. Gene:*COL7A1*. **Exons**: NM_000094.3:1-118. **Variants(10)**: c.7345-1G>A, c.6527_6528insC, c.4783G>C, c.6859G>A, c.5287C>T, c.706C>T, c.5443G>C, c.8440C>T, c.6266_6269delCCCC, c.8393T>A

Epidermolysis Bullosa Pruriginosa. Gene:*COL7A1*. **Exons**: NM_000094.3:1-118. **Variants(18)**: c.2471_2472insG, c.7411C>T, c.4119+1G>T, c.7787delG, c.8479C>T, c.6091G>A, c.4039G>C, c.425A>G, c.427-2A>G, c.3861delG, c.8245G>A, c.5532+1G>A, c.6187C>T, c.933C>A, c.5821-1G>A, c.4888C>T, c.6205C>T, c.5819delC

Epidermolysis Bullosa Simplex with Pyloric Atresia. Gene:*PLEC*. **Exons**: NM_000445.3:2-31,33. **Variants(4)**: c.6955C>T, c.913C>T, c.12043_12044insG, c.9085C>T

Ethylmalonic Encephalopathy. Gene:*ETHE1*. **Exons**: NM_014297.3:2-7. **Variants(5)**: c.604dupG, c.487C>T, c.221dupA, c.440_450delACAGCATGGCC, c.554T>G

Factor V Deficiency. Gene:*F5*. **Exons**: NM_000130.4:1-25. **Variants(5)**: c.1160T>C, c.5189A>G, c.2401C>T, c.6304C>T, c.439G>T

Familial Dysautonomia. Gene:*IKBKAP*. **Exons**: NM_003640.3:2-37. **Variants(3)**: c.2087G>C, c.2741C>T, c.2204+6T>C

Familial Exudative Vitreoretinopathy 4. Gene:*LRP5*. **Exons**: NM_002335.2:2-23. **Variants(5)**: c.804_813delGGGGAAGAGG, c.2254C>G, c.4099G>A, c.1709G>A, c.1828G>A

Familial Mediterranean Fever. Gene:*MEFV*. **Exons**: NM_000243.2:1-10. **Variants(11)**: c.2040G>A, c.1437C>G, c.2040G>C, c.1958G>A, c.2082G>A, c.2080A>G, c.2177T>C, c.800C>T, c.2282G>A, c.2230G>T, c.2076_2078delAAT

Fanconi anemia, complementation group A. Gene:*FANCA*. **Exons**: NM_000135.2:2-43. **Variants(5)**: c.2574C>G, c.1115_1118delTTGG, c.233_236delTTGA, c.4130C>G, c.3788_3790delTCT

Fanconi anemia, complementation group C. Gene:*FANCC*. **Exons**: NM_000136.2:2-15. **Variants(6)**: c.1487T>G, c.1642C>T, c.67delG, c.553C>T, c.37C>T, c.456+4A>T

Fanconi anemia, complementation group D1. Gene:*BRCA2*. **Exons**: NM_000059.3:2-27. **Variants(8)**: c.8488-1G>A, c.9900dupA, c.4648G>T, c.7691_7692insAT, c.658_659delGT, c.631+1G>A, c.5837_5838delCAinsAG, c.631+2T>G

Fanconi anemia, complementation group E. Gene:*FANCE*. **Exons**: NM_021922.2:2-10. **Variants(3)**: c.1114-8G>A, c.355C>T, c.421C>T

Fanconi anemia, complementation group G. Gene:*FANCG*. **Exons**: NM_004629.1:1-14. **Variants(8)**: c.1066C>T, c.307+1G>C, c.1795_1804delTGGATCCGTC, c.637_643delTACCGCC, c.1480+1G>C, c.1183_1192delGAGGTGTTTT, c.925-2A>G, c.313G>T

Fanconi anemia, complementation group I. Gene:*FANCI*. **Exons**: NM_001113378.1:2-38. **Variants(2)**: c.3853C>T, c.3854G>A

Fanconi anemia, complementation group J. Gene:*BRIP1*. **Exons**: NM_032043.2:2-20. **Variants(2)**: c.1045G>C, c.2392C>T

Fanconi anemia, complementation group L. Gene:*FANCL*. **Exons**: NM_018062.3:1-14. **Variants(2)**: c.1096_1099dupATTA, c.1007_1009delTAT

Fanconi anemia, complementation group M. Gene:*FANCM*. **Exons**: NM_020937.2:1-23. **Variants(1)**: c.2171C>A

Fanconi anemia, complementation group N. Gene:*PALB2*. **Exons**: NM_024675.3:1-13. **Variants(4)**: c.1653T>A, c.3116delA, c.2962C>T, c.3549C>G

Fanconi anemia, complementation group O. Gene:*RAD51C*. **Exons**: NM_058216.1:1-9. **Variants(1)**: c.773G>A

Fanconi anemia, complementation group P. Gene:*SLX4*. **Exons**: NM_032444.2:2-15. **Variants(5)**: c.286delA, c.1093delC, c.514delC, c.1163+3_1163+4insT, c.1163+2T>A

Fetal Akinesia Deformation Sequence. Gene:*RAPSN*. **Exons**: NM_005055.4:1-8. **Variants(1)**: c.566C>T

FGA-Related Congenital Afibrinogenemia. Gene:*FGA*. **Exons**: NM_021871.2:1-5. **Variants(3)**: c.1359dupC, c.1039C>T, c.510+1G>T

FGB-Related Congenital Afibrinogenemia. Gene:*FGB*. **Exons**: NM_005141.4:1-8. **Variants(3)**: c.1148T>G, c.1289G>A, c.794C>T

Fibrochondrogenesis. Gene:*COL11A1*. **Exons**: NM_001854.3:1-67. **Variants(2)**: c.2350G>C, c.1750dupG

Focal Segmental Glomerulosclerosis 3. Gene:*CD2AP*. **Exons**: NM_012120.2:1-18. **Variants(2)**: c.730-1delGinsCT, c.1834C>T

FRAS1-Related Fraser Syndrome. Gene:*FRAS1*. **Exons**: NM_025074.6:1-74. **Variants(7)**: c.8602C>T, c.6991_6992insGG, c.5605_5606insT, c.4271C>G, c.9013C>T, c.7682+1G>T, c.3799C>T

FREM2-Related Fraser Syndrome. Gene:*FREM2*. **Exons**: NM_207361.4:1-24. **Variants(2)**: c.5914G>A, c.7519+1G>A

Friedreich Ataxia. Gene:*FXN*. **Exons**: NM_000144.4:2-5. **Variants(5)**: c.317T>G, c.517T>G, c.460A>T, c.389G>T, c.385-2A>G

Frontometaphyseal Dysplasia. Gene:*FLNA*. **Exons**: NM_001456.3:2-47. **Variants(2)**: c.3557C>T, c.3476A>C

Fucosidosis. Gene:*FUCA1*. **Exons**: NM_000147.4:1-8. **Variants(6)**: c.1229T>G, c.244C>T, c.1160G>A, c.1138G>T, c.1279C>T, c.648C>A

Fuhrmann Dyndrome. Gene:*WNT7A*. **Exons**: NM_004625.3:1-4. **Variants(1)**: c.325G>A

Fumarate Hydratase Deficiency. Gene:*FH*. **Exons**: NM_000143.3:1-10. **Variants(2)**: c.1127A>C, c.521C>G

Galactosemia. Gene:*GALT*. **Exons**: NM_000155.3:1-11. **Variants(85)**: c.512T>C, c.563A>G, c.957C>G, c.1048A>G, c.1030C>T, c.337G>A, c.404C>T, c.221T>C, c.997C>G, c.379A>G, c.855G>T, c.938G>A, c.667C>A, c.1138T>C, c.595G>A, c.505C>A, c.610C>T, c.980T>C, c.692G>A, c.920C>A, c.425T>A, c.462G>A, c.677T>C, c.844C>G, c.428C>T, c.989C>T, c.1001A>G, c.160C>T, c.404C>G, c.1014C>G, c.290A>G, c.507+2T>C, c.580T>C, c.424A>G, c.619C>T, c.974C>T, c.238C>T, c.626A>G, c.199C>T, c.611G>C, c.377+1G>A, c.382G>A, c.1098C>A, c.691C>T, c.949delC, c.998G>A, c.948G>A, c.815G>A, c.899G>A, c.253-2A>G, c.776G>A, c.536G>A, c.1140A>C, c.524G>A, c.881T>A, c.452T>C, c.747G>A, c.602G>A, c.775C>T, c.855G>C, c.200G>A, c.82+2T>A, c.329-2A>C, c.18delC, c.564+1G>A, c.130G>A, c.616C>T, c.821-2A>G, c.634C>T, c.490delC, c.967T>C, c.413C>T, c.824delT, c.1075A>T, c.626A>C, c.687G>T, c.947G>A, c.341A>C, c.25C>T, c.443G>A, c.958G>A, c.607G>A, c.122G>A, c.1030C>A, c.584T>C

Gaucher Disease, Atypical. Gene:*PSAP*. **Exons**: NM_002778.2:1-14. **Variants(4)**: c.1288C>T, c.1145G>T, c.1144T>G, c.1046T>C

Gaucher Disease. Gene:*GBA*. **Exons**: NM_001005741.2:.. **Variants(4)**: c.1226A>G, c.115+1G>A, c.84dupG, c.1448T>C

Generalized arterial calcification of infancy 1. Gene:*ENPP1*. **Exons**: NM_006208.2:2-25. **Variants(6)**: c.1112A>T, c.913C>A, c.783C>G, c.1612G>C, c.2677G>T, c.1025G>T

Giant Axonal Neuropathy. Gene:*GAN*. **Exons**: NM_022041.3:2-11. **Variants(7)**: c.505G>A, c.1268T>C, c.1456G>A, c.413G>A, c.1447C>T, c.1429C>T, c.601C>T

GLDC-Related Glycine Encephalopathy. Gene:*GLDC*. **Exons**: NM_000170.2:2-25. **Variants(4)**: c.1705G>A, c.1691G>T, c.2216G>A, c.1166C>T

Glomerulosclerosis, Focal segmental,1. Gene:*ACTN4*. **Exons**: NM_004924.4:2-21. **Variants(3)**: c.776C>T, c.763A>G, c.784T>C

Glutaric Acidemia I. Gene:*GCDH*. **Exons**: NM_000159.2:2-12. **Variants(7)**: c.680G>C, c.1198G>A, c.1204C>T, c.1093G>A, c.1262C>T, c.883T>C, c.877G>A

Glutaric Acidemia IIA. Gene:*ETFA*. **Exons**: NM_000126.3:1-12. **Variants(3)**: c.470T>G, c.797C>T, c.346G>A

Glutaric Acidemia IIB. Gene:*ETFB*. **Exons**: NM_001985.2:1-6. **Variants(2)**: c.491G>A, c.382G>A

Glutaric Acidemia IIC. Gene:*ETFDH*. **Exons**: NM_004453.2:1-13. **Variants(7)**: c.1448C>T, c.250G>A, c.380T>A, c.524G>A, c.1130T>C, c.524G>T, c.2T>C

Glutathione Synthetase Deficiency. Gene:*GSS*. **Exons**: NM_000178.2:2-13. **Variants(6)**: c.4delG, c.847C>T, c.799C>T, c.656A>G, c.491G>A, c.656A>C

Glycogen Storage Disease Type Ia. Gene:*G6PC*. **Exons**: NM_000151.3:1-5. **Variants(12)**: c.1039C>T, c.247C>T, c.370G>A, c.229T>C, c.551G>A, c.248G>A, c.883C>T, c.328G>A, c.380_381insTA, c.562G>C, c.497T>G, c.113A>T

Glycogen Storage Disease Type Ib/Ic. Gene:*SLC37A4*. **Exons**: NM_001164278.1:3-11. **Variants(9)**: c.1108_1109delCT, c.1129G>T, c.83G>A, c.352T>C, c.1309C>T, c.1082G>A, c.706_708delGTG, c.287G>A, c.1081G>T

Glycogen Storage Disease Type II. Gene:*GAA*. **Exons**: NM_000152.3:2-20. **Variants(132)**: c.1843G>A, c.1636+5G>C, c.1856G>A, c.1798C>T, c.271delG, c.1115A>T, c.1796C>A, c.482_483delCC, c.896T>G, c.1333G>C, c.1826dupA, c.1703A>T, c.784G>A, c.1552-3C>G, c.1935C>A, c.546G>T, c.923A>C, c.1222A>G, c.1064T>C, c.2741delAinsCAG, c.2269C>T, c.1548G>A, c.1210G>A, c.692+1G>C, c.2173C>T, c.1364A>T, c.670C>T, c.2281delGinsAT, c.1755-1G>A, c.1725C>A, c.935T>G, c.340_341insT, c.643G>T, c.2012T>G, c.2380delC, c.1585_1586delTCinsGT, c.1076-1G>C, c.794delG, c.710C>T, c.1687C>T, c.2846T>A, c.1978C>T, c.2331+4A>G, c.989G>A, c.1564C>G, c.623T>C, c.1082C>T, c.1933G>A, c.1802C>G, c.971C>T, c.1432G>A, c.1905C>A, c.1735G>A, c.307T>G, c.377G>A, c.1941C>G, c.1551+1G>C, c.2185delC, c.685_686insCGGC, c.1316T>A, c.1556T>C, c.875A>G, c.2174G>C, c.1377_1379delCGA, c.2014C>T, c.2040G>A, c.2608C>T, c.1561G>A, c.2646+2T>A, c.573C>A, c.2605delG, c.2560C>T, c.1979G>A, c.1724A>C, c.716delT, c.2140delC, c.925G>A, c.2815_2816delGT, c.1120T>C, c.172C>T, c.1696T>C, c.2331+2T>C, c.1076-22T>G, c.953T>C, c.872T>C, c.2041-2A>C, c.118C>T, c.2303C>G, c.2702T>A, c.1214T>C, c.1634C>T, c.1100G>A, c.877G>A, c.1799G>A, c.1327-2A>G, c.1655T>C, c.2432delT, c.1836C>G, c.1194+2T>A, c.1326+1G>A, c.2219_2220delTG, c.1927G>A, c.2132C>G, c.1375G>A, c.854C>G, c.988T>G, c.2015G>A, c.1827delC, c.1080C>G, c.1128_1129delGGinsC, c.1942G>A, c.1438-1G>C, c.2104C>T, c.2188G>T, c.18_25delGCCCTGCT, c.2662G>T, c.379_380delTG, c.1411_1414delGAGA, c.2024_2026delACA, c.2639C>A, c.525delT, c.722_723delTT, c.1857C>G, c.1555A>G, c.1441T>C, c.1309C>T, c.309C>A, c.1456G>C, c.399C>A, c.1124G>T, c.1437+2T>C, c.-32-3C>A

Glycogen Storage Disease Type III. Gene:*AGL*. **Exons**: NM_000642.2:2-34. **Variants(13)**: c.2039G>A, c.3682C>T, c.3980G>A, c.16C>T, c.4529dupA, c.4260-12A>G, c.1999delC, c.18_19delGA, c.2590C>T, c.1222C>T, c.3965delT, c.4456delT, c.4342G>C

Glycogen Storage Disease Type IV. Gene:*GBE1*. **Exons**: NM_000158.3:1-16. **Variants(10)**: c.771T>A, c.1571G>A, c.986A>G, c.1883A>G, c.986A>C, c.1643G>A, c.1543C>T, c.1570C>T, c.671T>C, c.1774G>T

Glycogen Storage Disease Type IXc. Gene:*PHKG2*. **Exons**: NM_000294.2:2-10. **Variants(1)**: c.130C>T

Glycogen Storage Disease Type XI. Gene:*LDHA*. **Exons**: NM_005566.3:2-8. **Variants(2)**: c.126+1G>A, c.640_641delCT

Glycogen Storage Disease Type XII. Gene:*ALDOA*. **Exons**: NM_000034.3:7-14. **Variants(2)**: c.619G>A, c.386A>G

Glycogen Storage Disease Type XIII. Gene:*ENO3*. **Exons**: NM_053013.3:2-12. **Variants(2)**: c.467G>A, c.1121G>A

Glycogen Storage Disease Type XIV. Gene:*PGM1*. **Exons**: NM_002633.2:1-11. **Variants(2)**: c.1145-1G>C, c.343A>G

GM1-Gangliosidosis. Gene:*GLB1*. **Exons**: NM_000404.2:1-16. **Variants(14)**: c.622C>T, c.1772A>G, c.245C>T, c.145C>T, c.1445G>A, c.1369C>T, c.176G>A, c.601C>T, c.947A>G, c.152T>C, c.1370G>A, c.1771T>A, c.1051C>T, c.202C>T

GM2-Gangliosidosis, AB variant. Gene:*GM2A*. **Exons**: NM_000405.4:1-4. **Variants(4)**: c.410delA, c.160G>T, c.412T>C, c.262_264delAAG

GNE-Related Myopathy. Gene:*GNE*. **Exons**: NM_005476.5:2-12. **Variants(6)**: c.1727G>A, c.737G>A, c.1714G>T, c.2086G>A, c.2135T>C, c.909T>A

GRACILE Syndrome. Gene:*BCS1L*. **Exons**: NM_004328.4:3-9. **Variants(2)**: c.232A>G, c.166C>T

Greenberg dysplasia. Gene:*LBR*. **Exons**: NM_002296.3:2-14. **Variants(3)**: c.1748G>A, c.1402delT, c.32_35delTGGT

Griscelli Syndrome Type 1. Gene:*MYO5A*. **Exons**: NM_000259.3:2-41. **Variants(1)**: c.2332C>T

Griscelli Syndrome Type 2. Gene:*RAB27A*. **Exons**: NM_004580.4:2-6. **Variants(5)**: c.217T>G, c.352C>T, c.259G>C, c.389T>C, c.454G>C

Guanidinoaceteate Methyltransferase Deficiency. Gene:*GAMT*. **Exons**: NM_000156.5:2-6. **Variants(1)**: c.506G>A

Gyrate Atrophy Of the Choroid And Retina. Gene:*OAT*. **Exons**: NM_000274.3:2-10. **Variants(25)**: c.1172G>A, c.952delG, c.824G>A, c.596C>A, c.159delC, c.627T>A, c.897C>G, c.1201G>T, c.278G>T, c.3G>A, c.1276C>T, c.268C>G, c.994G>A, c.952G>A, c.1180T>C, c.677C>T, c.955C>T, c.1205T>C, c.539G>C, c.1250C>T, c.1186C>T, c.901-2A>G, c.192_193delAG, c.533G>A, c.812G>A

HADHA related Trifunctional Protein Deficiency. Gene:*HADHA*. **Exons**: NM_000182.4:1-20. **Variants(1)**: c.1793_1794delAT

HADHB related Trifunctional Protein Deficiency. Gene:*HADHB*. **Exons**: NM_000183.2:2-16. **Variants(2)**: c.1331G>A, c.1364T>G

HARP Syndrome. Gene:*PANK2*. **Exons**: NM_153638.2:1-7. **Variants(2)**: c.1441C>T, c.1413-1G>T

Hemophilia A. Gene:*F8*. **Exons**: NM_000132.3:1-26. **Variants(508)**: c.1498delA, c.680G>A, c.4770T>A, c.541G>A, c.1009+1G>A, c.755_756delCA, c.6350T>G, c.5562G>A, c.1311delG, c.5219+1G>A, c.1520C>G, c.524A>G, c.5894G>T, c.5561G>A, c.3385delC, c.5904C>A, c.670+5G>A, c.242C>A, c.6967C>G, c.2048A>G, c.3619_3622delCACA, c.5981T>C, c.1538-2A>G, c.1420G>A, c.6743G>C, c.6115+4A>G, c.6464_6465delAA, c.2939delG, c.5665C>T, c.5452G>T, c.195C>A, c.1357G>T, c.1718G>A, c.1831C>T, c.6084delG, c.6325C>T, c.1325A>G, c.1026T>A, c.935T>C, c.1266_1270delTGACA, c.729delT, c.4483delG, c.6226G>T, c.1750delC, c.6193T>C, c.4339delG, c.5389C>T, c.6393G>A, c.1230_1232delGGA, c.6724-1G>A, c.658G>C, c.2384_2388delGAACA, c.144-2A>G, c.901C>T,

c.5663G>T, c.1752+1G>A, c.1034T>C, c.1293G>T, c.2770G>T, c.6016G>T, c.3659delA, c.6046C>T, c.575T>C, c.1538-1G>T, c.980T>C, c.525C>A, c.2702C>A, c.6496C>G, c.6842T>C, c.6413C>A, c.4906C>T, c.2485delG, c.266G>A, c.2912T>G, c.292G>A, c.409A>G, c.6116-2A>G, c.1382_1389delTTCAGCAT, c.388+5G>A, c.1753-1G>A, c.665A>T, c.5568T>G, c.6721C>T, c.1475A>G, c.6797delG, c.6250A>T, c.5183A>G, c.5681A>G, c.388G>C, c.3736delC, c.854T>C, c.5374-2A>T, c.2633T>G, c.1752+5G>A, c.566C>T, c.5373+1G>A, c.6505C>T, c.1487delC, c.6881_6882delTT, c.2099C>A, c.1394C>G, c.4383_4384delCC, c.6412_6413delTC, c.5140delA, c.3991_3992delAA, c.5573_5574delCT, c.5998+1G>A, c.5564_5567delCTTA, c.977T>C, c.4899delT, c.6836T>G, c.1636C>T, c.514_515insTATCTTGA, c.4757G>A, c.202A>C, c.209_212delTTGT, c.1203G>A, c.3692delC, c.6748C>T, c.206T>C, c.6296_6297delTT, c.2032A>T, c.5536A>T, c.1202G>A, c.5869C>T, c.5575delG, c.1763A>G, c.1241A>G, c.1657T>C, c.6485C>T, c.350T>G, c.1337G>A, c.526T>C, c.556G>T, c.5113C>T, c.98G>A, c.1214T>G, c.3551delA, c.6273+1G>A, c.824A>G, c.144-1G>A, c.3143G>A, c.1813T>C, c.881C>T, c.1909A>G, c.1463_1464insTTGATTC, c.1702G>A, c.296T>A, c.764G>C, c.6665G>A, c.1406G>C, c.5934T>G, c.2649delG, c.5336G>A, c.1630G>A, c.5770delA, c.787+3A>G, c.6172G>C, c.5415T>A, c.2159G>A, c.1786T>C, c.683A>G, c.1522delA, c.6403C>T, c.6694C>T, c.1315G>A, c.5855_5856dupTA, c.519_523delTACCT, c.6865C>T, c.6989A>C, c.542T>A, c.6723+1G>A, c.1904-2A>G, c.1172G>A, c.6404G>C, c.3325C>T, c.6994T>C, c.5125delA, c.1640G>A, c.1244C>A, c.6208G>A, c.4926delA, c.2T>G, c.3409_3410delCT, c.1063C>T, c.4712_4715delAAAG, c.695_698delAGAA, c.2132G>A, c.5825G>C, c.6115+1G>A, c.516C>G, c.4072C>T, c.5766C>A, c.1809C>G, c.4103delC, c.2203_2204insTTCT, c.5557G>A, c.1407_1411delACCTT, c.6590T>A, c.7035C>A, c.1916_1917delAT, c.1595G>A, c.902G>A, c.1569G>T, c.2443C>T, c.6120T>A, c.4201C>T, c.5184T>A, c.3904C>T, c.328A>G, c.6972C>G, c.1043G>A, c.7016G>C, c.3860delT, c.4564A>T, c.2615C>G, c.2167G>A, c.1947_1950delTTTG, c.5879G>A, c.902G>T, c.5953C>T, c.1414_1418delCTTTA, c.7012delC, c.3344delT, c.3607G>T, c.5985C>A, c.3590delA, c.6321delT, c.4430_4431delAG, c.7021G>A, c.796G>T, c.3371C>A, c.4798A>T, c.1861C>T, c.822G>A, c.398A>G, c.6740_6741delAG, c.2214C>G, c.991_992delAT, c.6995G>A, c.3421C>T, c.761A>T, c.6360T>G, c.5220-1G>A, c.601+5G>A, c.606T>G, c.849delT, c.6494delC, c.2121G>A, c.3766G>T, c.923C>T, c.4265_4266delAT, c.1474T>C, c.3703delA, c.6547A>G, c.6520C>G, c.6593G>T, c.1348T>A, c.1754T>C, c.5219G>T, c.832G>A, c.3833delA, c.6870G>A, c.5677C>T, c.545A>C, c.5392G>C, c.3602delT, c.5177G>A, c.6443A>G, c.4720delG, c.5914_5915delAT, c.6794A>G, c.6713G>A, c.2354_2355delTA, c.5526G>A, c.6794_6795delAG, c.6744G>T, c.4519delA, c.1003C>T, c.4460delA, c.337C>T, c.1529T>A, c.671-2A>G, c.592T>G, c.5460delA, c.3851_3852delCA, c.640T>G, c.636delC, c.5697delC, c.605G>A, c.421G>A, c.2638G>T, c.1751A>G, c.5243delA, c.4662_4663delGA, c.4918G>T, c.6738delA, c.1812G>C, c.4425_4426delAA, c.4781_4787delTACCAAA, c.6671C>T, c.6115+5G>A, c.4561C>T, c.5220-2A>G, c.248C>G, c.602-1G>A, c.2962_2963delAG, c.5301C>A, c.1990_1991delCA, c.6356A>G, c.1200_1201delTT, c.4035delA, c.6574+1G>A, c.1226A>G, c.5618C>G, c.6200C>T, c.1804C>G, c.3772delT, c.4423C>T, c.1430G>T, c.2842delC, c.958_960delATG, c.7034G>A, c.5372T>C, c.4805_4806delAA, c.3060_3063delCAAA, c.4296_4300delTTCTC, c.857A>C, c.4121_4124delTAGA, c.5213_5216delGAAA, c.3292C>T, c.1408C>A, c.6082G>A, c.2149C>T, c.2893A>T, c.953T>C, c.4858delC, c.2440C>T, c.1078_1079delGA, c.4280delT, c.4328_4331delAAGA, c.1033G>C, c.5726A>G, c.741G>A, c.5878C>T, c.4848delC, c.6610delA, c.270delG, c.560T>A, c.590T>G, c.6563G>A, c.3203_3204delGA, c.5882G>A, c.5433T>A, c.2235_2242delTTCAGCAT, c.4999delC, c.1976T>C, c.1798G>T, c.403G>T, c.1726G>T, c.6968G>A, c.1364T>G, c.265+1G>A, c.1442_1443insT, c.3490delT, c.7054delT, c.1271+1G>A, c.4934G>A, c.3493G>T, c.5883G>A, c.3416_3417delCT, c.4272delC, c.1957G>A, c.6501delC, c.6506G>A, c.974_975delTT, c.5071_5075delATGAA, c.1434C>A, c.3827C>G, c.1293delG, c.2936G>A, c.558C>A, c.2246T>A, c.1616G>A, c.6429+5G>T, c.2320C>T, c.410C>T, c.2610delT, c.71A>G, c.4658delA, c.5719A>T, c.1917T>A, c.755C>T, c.2409delT, c.1285C>T, c.2369delC, c.5399G>A, c.4935G>A, c.797G>A, c.2244C>A, c.205_206delCT, c.940A>G, c.2114-1G>A, c.2120G>A, c.5752delT, c.173delC, c.2110C>T, c.5508G>A, c.1485C>G, c.1621A>T, c.6429+2T>A, c.3844A>T, c.1696C>T, c.4492_4493delGT, c.266-1G>A, c.4318delT, c.6394C>T, c.6683G>A, c.3232_3239delTTGAGGCT, c.1941_1944delAGTT, c.5260T>C, c.6575-1G>A, c.6977G>A, c.415C>T, c.1750C>A, c.2958delA, c.6997delG, c.118delC, c.1444-2A>G, c.1781T>A, c.1965C>G, c.57T>G, c.3548_3549delAA, c.1804C>T, c.6595delA, c.1443+1G>A, c.5606G>A, c.5587-1G>A, c.1454A>C, c.6876_6877delCT, c.6385A>T, c.515G>T, c.6967C>T, c.364_365delGT, c.6760delC, c.1760C>A, c.2089_2090delGT, c.5219+3A>G, c.968G>A, c.6515C>A, c.1803C>G, c.1933C>T, c.360delT, c.5443C>T, c.6497delG, c.1492G>A, c.5398C>G, c.6116-1G>A, c.5374-1G>A, c.5822A>G, c.6374G>C, c.530A>G, c.6970T>A, c.4796G>A, c.6554T>C, c.3994_3997delAGAG, c.6053A>G, c.406C>T, c.2526_2527delAG, c.1899G>T, c.4687delG, c.3289C>T, c.491G>A, c.4987A>T, c.5428T>C, c.1281_1282delAA, c.2496T>G, c.5888T>C, c.5815+2T>C, c.3967C>T, c.2937G>A, c.3091_3094delAAGA, c.5038G>T, c.6682C>G, c.2373G>A, c.3702_3705delTACA, c.224delA, c.1478delA, c.43C>T, c.6194G>A, c.6253G>T, c.6915T>G, c.872A>G, c.595A>T, c.688_689delGA, c.5335G>A, c.3381G>A, c.5881T>C, c.6545G>A, c.5553C>A, c.2360delA, c.4662G>A, c.6623A>G, c.472C>T, c.938T>G, c.6266G>A, Intron 22 inversion, Intron 1 inversion

Hemophilia B. Gene:*F9*. **Exons**: NM_000133.3:1-8. **Variants(301)**: c.392-2A>G, c.60delA, c.1295G>A, c.1278delT, c.985delA, c.1342delT, c.1024delA, c.420A>T, c.272A>G, c.464G>A, c.520+2T>G, c.162G>C, c.545_546delCT, c.505T>C, c.281G>T, c.268C>T, c.774T>G, c.1256T>A, c.1067G>A, c.422G>A, c.407T>C, c.907delC, c.400T>C, c.174delG, c.80_83delAATG, c.1322A>G, c.229delG, c.782G>A, c.1173T>A, c.1097C>A, c.698C>A, c.280G>A, c.501G>T, c.735delT, c.572G>A, c.1136G>A, c.50T>A, c.260T>G, c.941A>G, c.416G>A, c.219A>C, c.914A>G, c.263G>A, c.284delA, c.179_180delTT, c.155T>C, c.237A>C, c.392-1G>T, c.534_535delTG, c.487dupC, c.679G>T, c.724-2A>G, c.990C>A, c.1031T>A, c.1187G>A, c.758G>A, c.344A>G, c.681_682insT, c.128G>A, c.317G>A, c.205T>C, c.781T>C, c.1070G>A, c.757G>A, c.1220G>A, c.138G>C, c.350G>A, c.862G>T, c.349T>A, c.412A>C, c.1361T>C, c.688G>A, c.881G>A, c.936C>A, c.127C>T, c.1064G>A, c.754T>C, c.603T>G, c.1142C>A, c.88G>A, c.1105C>G, c.1275A>C, c.214G>C, c.482A>G, c.236A>C, c.1068G>A, c.173G>A, c.1018G>T, c.305G>A, c.1235G>A, c.1301_1302delAG, c.278-3A>G, c.277G>A, c.687_689delTGG, c.1023C>A, c.278-12C>T, c.1304G>A, c.1025C>A, c.1146T>A, c.277+5G>C, c.427C>G, c.909delC, c.423C>A, c.478G>A, c.786T>G, c.547_548delGT, c.109G>A, c.1293G>A, c.967_974delGAACCCTT, c.682G>C, c.413A>G, c.775G>T, c.1144T>C, c.520+13A>G, c.83G>A, c.1088G>A, c.247_248insA, c.96delT, c.172G>A, c.1323T>G, c.722delA, c.917A>G, c.1213G>A, c.1343_1350delCCCGGTAT, c.838+2T>G, c.55delC, c.862delG, c.1188T>A, c.796G>A, c.1151G>C, c.892C>T, c.804T>G, c.454A>T, c.655C>T, c.1084A>G, c.1346G>A, c.1276A>C, c.471T>A, c.493G>T, c.1349A>G, c.291T>G, c.1325G>A, c.1274T>G, c.1109A>C, c.838+1G>C, c.533G>A, c.799delC, c.1357T>A, c.947T>C, c.157G>A, c.253-2A>G, c.676C>G, c.536G>A, c.827_828delCA, c.1219T>A, c.165_169delTGTTC, c.1181T>A, c.1185C>G, c.155delT, c.622G>T, c.212T>C, c.185_188delGAGA, c.874delC, c.223C>T, c.946A>T, c.1318A>G, c.689_691delGAG, c.905A>G, c.856_858delGAG, c.322delT, c.532T>C, c.723+2T>C, c.1328T>C, c.707G>A, c.199G>A, c.277+4A>G, c.800A>G, c.1217C>G, c.755G>A, c.1147C>A, c.799C>T, c.484C>A, c.373G>A, c.1241C>A, c.389delT, c.556delA, c.401G>A, c.324C>A, c.316G>A, c.89-6T>G, c.1231A>G, c.509G>A, c.370G>A, c.88+5G>A, c.145T>C, c.1151_1154delGATC, c.1150C>T, c.922delG, c.1244A>G, c.835G>A, c.813delT, c.1145G>A, c.1132G>T, c.1240C>A, c.83delG, c.723G>C, c.1058T>C, c.224G>A, c.1306G>A, c.479G>A, c.127delC, c.1294G>A, c.278A>G, c.1135C>G, c.1174A>G, c.252+5G>A, c.415G>A, c.1175delA, c.413delA, c.1129G>T, c.352T>A, c.226G>A, c.279T>A, c.1113C>A, c.59T>A, c.1183T>C, c.418dupA, c.568_569delAC, c.133_134insT, c.720G>A, c.808G>T, c.466T>C, c.942T>G, c.197A>T, c.219_220insA, c.253-3T>G, c.723+1G>T, c.1169T>C, c.252+1G>A, c.1069G>A, c.292G>T, c.957_958delGGinsC, c.1139C>A, c.1270T>C, c.677G>A, c.724-1G>A, c.1077_1078delCT, c.161_162delAG, c.932delA, c.761G>A, c.1348T>C, c.659delC, c.1324G>A, c.391+1G>C, c.719G>A, c.520+1G>T, c.1066T>A, c.1258G>T, c.783G>T, c.1297G>A, c.235G>A,

c.287A>C, c.88+5_88+8delGTTT, c.449delA, c.414T>A, c.356G>A, c.470G>A, c.206G>A, c.82T>C, c.871G>A, c.328G>A, c.944A>G, c.1358G>A, c.148G>A, c.1189G>C, c.1168A>T, c.191G>A, c.278-1G>A, c.163T>A, c.392delA, c.668delA, c.278-2A>T, c.184A>T, c.286C>T, c.1052G>A, c.785T>C, c.659C>A, c.353G>A, c.1138G>A, c.190T>C, c.1074_1075delAG, c.1182G>A, c.434G>A, c.956T>C, c.453_454delCA, c.277+2T>C, c.508T>C, c.30delA, c.1183T>A, c.255_256delTG, c.391+2T>C
Hereditary Fructose Intolerance. Gene:*ALDOB*. **Exons**: NM_000035.3:2-9. **Variants(7)**: c.10C>T, c.1005C>G, c.720C>A, c.178C>T, c.524C>A, c.360_363delCAAA, c.448G>C
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum. Gene:*SLC12A6*. **Exons**: NM_133647.1:1-25. **Variants(5)**: c.619C>T, c.2436+1delG, c.3031C>T, c.1584_1585delCTinsG, c.2023C>T
Hereditary Sensory and Autonomic Neuropathy IV. Gene:*NTRK1*. **Exons**: NM_001012331.1:2-16. **Variants(9)**: c.1076A>G, c.1711G>C, c.1741A>G, c.2066C>T, c.851-33T>A, c.1908_1909insT, c.1709delT, c.1834G>T, c.2321G>C
Hermansky-Pudlak Syndrome 1. Gene:*HPS1*. **Exons**: NM_000195.3:3-20. **Variants(6)**: c.1472_1487dupCCAGCAGGGGAGGCCC, c.397G>T, c.972delC, c.398+5G>A, c.972_973insC, c.1996G>T
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency. Gene:*CBS*. **Exons**: NM_000071.2:3-17. **Variants(12)**: c.1330G>A, c.341C>T, c.430G>A, c.434C>T, c.502G>A, c.572C>T, c.1058C>T, c.797G>A, c.919G>A, c.1397C>T, c.1150A>G, c.1265C>T
Hydrolethalus Syndrome 1. Gene:*HYLS1*. **Exons**: NM_145014.2:4. **Variants(1)**: c.632A>G
Hydrolethalus Syndrome 2. Gene:*KIF7*. **Exons**: NM_198525.2:2-4,6-19. **Variants(1)**: c.2896_2897delGC
Hyper-IgD syndrome. Gene:*MVK*. **Exons**: NM_000431.2:2-11. **Variants(5)**: c.803T>C, c.829C>T, c.1129G>A, c.59A>C, c.494C>T
Hypermethioninemia. Gene:*MAT1A*. **Exons**: NM_000429.2:1-9. **Variants(9)**: c.1006G>A, c.164C>A, c.966T>G, c.790C>T, c.914T>C, c.1043_1044delTG, c.538_539insTG, c.1070C>T, c.827_828insG
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome. Gene:*SLC25A15*. **Exons**: NM_014252.3:2-7. **Variants(11)**: c.658G>A, c.562_564delTTC, c.824G>A, c.79G>A, c.815C>T, c.212T>A, c.535C>T, c.110T>G, c.95C>G, c.538G>A, c.569G>A
Hyperprolinemia Type II. Gene:*ALDH4A1*. **Exons**: NM_003748.3:2-15. **Variants(1)**: c.1055C>T
Hypomagnesemia 5. Gene:*CLDN19*. **Exons**: NM_148960.2:1-5. **Variants(3)**: c.169C>G, c.59G>A, c.269T>C
Hypomyelination and Congenital Cataract. Gene:*FAM126A*. **Exons**: NM_032581.3:2-11. **Variants(1)**: c.158T>C
Hypoparathyroidism-Retardation-Dysmorphism Syndrome. Gene:*TBCE*. **Exons**: NM_003193.3:2-17. **Variants(2)**: c.1113T>A, c.66_67delAG
Hypophosphatasia. Gene:*ALPL*. **Exons**: NM_000478.4:2-12. **Variants(20)**: c.979T>C, c.746G>T, c.211C>T, c.881A>C, c.535G>A, c.1366G>A, c.620A>C, c.571G>A, c.1559delT, c.1133A>T, c.98C>T, c.814C>T, c.346G>A, c.892G>A, c.1306T>C, c.1250A>G, c.1001G>A, c.526G>A, c.407G>A, c.212G>C
Hypotrichosis, congenital, with juvenile macular dystrophy. Gene:*CDH3*. **Exons**: NM_001793.4:1-16. **Variants(1)**: c.981delG
Ichthyosis Follicularis-Atrichia-Photophobia Syndrome. Gene:*MBTPS2*. **Exons**: NM_015884.3:1-11. **Variants(4)**: c.1286G>A, c.677G>T, c.1424T>C, c.261G>A
IGF1 Deficiency. Gene:*IGF1*. **Exons**: NM_000618.3:1-4. **Variants(1)**: c.274G>A
Infantile Sialic acid Storage Disorder. Gene:*SLC17A5*. **Exons**: NM_012434.4:1-11. **Variants(3)**: c.1001C>G, c.548A>G, c.918T>G
Infantile Striatonigral Degeneration. Gene:*NUP62*. **Exons**: NM_153719.3:3. **Variants(1)**: c.1172A>C
Infantile-Onset Spinocerebellar Ataxia. Gene:*C10orf2*. **Exons**: NM_021830.4:1-5. **Variants(5)**: c.955A>G, c.1370C>T, c.1287C>T, c.952G>A, c.1523A>G
Isolated Growth Hormone Deficiency. Gene:*BTK*. **Exons**: NM_000061.2:2-19. **Variants(2)**: c.1625T>C, c.1125T>G
Isolated Microphthalmia 3. Gene:*RAX*. **Exons**: NM_013435.2:1-2. **Variants(3)**: c.575G>A, c.439C>T, c.909C>G
Isolated Microphthalmia 5. Gene:*MFRP*. **Exons**: NM_031433.2:1-13. **Variants(1)**: c.499_500insC
Isovaleric Acidemia. Gene:*IVD*. **Exons**: NM_002225.3:1-12. **Variants(5)**: c.941C>T, c.1188delT, c.157C>T, c.605G>T, c.134T>C
ITGA6-Related Epidermolysis Bullosa with Pyloric Atresia. Gene:*ITGA6*. **Exons**: NM_000210.2:1-25. **Variants(1)**: c.791delC
ITGB4-Related Epidermolysis Bullosa with Pyloric Atresia. Gene:*ITGB4*. **Exons**: NM_001005731.1:2-39. **Variants(14)**: c.112T>C, c.1684T>C, c.3841C>T, c.4410delG, c.4433G>A, c.467T>C, c.3977-19T>A, c.3674G>A, c.3801_3802insT, c.1150delG, c.3793+1G>A, c.1660C>T, c.182G>A, c.2792G>A
Johanson-Blizzard Syndrome. Gene:*UBR1*. **Exons**: NM_174916.2:1-47. **Variants(2)**: c.407A>G, c.1537C>T
Joubert Syndrome 1. Gene:*INPP5E*. **Exons**: NM_019892.4:1-10. **Variants(4)**: c.1132C>T, c.1543C>T, c.1688G>A, c.1304G>A
Joubert Syndrome 10. Gene:*OFD1*. **Exons**: NM_003611.2:1-23. **Variants(2)**: c.2767delG, c.2844_2850delAGACAAA
Joubert syndrome 13. Gene:*TCTN1*. **Exons**: NM_001082538.2:1-14. **Variants(1)**: c.221-2A>G
Joubert Syndrome 2. Gene:*TMEM216*. **Exons**: NM_001173990.2:1-5. **Variants(2)**: c.218G>A, c.218G>T
Joubert Syndrome 3. Gene:*AHI1*. **Exons**: NM_017651.4:3-28. **Variants(10)**: c.1328T>A, c.1303C>T, c.985C>T, c.2168G>A, c.1052G>T, c.1765C>T, c.1484G>A, c.3263_3264delGG, c.1051C>T, c.2370dupT
Joubert Syndrome 5. Gene:*CEP290*. **Exons**: NM_025114.3:2-54. **Variants(5)**: c.21G>T, c.5668G>T, c.2249T>G, c.4723A>T, c.4656delA
Joubert Syndrome 6. Gene:*TMEM67*. **Exons**: NM_153704.5:1-28. **Variants(3)**: c.755T>C, c.130C>T, c.1538A>G
Joubert Syndrome 7. Gene:*RPGRIP1L*. **Exons**: NM_015272.2:2-22,24-27. **Variants(7)**: c.1975T>C, c.697A>T, c.1843A>C, c.2269delA, c.2413C>T, c.757C>T, c.2050C>T
Joubert Syndrome 8. Gene:*ARL13B*. **Exons**: NM_182896.2:1-10. **Variants(3)**: c.236G>A, c.598C>T, c.246G>A
Joubert Syndrome 9. Gene:*CC2D2A*. **Exons**: NM_001080522.2:3-38. **Variants(5)**: c.4582C>T, c.3289delG, c.4667A>T, c.3364C>T, c.2848C>T

Kanzaki Disease. Gene:*NAGA*. **Exons**: NM_000262.2:1-9. **Variants(3)**: c.985C>T, c.577G>T, c.986G>A

KCNJ11-Related Congenital Hyperinsulinism. Gene:*KCNJ11*. **Exons**: NM_000525.3:1. **Variants(4)**: c.761C>T, c.440T>C, c.776A>G, c.36C>A

Kelley-Seegmiller Syndrome. Gene:*HPRT1*. **Exons**: NM_000194.2:2-9. **Variants(4)**: c.193C>T, c.602A>G, c.239A>T, c.329C>T

Knobloch syndrome. Gene:*COL18A1*. **Exons**: NM_030582.3:1-41. **Variants(2)**: c.3517_3518delCC, c.3618_3619delGG

Krabbe Disease. Gene:*GALC*. **Exons**: NM_000153.3:2-17. **Variants(5)**: c.1153G>T, c.1586C>T, c.1630G>A, c.857G>A, c.1796T>G

L1 Syndrome. Gene:*L1CAM*. **Exons**: NM_000425.3:1-28. **Variants(14)**: c.536T>G, c.924C>T, c.1792G>A, c.1354G>A, c.719C>T, c.3489_3490delTG, c.2432-19A>C, c.2254G>A, c.551G>A, c.791G>A, c.3581C>T, c.1108G>A, c.1939+5G>A, c.3458-1G>C

LAMA2-Related Congenital Muscular Dystrophy. Gene:*LAMA2*. **Exons**: NM_000426.3:1-65. **Variants(10)**: c.2098_2099delTT, c.8314delA, c.2901C>A, c.825delC, c.3718C>T, c.7732C>T, c.2049_2050delAG, c.7147C>T, c.9253C>T, c.4645C>T

LAMA3-Related Junctional Epidermolysis Bullosa. Gene:*LAMA3*. **Exons**: NM_000227.3:1-38. **Variants(4)**: c.2116A>T, c.335delG, c.4135C>T, c.1981C>T

LAMB3-Related Junctional Epidermolysis Bullosa. Gene:*LAMB3*. **Exons**: NM_000228.2:2-23. **Variants(10)**: c.1587_1588delAG, c.496C>T, c.727C>T, c.2806C>T, c.904delT, c.1903C>T, c.1830G>A, c.1438_1442delCCGTG, c.628G>A, c.124C>T

LAMC2-Related Junctional Epidermolysis Bullosa. Gene:*LAMC2*. **Exons**: NM_005562.2:1-23. **Variants(8)**: c.1065C>G, c.1659C>A, c.3512_3513insA, c.2137_2143delCAGAACC, c.405-1G>A, c.1067-1G>A, c.283C>T, c.733C>T

Lathosterolosis. Gene:*SC5DL*. **Exons**: NM_006918.4:2-5. **Variants(1)**: c.86G>A

Leber Congenital Amaurosis 1. Gene:*GUCY2D*. **Exons**: NM_000180.3:3-19. **Variants(3)**: c.1694T>C, c.2945delG, c.622delC

Leber congenital amaurosis 10. Gene:*CEP290*. **Exons**: NM_025114.3:2-54. **Variants(1)**: c.3185delT

Leber Congenital Amaurosis 13. Gene:*RDH12*. **Exons**: NM_152443.2:3-9. **Variants(13)**: c.379G>T, c.377C>T, c.295C>A, c.451C>G, c.152T>A, c.523T>C, c.184C>T, c.146C>T, c.806_810delCCCTG, c.464C>T, c.565C>T, c.677A>G, c.451C>A

Leber Congenital Amaurosis 14. Gene:*LRAT*. **Exons**: NM_004744.3:2-3. **Variants(2)**: c.525T>A, c.217_218delAT

Leber Congenital Amaurosis 15. Gene:*TULP1*. **Exons**: NM_003322.3:1-15. **Variants(2)**: c.1204G>T, c.1198C>T

Leber Congenital Amaurosis 16. Gene:*KCNJ13*. **Exons**: NM_002242.4:2-3. **Variants(2)**: c.496C>T, c.722T>C

Leber Congenital Amaurosis 2. Gene:*RPE65*. **Exons**: NM_000329.2:1-14. **Variants(4)**: c.700C>T, c.907A>T, c.1067delA, c.1292A>G

Leber Congenital Amaurosis 4. Gene:*AIPL1*. **Exons**: NM_014336.3:1-6. **Variants(5)**: c.244C>T, c.715T>C, c.905G>T, c.589G>C, c.834G>A

Leber Congenital Amaurosis 7. Gene:*CRX*. **Exons**: NM_000554.4:2-4. **Variants(2)**: c.268C>T, c.529delG

Leber Congenital Amaurosis 8. Gene:*CRB1*. **Exons**: NM_201253.2:1-12. **Variants(4)**: c.2688T>A, c.2843G>A, c.3299T>G, c.3997G>T

Leber Congenital Amaurosis 9. Gene:*NMNAT1*. **Exons**: NM_022787.3:2-5. **Variants(9)**: c.769G>A, c.457C>G, c.817A>G, c.25G>A, c.619C>T, c.710G>T, c.451G>T, c.507G>A, c.838T>C

Leigh Syndrome, French-Canadian Type. Gene:*LRPPRC*. **Exons**: NM_133259.3:2-38. **Variants(2)**: c.1061C>T, c.3830_3839delGTGGTGCAATinsAG

Lesch-Nyhan Syndrome. Gene:*HPRT1*. **Exons**: NM_000194.2:2-9. **Variants(4)**: c.508C>T, c.170T>C, c.143G>A, c.212_213insG

Lethal Acantholytic Epidermolysis Bullosa. Gene:*DSP*. **Exons**: NM_004415.2:1-24. **Variants(2)**: c.6370_6371delCT, c.5800C>T

Lethal Congenital Contracture Syndrome 1. Gene:*GLE1*. **Exons**: NM_001003722.1:1-16. **Variants(2)**: c.1849G>A, c.2051T>C

LIG4 Syndrome. Gene:*LIG4*. **Exons**: NM_002312.3:2. **Variants(4)**: c.1738C>T, c.1406G>A, c.2440C>T, c.833G>A

Limb-Girdle Muscular Dystrophy Type 2A. Gene:*CAPN3*. **Exons**: NM_000070.2:1-24. **Variants(9)**: c.956C>T, c.1469G>A, c.1795_1796insA, c.257C>T, c.2362_2363delAGinsTCATCT, c.328C>T, c.1715G>A, c.550delA, c.2306G>A

Limb-Girdle Muscular Dystrophy type 2B. Gene:*DYSF*. **Exons**: NM_003494.3:1-35,37-55. **Variants(5)**: c.2372C>G, c.5201A>G, c.895G>A, c.200_201delTGinsAT, c.1873G>T

Limb-Girdle Muscular Dystrophy type 2C. Gene:*SGCG*. **Exons**: NM_000231.2:2-8. **Variants(2)**: c.787G>A, c.848G>A

Limb-Girdle Muscular Dystrophy type 2D. Gene:*SGCA*. **Exons**: NM_000023.2:1-9. **Variants(4)**: c.293G>A, c.574C>T, c.739G>A, c.229C>T

Limb-Girdle Muscular Dystrophy type 2E. Gene:*SGCB*. **Exons**: NM_000232.4:2-6. **Variants(7)**: c.272G>T, c.452C>G, c.299T>A, c.323T>G, c.552T>G, c.341C>T, c.272G>C

Limb-Girdle Muscular Dystrophy type 2G. Gene:*TCAP*. **Exons**: NM_003673.3:1-2. **Variants(1)**: c.157C>T

Limb-Girdle Muscular Dystrophy type 2H. Gene:*TRIM32*. **Exons**: NM_012210.3:2. **Variants(3)**: c.1560delC, c.1181G>A, c.1459G>A

Limb-Girdle Muscular Dystrophy Type 2L. Gene:*ANO5*. **Exons**: NM_213599.2:1-22. **Variants(6)**: c.2311_2312delCA, c.1407+5G>A, c.191_192insA, c.1295C>G, c.2272C>T, c.692G>T

Lipoid Congenital Adrenal Hyperplasia. Gene:*STAR*. **Exons**: NM_000349.2:1-7. **Variants(8)**: c.562C>T, c.650G>C, c.545G>A, c.772C>T, c.559G>A, c.577C>T, c.749G>A, c.545G>T

Lissencephaly with Cerebellar Hypoplasia. Gene:*RELN*. **Exons**: NM_005045.3:1-65. **Variants(1)**: c.5615-1G>A

Long-Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency. Gene:*HADHA*. **Exons**: NM_000182.4:1-20. **Variants(4)**: c.2132_2133insC, c.1528G>C, c.1132C>T, c.1678C>T

Lujan-Fryns Syndrome. Gene:*MED12*. **Exons**: NM_005120.2:1-41,43-45. **Variants(1)**: c.3020A>G

Macular Corneal Dystrophy. Gene:*CHST6*. **Exons**: NM_021615.4:3. **Variants(2)**: c.609C>A, c.599T>G

Malonyl-CoA Decarboxylase Deficiency. Gene:*MLYCD*. **Exons**: NM_012213.2:2-5. **Variants(1)**: c.560C>G

Mandibuloacral Dysplasia with type A lipodystrophy. Gene:*ZMPSTE24*. **Exons**: NM_005857.4:1-10. **Variants(5)**: c.1018T>C, c.743C>T, c.1349G>A, c.121C>T, c.1085_1086insT

Mandibuloacral Dysplasia with type B lipodystrophy. Gene:*LMNA*. **Exons**: NM_170707.3:1-12. **Variants(6)**: c.1586C>T, c.1626G>C, c.1411C>T, c.1318G>A, c.1580G>A, c.1579C>T

Maple Syrup Urine Disease Type 1A. Gene:*BCKDHA*. **Exons**: NM_000709.3:1-9. **Variants(3)**: c.1312T>A, c.117delC, c.868G>A

Maple Syrup Urine Disease Type 1B. Gene:*BCKDHB*. **Exons**: NM_183050.2:1-10. **Variants(6)**: c.356T>G, c.1039-7_1039-4delTCTG, c.832G>A, c.616C>T, c.1114G>T, c.548G>C

Maple Syrup Urine Disease Type 2. Gene:*DBT*. **Exons**: NM_001918.3:1-11. **Variants(5)**: c.1355A>G, c.1448G>T, c.294C>G, c.581C>G, c.827T>G

Maple Syrup Urine Disease Type 3. Gene:*DLD*. **Exons**: NM_000108.3:1-14. **Variants(3)**: c.105_106insA, c.685G>T, c.1483A>G

Marinesco-Sj?gren syndrome. Gene:*SIL1*. **Exons**: NM_022464.4:2-10. **Variants(3)**: c.1370T>C, c.1312C>T, c.331C>T

Martsolf Syndrome. Gene:*RAB3GAP2*. **Exons**: NM_012414.3:1-35. **Variants(1)**: c.3154G>T

McKusick-Kaufman Syndrome. Gene:*MKKS*. **Exons**: NM_018848.3:3-6. **Variants(3)**: c.724G>T, c.110A>G, c.250C>T

Meckel Syndrome 1. Gene:*MKS1*. **Exons**: NM_017777.3:1-18. **Variants(2)**: c.51_55dupCCGGG, c.80+2T>C

Meckel Syndrome 10. Gene:*B9D2*. **Exons**: NM_030578.3:2-4. **Variants(1)**: c.301A>C

Meckel Syndrome 2. Gene:*TMEM216*. **Exons**: NM_001173990.2:1-5. **Variants(3)**: c.341T>G, c.253C>T, c.230G>C

Meckel Syndrome 3. Gene:*TMEM67*. **Exons**: NM_153704.5:1-28. **Variants(2)**: c.1127A>C, c.622A>T

Meckel Syndrome 4. Gene:*CEP290*. **Exons**: NM_025114.3:2-54. **Variants(2)**: c.384_387delTAGA, c.613C>T

Meckel Syndrome 5. Gene:*RPGRIP1L*. **Exons**: NM_015272.2:2-22,24-27. **Variants(3)**: c.2614C>T, c.1033C>T, c.394A>T

MECP2-Related Severe Neonatal Encephalopathy. Gene:*MECP2*. **Exons**: NM_004992.3:2-4. **Variants(6)**: c.410A>G, c.964C>T, c.419C>T, c.1282G>A, c.1180G>T, c.674C>T

Megalencephalic Leukoencephalopathy with Subcortical Cysts 1. Gene:*MLC1*. **Exons**: NM_015166.3:2-12. **Variants(8)**: c.278C>T, c.422A>G, c.274C>T, c.839C>T, c.423C>A, c.206C>T, c.135_136insC, c.178-10T>A

Metachromatic Leukodystrophy due to Arylsulfatase A. Gene:*ARSA*. **Exons**: NM_000487.5:1-8. **Variants(12)**: c.1283C>T, c.769G>C, c.465+1G>A, c.257G>A, c.1401_1411delGTTAGACGCAG, c.739G>A, c.1232C>T, c.1210+1G>A, c.293C>T, c.641C>T, c.542T>G, c.302G>A

Metachromatic Leukodystrophy due to Saposin B Deficiency. Gene:*PSAP*. **Exons**: NM_002778.2:1-14. **Variants(2)**: c.722G>C, c.643A>C

Methylmalonic Aciduria and Homocystinuria CblC type. Gene:*MMACHC*. **Exons**: NM_015506.2:1-4. **Variants(6)**: c.331C>T, c.394C>T, c.440G>C, c.482G>A, c.347T>C, c.271dupA

Methylmalonic Aciduria and Homocystinuria CblD type. Gene:*MMADHC*. **Exons**: NM_015702.2:2-8. **Variants(7)**: c.746A>G, c.748C>T, c.545C>A, c.160C>T, c.776T>C, c.419dupA, c.57_64delCTCTTTAG

Mevalonic Aciduria. Gene:*MVK*. **Exons**: NM_000431.2:2-11. **Variants(3)**: c.1000G>A, c.902A>C, c.928G>A

Microphthalmia with coloboma 8/Syndromic Microphthalmia 9. Gene:*STRA6*. **Exons**: NM_022369.3:2-19. **Variants(12)**: c.527_528insG, c.1678G>C, c.910_911delGGinsAA, c.1931C>T, c.878C>T, c.1521-1G>A, c.52_53delTAinsC, c.1964G>A, c.1963C>T, c.147delC, c.69G>A, c.277_278insCC

Minicore Myopathy With External Ophthalmoplegia. Gene:*RYR1*. **Exons**: NM_000540.2:1-90,92-106. **Variants(6)**: c.1739_1742dupATCA, c.10343C>T, c.7268T>A, c.325C>T, c.14365-2A>T, c.5726_5727delAG

Mitochondrial Complex III Deficiency Nuclear Type 1. Gene:*BCS1L*. **Exons**: NM_004328.4:3-9. **Variants(7)**: c.830G>A, c.296C>T, c.1057G>A, c.547C>T, c.464G>C, c.148A>G, c.133C>T

Mitochondrial Complex III Deficiency nuclear Type 3. Gene:*UQCRB*. **Exons**: NM_006294.4:1-4. **Variants(1)**: c.306_309delAAAA

Mitochondrial Complex III Deficiency nuclear Type 4. Gene:*UQCRQ*. **Exons**: NM_014402.4:2-3. **Variants(1)**: c.134C>T

Mitochondrial DNA depletion Syndrome 2. Gene:*TK2*. **Exons**: NM_004614.4:2-10. **Variants(5)**: c.323C>T, c.361C>A, c.159C>G, c.268C>T, c.635T>A

Mitochondrial DNA depletion Syndrome 3. Gene:*DGUOK*. **Exons**: NM_080916.2:1-7. **Variants(4)**: c.679G>A, c.425G>A, c.763G>T, c.313C>T

Mitochondrial DNA depletion Syndrome 6. Gene:*MPV17*. **Exons**: NM_002437.4:2-8. **Variants(5)**: c.498C>A, c.149G>A, c.359G>A, c.148C>T, c.70G>T

Mitochondrial DNA depletion Syndrome 9. Gene:*SUCLG1*. **Exons**: NM_003849.3:1-8. **Variants(3)**: c.626C>A, c.97+3G>C, c.152_153delAT

Miyoshi Myopathy. Gene:*DYSF*. **Exons**: NM_003494.3:1-35,37-55. **Variants(7)**: c.895G>T, c.6124C>T, c.1555G>A, c.1813C>T, c.5713C>T, c.2997G>T, c.3137G>A

MMAA-Related Methylmalonic Acidemia. Gene:*MMAA*. **Exons**: NM_172250.2:2-7. **Variants(4)**: c.503delC, c.433C>T, c.283C>T, c.620A>G

MMAB-Related Methylmalonic Acidemia. Gene:*MMAB*. **Exons**: NM_052845.3:1-9. **Variants(3)**: c.548A>T, c.556C>T, c.569G>A

Mohr-Tranebjaerg Syndrome. Gene:*TIMM8A*. **Exons**: NM_004085.3:1-2. **Variants(3)**: c.112C>T, c.238C>T, c.198C>G

Molybdenum Cofactor Deficiency A. Gene:*MOCS1*. **Exons**: NM_005943.5:1-9. **Variants(2)**: c.956G>A, c.217C>T

Molybdenum Cofactor Deficiency B. Gene:*MOCS2*. **Exons**: NM_176806.2:1-3. **Variants(3)**: c.*422G>A, c.16C>T, c.*487A>C

MORM Syndrome. Gene:*INPP5E*. **Exons**: NM_019892.4:1-10. **Variants(1)**: c.1879C>T

MTHFR deficiency. Gene:*MTHFR*. **Exons**: NM_005957.4:2-12. **Variants(6)**: c.1743G>A, c.1015T>G, c.968T>C, c.547C>T, c.1129C>T, c.971A>G

Mucolipidosis II. Gene:*GNPTAB*. **Exons**: NM_024312.4:1-21. **Variants(6)**: c.3173C>G, c.2681G>A, c.3503_3504delTC, c.1196C>T, c.310C>T, c.3565C>T

Mucolipidosis IV. Gene:*MCOLN1*. **Exons**: NM_020533.2:2-14. **Variants(5)**: c.1084G>T, c.1207C>T, c.964C>T, c.304C>T, c.406-2A>G

Mucopolysaccharidosis Type I. Gene:*IDUA*. **Exons**: NM_000203.3:2-8,12-14. **Variants(8)**: c.1960T>G, c.1037T>G, c.1855C>G, c.266G>A, c.1206G>A, c.1861C>T, c.208C>T, c.192C>A

Mucopolysaccharidosis Type II. Gene:*IDS*. **Exons**: NM_000202.5:1-2,4-9. **Variants(9)**: c.1425G>A, c.1464G>T, c.1327C>T, c.514C>T, c.1466G>C, c.1402C>T, c.1403G>T, c.1403G>A, c.998C>T

Mucopolysaccharidosis Type IIIA. Gene:*SGSH*. **Exons**: NM_000199.3:2-8. **Variants(10)**: c.1105G>A, c.892T>C, c.197C>G, c.1298G>A, c.383C>T, c.449G>A, c.1339G>A, c.220C>T, c.734G>A, c.617G>C

Mucopolysaccharidosis Type IIIC. Gene:*HGSNAT*. **Exons**: NM_152419.2:2-18. **Variants(8)**: c.1030C>T, c.493+1G>A, c.962T>G, c.1345dupG, c.525dupT, c.1553C>T, c.848C>T, c.372-2A>G

Mucopolysaccharidosis Type IIID. Gene:*GNS*. **Exons**: NM_002076.3:1-14. **Variants(4)**: c.1169delA, c.1063C>T, c.1226dupG, c.1168C>T

Mucopolysaccharidosis Type IVB. Gene:*GLB1*. **Exons**: NM_000404.2:1-16. **Variants(6)**: c.1527G>T, c.818G>T, c.1313G>A, c.1223A>C, c.1444C>T, c.1498A>G

Mucopolysaccharidosis Type VI. Gene:*ARSB*. **Exons**: NM_000046.3:2-8. **Variants(6)**: c.629A>G, c.349T>C, c.410G>T, c.707T>C, c.1178A>C, c.1214G>A

Mucopolysaccharidosis Type VII. Gene:*GUSB*. **Exons**: NM_000181.3:1-12. **Variants(15)**: c.442C>T, c.646C>T, c.526C>T, c.1061C>T, c.1144C>T, c.1831C>T, c.1244+1G>A, c.1338G>A, c.1069C>T, c.1484A>G, c.1050G>C, c.1881G>T, c.1856C>T, c.1730G>T, c.1521G>A

Mulibrey Nanism. Gene:*TRIM37*. **Exons**: NM_015294.3:1-24. **Variants(14)**: c.860G>A, c.2056C>T, c.227T>C, c.2212delG, c.1411C>T, c.1894_1895delGA, c.965G>T, c.855_860+2delTTTCAGGT, c.1346_1347insA, c.838_842delACTTT, c.326G>C, c.1037_1040dupAGAT, c.745C>T, c.496_500delAGGAA

Muscular Dystrophy-Dystroglycanopathy (congenital with brain and eye anomalies) Type A1. Gene:*POMT1*. **Exons**: NM_007171.3:2-20. **Variants(2)**: c.226G>A, c.907C>T

Muscular Dystrophy-Dystroglycanopathy (congenital with brain and eye anomalies) Type A2. Gene:*POMT2*. **Exons**: NM_013382.5:1-21. **Variants(5)**: c.1117G>T, c.2177G>A, c.1238G>C, c.1912C>T, c.1057G>A

Muscular Dystrophy-Dystroglycanopathy (congenital with brain and eye anomalies) Type A3. Gene:*POMGNT1*. **Exons**: NM_017739.3:2-22. **Variants(7)**: c.932G>A, c.1425G>A, c.1539+1G>A, c.1539+1G>T, c.187C>T, c.652+1G>A, c.1864delC

Muscular Dystrophy-Dystroglycanopathy (congenital with brain and eye anomalies) Type A4. Gene:*FKTN*. **Exons**: NM_001079802.1:3-11. **Variants(2)**: c.509C>A, c.1112A>G

Muscular Dystrophy-Dystroglycanopathy (congenital with brain and eye anomalies) type A6. Gene:*LARGE*. **Exons**: NM_004737.4:3-16. **Variants(2)**: c.1483T>C, c.992C>T

Muscular Dystrophy-Dystroglycanopathy (Congenital with Mental retardation) Type B1. Gene:*POMT1*. **Exons**: NM_007171.3:2-20. **Variants(6)**: c.2163C>A, c.2005G>A, c.1540C>T, c.1746G>C, c.193G>A, c.1770G>C

Muscular Dystrophy-Dystroglycanopathy (congenital with mental retardation) Type B2. Gene:*POMT2*. **Exons**: NM_013382.5:1-21. **Variants(4)**: c.1941G>A, c.1997A>G, c.2242T>C, c.1445G>T

Muscular Dystrophy-Dystroglycanopathy (congenital with mental retardation) Type B3. Gene:*POMGNT1*. **Exons**: NM_017739.3:2-22. **Variants(2)**: c.1469G>A, c.1814G>C

Muscular Dystrophy-Dystroglycanopathy (congenital with mental retardation) type B6. Gene:*LARGE*. **Exons**: NM_004737.4:3-16. **Variants(1)**: c.1525G>A

Muscular Dystrophy-Dystroglycanopathy (limb-girdle) Type C1. Gene:*POMT1*. **Exons**: NM_007171.3:2-20. **Variants(1)**: c.598G>C

Muscular Dystrophy-Dystroglycanopathy (limb-girdle) Type C2. Gene:*POMT2*. **Exons**: NM_013382.5:1-21. **Variants(2)**: c.551C>T, c.2243G>C

Muscular Dystrophy-Dystroglycanopathy (limb-girdle) Type C4. Gene:*FKTN*. **Exons**: NM_001079802.1:3-11. **Variants(3)**: c.340G>A, c.527T>C, c.920G>A

MUT-Related Methylmalonic Acidemia. Gene:*MUT*. **Exons**: NM_000255.3:2-13. **Variants(10)**: c.2150G>T, c.2080C>T, c.313T>C, c.655A>T, c.1105C>T, c.278G>A, c.1867G>A, c.322C>T, c.643G>A, c.682C>T

Myopathy with Deficiency of ISCU. Gene:*ISCU*. **Exons**: NM_213595.2:2-5. **Variants(1)**: c.149G>A

Myotonia Congenita. Gene:*CLCN1*. **Exons**: NM_000083.2:1-23. **Variants(5)**: c.501C>G, c.1238T>G, c.1453A>G, c.871G>A, c.2680C>T

N-acetylglutamate synthase deficiency. Gene:*NAGS*. **Exons**: NM_153006.2:2-7. **Variants(6)**: c.971G>A, c.916-2A>T, c.1025delG, c.1299G>C, c.1307dupT, c.1289T>C

Nanophthalmos 2. Gene:*MFRP*. **Exons**: NM_031433.2:1-13. **Variants(4)**: c.1150_1151insC, c.545T>C, c.523C>T, c.498delC

Nemaline Myopathy 2. Gene:*NEB*. **Exons**: NM_004543.4:3-56,58-60,62-65,68-150. **Variants(3)**: c.18318_18319delAG, c.19606G>T, c.19119_19120delGA

Nemaline Myopathy 5. Gene:*TNNT1*. **Exons**: NM_003283.4:2-3,5-14. **Variants(1)**: c.538G>T

Nephronophthisis 1. Gene:*NPHP1*. **Exons**: NM_000272.3:1-20. **Variants(2)**: c.1884+1G>T, c.80T>A

Nephronophthisis 11. Gene:*TMEM67*. **Exons**: NM_153704.5:1-28. **Variants(4)**: c.2461G>A, c.1843T>C, c.869G>T, c.2461G>C

Nephronophthisis 2. Gene:*INVS*. **Exons**: NM_014425.3:2-17. **Variants(5)**: c.1453delC, c.2695C>T, c.2719C>T, c.1478T>C, c.1807C>T

Nephropathic Cystinosis. Gene:*CTNS*. **Exons**: NM_004937.2:3-12. **Variants(10)**: c.969C>G, c.397_398delAT, c.357_360delCAGC, c.414G>A, c.853-3C>G, c.329G>T, c.1015G>A, c.416C>T, c.473T>C, c.283G>T

Nephrotic Syndrome Type 1. Gene:*NPHS1*. **Exons**: NM_004646.3:1-29. **Variants(10)**: c.1481delC, c.2456A>T, c.121_122delCT, c.1307_1308dupAC, c.3250_3251insG, c.2464G>A, c.3478C>T, c.3325C>T, c.3250delG, c.793T>C

Nephrotic Syndrome Type 2. Gene:*NPHS2*. **Exons**: NM_014625.2:1-8. **Variants(1)**: c.413G>A

Nephrotic Syndrome Type 3. Gene:*PLCE1*. **Exons**: NM_016341.3:2-32. **Variants(8)**: c.4846C>T, c.3736C>T, c.961C>T, c.3846delG, c.3346C>T, c.1477C>T, c.4451C>T, c.5560C>T

Neurodegeneration With Brain Iron Accumulation 2. Gene:*PLA2G6*. **Exons**: NM_003560.2:2-17. **Variants(6)**: c.2370T>G, c.1894C>T, c.1634A>C, c.109C>T, c.929T>A, c.238G>A

Neuronal Ceroid-Lipofuscinoses 1. Gene:*PPT1*. **Exons**: NM_000310.3:1-9. **Variants(7)**: c.29T>A, c.223A>C, c.169_170insA, c.322G>C, c.236A>G, c.451C>T, c.364A>T

Neuronal Ceroid-Lipofuscinoses 10. Gene:*CTSD*. **Exons**: NM_001909.4:2-9. **Variants(2)**: c.1149G>C, c.685T>A

Neuronal Ceroid-Lipofuscinoses 2. Gene:*TPP1*. **Exons**: NM_000391.3:1-13. **Variants(8)**: c.1340G>A, c.616C>T, c.857A>G, c.509-1G>C, c.1094G>A, c.622C>T, c.1093T>C, c.851G>T

Neuronal Ceroid-Lipofuscinoses 3. Gene:*CLN3*. **Exons**: NM_001042432.1:2-16. **Variants(2)**: c.597C>A, c.883G>A

Neuronal Ceroid-Lipofuscinoses 4A. Gene:*CLN6*. **Exons**: NM_017882.2:2-7. **Variants(3)**: c.308G>A, c.139C>T, c.200T>C

Neuronal Ceroid-Lipofuscinoses 5. Gene:*CLN5*. **Exons**: NM_006493.2:1-4. **Variants(2)**: c.1175_1176delAT, c.1054G>T

Neuronal Ceroid-Lipofuscinoses 6. Gene:*CLN6*. **Exons**: NM_017882.2:2-7. **Variants(3)**: c.316_317insC, c.214G>T, c.663C>G

Neuronal Ceroid-Lipofuscinoses 7. Gene:*MFSD8*. **Exons**: NM_152778.2:2-13. **Variants(6)**: c.1235C>T, c.1286G>A, c.362A>G, c.894T>G, c.881C>A, c.929G>A

Neuronal Ceroid-Lipofuscinoses 8. Gene:*CLN8*. **Exons**: NM_018941.3:2-3. **Variants(5)**: c.611G>T, c.88G>C, c.789G>C, c.88delG, c.70C>G

Niemann-Pick Disease Type A. Gene:*SMPD1*. **Exons**: NM_000543.4:1-6. **Variants(5)**: c.1493G>T, c.996delC, c.1152G>A, c.911T>C, c.788T>A

Niemann-Pick Disease Type B. Gene:*SMPD1*. **Exons**: NM_000543.4:1-6. **Variants(3)**: c.1327C>T, c.1829_1831delGCC, c.1267C>T

Niemann-Pick Disease Type C1. Gene:*NPC1*. **Exons**: NM_000271.4:2-25. **Variants(16)**: c.3467A>G, c.3662delT, c.3263A>G, c.337T>C, c.2974G>A, c.2848G>A, c.3611_3614delTTAC, c.3107C>T, c.2873G>A, c.3104C>T, c.3175C>T, c.2932C>T, c.530G>A, c.2974G>T, c.3591+1G>A, c.3182T>C

Niemann-Pick Disease Type C2. Gene:*NPC2*. **Exons**: NM_006432.3:1-5. **Variants(11)**: c.441+1G>A, c.358C>T, c.27delG, c.111delG, c.199T>C, c.436C>T, c.82+2T>C, c.58G>T, c.352G>T, c.190+5G>A, c.115G>A

Night blindness, congenital stationary (complete) 1B, Autosomal Recessive. Gene:*GRM6*. **Exons**: NM_000843.3:2-10. **Variants(7)**: c.2341G>A, c.719_720insG, c.727_728insG, c.2122C>T, c.1214T>C, c.1565G>A, c.1861C>T

Night blindness, congenital stationary (complete) 1E, Autosomal Recessive. Gene:*GPR179*. **Exons**: NM_001004334.2:1-11. **Variants(6)**: c.278delC, c.187delC, c.598C>T, c.1784+1G>A, c.984delC, c.1807C>T

Night blindness, congenital stationary (complete)1A. Gene:*NYX*. **Exons**: NM_022567.2:1. **Variants(1)**: c.1049G>A

Night blindness, congenital stationary (complete)1D. Gene:*SLC24A1*. **Exons**: NM_004727.2:2-10. **Variants(1)**: c.1613_1614delTT

Nijmegen breakage syndrome. Gene:*NBN*. **Exons**: NM_002485.4:1-16. **Variants(1)**: c.657_661delACAAA

Oculocutaneous Albinism Type 1. Gene:*TYR*. **Exons**: NM_000372.4:1-4. **Variants(30)**: c.823G>T, c.1118C>A, c.265T>C, c.1012_1013insC, c.272G>A, c.1501dupC, c.896G>A, c.1A>G, c.230G>A, c.1164delT, c.572delG, c.707G>A, c.1209G>T, c.1265G>A, c.1336G>A, c.164G>A, c.140G>A, c.649C>T, c.1217C>T, c.1255G>A, c.1147G>A, c.616G>A, c.1146C>A, c.650G>A, c.1467dupT, c.242C>T, c.1342G>A, c.533G>A, c.732_733delTG, c.286dupA

Oculocutaneous Albinism Type 2. Gene:*OCA2*. **Exons**: NM_000275.2:2-24. **Variants(8)**: c.79G>A, c.2037G>C, c.1465A>G, c.1960delG, c.1842+1G>T, c.1327G>A, c.2228C>T, c.1182G>A

Oculocutaneous Albinism Type 3. Gene:*TYRP1*. **Exons**: NM_000550.2:2-8. **Variants(6)**: c.1120C>T, c.1103delA, c.107delT, c.497C>G, c.1057_1060delAACA, c.1067G>A

Oculocutaneous Albinism Type 4. Gene:*SLC45A2*. **Exons**: NM_016180.3:1-7. **Variants(3)**: c.986delC, c.469G>A, c.1121delT

Odontoonychodermal Dysplasia. Gene:*WNT10A*. **Exons**: NM_025216.2:1-3. **Variants(3)**: c.321C>A, c.697G>T, c.383G>A

Oguchi Disease-1. Gene:*SAG*. **Exons**: NM_000541.4:2-16. **Variants(5)**: c.926delA, c.577C>T, c.523C>T, c.874C>T, c.916G>T

Ohdo Syndrome. Gene:*MED12*. **Exons**: NM_005120.2:1-41,43-45. **Variants(3)**: c.5185C>A, c.3443G>A, c.3493T>C

Ornithine Transcarbamylase Deficiency. Gene:*OTC*. **Exons**: NM_000531.5:1-10. **Variants(14)**: c.386G>A, c.77G>A, c.829C>T, c.617T>G, c.332T>C, c.119G>A, c.646C>G, c.717+2T>C, c.259G>A, c.118C>T, c.148G>T, c.674C>T, c.460G>T, c.134T>C

Osteogenesis Imperfecta Type VII. Gene:*CRTAP*. **Exons**: NM_006371.4:2-7. **Variants(2)**: c.561T>G, c.826C>T

Osteogenesis Imperfecta Type VIII. Gene:*LEPRE1*. **Exons**: NM_022356.3:1-15. **Variants(6)**: c.747delC, c.1080+1G>T, c.1473+1G>T, c.1102C>T, c.1365_1366delAGinsC, c.1656C>A

Osteoporosis-Pseudoglioma Syndrome. Gene:*LRP5*. **Exons**: NM_002335.2:2-23. **Variants(9)**: c.1282C>T, c.1481G>A, c.1468delG, c.2202G>A, c.1453G>T, c.2305delG, c.1708C>T, c.1584+1G>A, c.2557C>T

Pancreatic Agenesis. Gene:*PDX1*. **Exons**: NM_000209.3:1. **Variants(3)**: c.492G>T, c.533A>G, c.532G>A

Pantothenate Kinase-Associated Neurodegeneration. Gene:*PANK2*. **Exons**: NM_153638.2:1-7. **Variants(5)**: c.790C>T, c.570C>G, c.1583C>T, c.1561G>A, c.533C>A

PCCA-Related Propionic Acidemia. Gene:*PCCA*. **Exons**: NM_000282.3:1-24. **Variants(3)**: c.1891G>C, c.412G>A, c.862A>T

PCCB-Related Propionic Acidemia. Gene:*PCCB*. **Exons**: NM_000532.4:1-15. **Variants(9)**: c.1283C>T, c.1173_1174insT, c.1228C>T, c.737G>T, c.1495C>T, c.502G>A, c.1534C>T, c.1304A>G, c.1540_1541insCCC

Pelizaeus-Merzbacher Disease. Gene:*PLP1*. **Exons**: NM_000533.3:1-7. **Variants(3)**: c.169G>T, c.725C>T, c.3G>A

Peroxisomal acyl-CoA oxidase deficiency. Gene:*ACOX1*. **Exons**: NM_004035.6:1-14. **Variants(4)**: c.442C>T, c.832A>G, c.926A>G, c.532G>T

Peroxisome biogenesis disorder 1. Gene:*PEX1*. **Exons**: NM_000466.2:1-24. **Variants(4)**: c.2097_2098insT, c.1991T>C, c.2916delA, c.2528G>A

Peroxisome biogenesis disorder 2. Gene:*PEX5*. **Exons**: NM_001131025.1:2-16. **Variants(2)**: c.1279C>T, c.1578T>G

Peroxisome biogenesis disorder 3. Gene:*PEX12*. **Exons**: NM_000286.2:1-3. **Variants(3)**: c.691A>T, c.538C>T, c.959C>T

Peroxisome biogenesis disorder 5. Gene:*PEX2*. **Exons**: NM_000318.2:4. **Variants(2)**: c.163G>A, c.355C>T

Peroxisome biogenesis disorder 6. Gene:*PEX10*. **Exons**: NM_153818.1:2-6. **Variants(2)**: c.704_705insA, c.373C>T

Peroxisome biogenesis disorder 7. Gene:*PEX26*. **Exons**: NM_017929.5:2-6. **Variants(5)**: c.254dupT, c.265G>A, c.34_35insC, c.292C>T, c.2T>C

Phenylketonuria. Gene:*PAH*. **Exons**: NM_000277.1:1-13. **Variants(203)**: c.165T>G, c.208_210delTCT, c.464G>A, c.932T>C, c.727C>T, c.805A>C, c.1021A>T, c.618C>A, c.441+1G>A, c.168+1G>A, c.896T>G, c.353-1G>C, c.204A>T, c.1A>G, c.1056delT, c.694C>T, c.728G>A, c.754C>T, c.122T>C, c.581T>C, c.1157A>G, c.782G>A, c.662A>G, c.943G>T, c.678G>C, c.763T>G, c.977G>A, c.901C>T, c.559T>C, c.1089delG, c.721C>T, c.913-7A>G, c.775G>A, c.611A>G, c.997C>T, c.1200-8G>A, c.1084C>A, c.814G>T, c.1112A>G, c.1045T>C, c.975C>G, c.1232C>G, c.168+5G>A, c.558_559delAT, c.912+2T>C, c.818C>T, c.284_286delTCA, c.941C>A, c.1129delT, c.1055delG, c.608G>A, c.745C>T, c.442G>A, c.442-1G>A, c.841C>G, c.1197A>T, c.926C>A, c.136G>A, c.1161C>A, c.829T>G, c.183C>G, c.667A>T, c.691T>C, c.143T>C, c.1042C>G, c.1238G>C, c.737delC, c.1315+1G>A, c.911A>G, c.619A>G, c.1159T>C, c.842+5G>A, c.715G>A, c.491T>C, c.1315+6T>A, c.722delG, c.165delT, c.47_48delCT, c.1127delA, c.1172G>T, c.119C>T, c.439C>T, c.193A>G, c.671T>C, c.1196_1199delTAAG, c.1114A>T, c.521T>C, c.1220delC, c.1144T>C, c.482T>C, c.1315+2T>C, c.842+2T>A, c.398_401delATCA, c.355C>T, c.719T>C, c.838G>A, c.509+1G>A, c.58C>T, c.502T>C, c.806delT, c.826A>G, c.842+3G>C, c.740G>A, c.1289T>C, c.169-13T>G, c.1066-11G>A, c.241A>C, c.194T>A, c.968_970delCAA, c.442-2A>C, c.1200-1G>A, c.1315+4A>G, c.964G>A, c.960G>C, c.1200delG, c.111_112insC, c.503delA, c.1012G>T, c.776C>T, c.1199+1G>A, c.632delC, c.707-1G>A, c.789C>G, c.1180G>C, c.755G>A, c.692C>T, c.117C>G, c.884C>G, c.1199+2T>C, c.169-2A>G, c.199T>C, c.856G>A, c.529G>A, c.664_665delGA, c.953T>C, c.514C>T, c.970-2A>C, c.969+1G>A, c.724C>T, c.1183G>C, c.1006C>T, c.712A>C, c.441+3G>C, c.739G>A, c.259A>C, c.731C>T, c.847A>T, c.941delC, c.781C>T, c.1340C>A, c.580_581delCT, c.137delG, c.569T>C, c.635T>C, c.734T>A, c.1199G>A, c.733G>C, c.1033G>A, c.810A>T, c.473G>A, c.490A>G, c.208T>C, c.601C>T, c.916delA, c.665A>G, c.1229T>C, c.116_118delTCT, c.809G>A, c.648C>G, c.359G>A, c.264dupG, c.250G>T, c.1057delG, c.226G>T, c.842+1G>A, c.533A>G, c.907T>C, c.1301C>A, c.1222C>T, c.1024G>A, c.194T>C, c.441+5G>T, c.1024delG, c.508C>G, c.1065+1G>A, c.535T>C, c.1219C>T, c.1076C>G, c.331C>T, c.974A>G, c.889C>T, c.673C>A, c.311C>A, c.912+1G>A, c.1066-1G>A, c.764T>C, c.638T>C, c.890G>A, c.1220C>T, c.1068C>A, c.472C>T, c.1200-2A>G, c.842C>T

Phosphoribosylpyrophosphate Synthetase Superactivity. Gene:*PRPS1*. **Exons**: NM_002764.3:1-7. **Variants(2)**: c.398A>C, c.455T>C

Phosphoserine Aminotransferase Deficiency. Gene:*PSAT1*. **Exons**: NM_058179.2:1-9. **Variants(1)**: c.299A>C

POLG-Related Disorders. Gene:*POLG*. **Exons**: NM_002693.2:2-23. **Variants(18)**: c.695G>A, c.2591A>G, c.1879C>T, c.3218C>T, c.911T>G, c.2209G>C, c.679C>T, c.1399G>A, c.2557C>T, c.3630dupC, c.2243G>C, c.8G>C, c.752C>T, c.1491G>C, c.2794C>T, c.2T>C, c.2542G>A, c.2617G>T

Pontocerebellar Hypoplasia. Gene:*TSEN54*. **Exons**: NM_207346.2:3-11. **Variants(3)**: c.919G>T, c.736C>T, c.1027C>T

Primary Autosomal Recessive Microcephaly 1. Gene:*MCPH1*. **Exons**: NM_024596.3:1-14. **Variants(5)**: c.74C>G, c.566_567insA, c.215C>T, c.302C>G, c.427_428insA

Primary Autosomal Recessive Microcephaly 2. Gene:*WDR62*. **Exons**: NM_001083961.1:1-32. **Variants(5)**: c.1313G>A, c.1531G>A, c.671G>C, c.193G>A, c.1408C>T

Primary Autosomal Recessive Microcephaly 3. Gene:*CDK5RAP2*. **Exons**: NM_018249.4:1-38. **Variants(6)**: c.4441C>T, c.4672C>T, c.246T>A, c.524_528delAGGCA, c.700G>T, c.4546G>T

Primary Autosomal Recessive Microcephaly 5. Gene:*ASPM*. **Exons**: NM_018136.4:1-28. **Variants(64)**: c.4795C>T, c.9677dupG, c.2389C>T, c.7491_7495delTATTA, c.3527C>G, c.1729_1730delAG, c.3477_3481delCGCTA, c.9697C>T, c.3188T>G, c.6189T>G, c.9730C>T, c.1590delA, c.6337_6338delAT, c.1366G>T, c.5136C>A, c.1406_1413delATCCTAAA, c.8844delC, c.7894C>T, c.7761T>G, c.9178C>T, c.3796G>T, c.9595A>T, c.719_720delCT, c.1002delA, c.9238A>T, c.577C>T, c.9539A>C, c.3811C>T, c.9190C>T, c.7782_7783delGA, c.5149delA, c.9557C>G, c.440delA, c.8131_8132delAA, c.10059C>A, c.8668C>T, c.9747_9748delCT, c.1260_1266delTCAAGTC, c.1179delT, c.3055C>T, c.3082G>A, c.9789T>A, c.1990C>T, c.2938C>T, c.3663delG, c.2967G>A, c.9492T>G, c.4858_4859delAT, c.9159delA, c.9319C>T, c.8378delT, c.3710C>G, c.8508_8509delGA, c.4195dupA, c.9115_9118dupCATT, c.1154_1155delAG, c.6732delA, c.9685delA, c.7860_7861delGA, c.3978G>A, c.349C>T, c.1959_1962delCAAA, c.9754delA, c.4583delA

Primary Autosomal Recessive Microcephaly 6. Gene:*CENPJ*. **Exons**: NM_018451.4:2-17. **Variants(3)**: c.3704A>T, c.3243_3246delTCAG, c.18delC

Primary Autosomal Recessive Microcephaly 7. Gene:*STIL*. **Exons**: NM_003035.2:2-17. **Variants(4)**: c.2826+1G>A, c.3715C>T, c.2392T>G, c.3655delG

Primary Autosomal Recessive Microcephaly 9. Gene:*CEP152*. **Exons**: NM_014985.3:2-26. **Variants(1)**: c.794A>C

Primary Coenzyme Q10 deficiency 1. Gene:*COQ2*. **Exons**: NM_015697.7:2-7. **Variants(3)**: c.890A>G, c.683A>G, c.590G>A

Primary Coenzyme Q10 deficiency 2. Gene:*PDSS1*. **Exons**: NM_014317.3:3-12. **Variants(1)**: c.924T>G

Primary Coenzyme Q10 deficiency 3. Gene:*PDSS2*. **Exons**: NM_020381.3:1-8. **Variants(2)**: c.964C>T, c.1145C>T

Primary Coenzyme Q10 deficiency 4. Gene:*ADCK3*. **Exons**: NM_020247.4:2-15. **Variants(7)**: c.1645G>A, c.1541A>G, c.637C>T, c.1813_1814insG, c.815G>T, c.1651G>A, c.815G>A

Primary Hyperoxaluria Type I. Gene:*AGXT*. **Exons**: NM_000030.2:1-11. **Variants(12)**: c.454T>A, c.322T>C, c.466G>A, c.731T>C, c.613T>C, c.560C>T, c.697C>T, c.121G>A, c.508G>A, c.738G>A, c.33_34insC, c.245G>A

Primary Hyperoxaluria Type II. Gene:*GRHPR*. **Exons**: NM_012203.1:1-9. **Variants(3)**: c.103delG, c.295C>T, c.403_404+2delAAGT

Progressive Myoclonic Epilepsy 1A. Gene:*CSTB*. **Exons**: NM_000100.3:1-3. **Variants(2)**: c.212A>C, c.202C>T

Pycnodysostosis. Gene:*CTSK*. **Exons**: NM_000396.3:2-8. **Variants(5)**: c.721C>T, c.154A>T, c.236G>A, c.990A>G, c.926T>C

Pyridoxal 5'-Phosphate-dependent Epilepsy. Gene:*PNPO*. **Exons**: NM_018129.3:1-7. **Variants(2)**: c.685C>T, c.784T>C

Pyruvate Carboxylase Deficiency. Gene:*PC*. **Exons**: NM_000920.3:3-22. **Variants(4)**: c.467G>A, c.1748G>T, c.434T>C, c.1828G>A

Pyruvate Dehydrogenase Phosphatase Deficiency. Gene:*PDP1*. **Exons**: NM_018444.3:2. **Variants(2)**: c.851_853delTTC, c.277G>T

Pyruvate Kinase Deficiency. Gene:*PKLR*. **Exons**: NM_000298.5:1-11. **Variants(8)**: c.110G>A, c.1261C>A, c.1675C>T, c.1529G>A, c.1151C>T, c.1436G>A, c.487C>T, c.1456C>T

Rabson-Mendenhall Syndrome. Gene:*INSR*. **Exons**: NM_000208.2:2-22. **Variants(1)**: c.3079C>T

RAG1-Related Omenn Syndrome. Gene:*RAG1*. **Exons**: NM_000448.2:2. **Variants(3)**: c.1682G>A, c.1681C>T, c.983G>A

RAG1-Related Severe Combined Immunodeficiency. Gene:*RAG1*. **Exons**: NM_000448.2:2. **Variants(7)**: c.2320G>T, c.2164G>A, c.2326C>T, c.940C>T, c.2923C>T, c.2333G>A, c.2814T>G

RAG2-Related Severe Combined Immunodeficiency. Gene:*RAG2*. **Exons**: NM_000536.3:2. **Variants(4)**: c.115A>G, c.1352G>C, c.230C>A, c.686G>A

Raine Syndrome. Gene:*FAM20C*. **Exons**: NM_020223.3:2-10. **Variants(6)**: c.1351G>A, c.1163T>G, c.1093G>C, c.1645C>T, c.838G>A, c.773T>A

RAPSN-Related Congenital Myasthenic Syndrome. Gene:*RAPSN*. **Exons**: NM_005055.4:1-8. **Variants(5)**: c.264C>A, c.807C>A, c.490C>T, c.848T>C, c.484G>A

Refsum Disease. Gene:*PHYH*. **Exons**: NM_006214.3:2-9. **Variants(13)**: c.497-2A>G, c.805A>C, c.678+5G>T, c.734G>A, c.135-2A>G, c.164delT, c.679-1G>T, c.823C>T, c.135-1G>C, c.824G>A, c.610G>A, c.530A>G, c.678+2T>G

Renal-Hepatic-Pancreatic Dysplasia. Gene:*NPHP3*. **Exons**: NM_153240.4:1-27. **Variants(3)**: c.3340C>T, c.1985+5G>A, c.1729C>T

REN-Related Renal Tubular Dysgenesis. Gene:*REN*. **Exons**: NM_000537.3:1-10. **Variants(4)**: c.689G>A, c.404C>A, c.127C>T, c.145C>T

Restrictive Dermopathy, Lethal. Gene:*ZMPSTE24*. **Exons**: NM_005857.4:1-10. **Variants(3)**: c.1085_1086insT, c.715G>T, c.54dupT

Retinal cone dystrophy 3B. Gene:*KCNV2*. **Exons**: NM_133497.3:1-2. **Variants(10)**: c.1016_1024delACCTGGTGG, c.8_11delAACA, c.916G>T, c.357_358insC, c.491T>C, c.767C>G, c.226C>T, c.1376G>A, c.427G>T, c.325C>T

Retinitis Pigmentosa 12. Gene:*CRB1*. **Exons**: NM_201253.2:1-12. **Variants(6)**: c.2290C>T, c.2983G>T, c.3299T>C, c.3541T>C, c.2401A>T, c.3122T>C

Retinitis Pigmentosa 14. Gene:*TULP1*. **Exons**: NM_003322.3:1-15. **Variants(6)**: c.1259G>C, c.1471T>C, c.1511_1521delTGCAGTTCGGC, c.99+1G>A, c.1376T>A, c.1444C>T

Retinitis Pigmentosa 19. Gene:*ABCA4*. **Exons**: NM_000350.2:1-50. **Variants(3)**: c.1938-1G>A, c.1848delA, c.4539+1G>T

Retinitis Pigmentosa 2. Gene:*RP2*. **Exons**: NM_006915.2:1-5. **Variants(7)**: c.358C>T, c.453delC, c.353G>T, c.305_306insT, c.353G>A, c.76C>T, c.453C>G

Retinitis Pigmentosa 20. Gene:*RPE65*. **Exons**: NM_000329.2:1-14. **Variants(7)**: c.1543C>T, c.1087C>A, c.1355T>G, c.271C>T, c.394G>A, c.1102T>C, c.1022T>C

Retinitis Pigmentosa 25. Gene:*EYS*. **Exons**: NM_001142800.1:4-43. **Variants(3)**: c.5928-2A>G, c.6714delT, c.5857G>T

Retinitis Pigmentosa 26. Gene:*CERKL*. **Exons**: NM_201548.4:1-13. **Variants(4)**: c.420delT, c.598A>T, c.769C>T, c.780delT

Retinitis Pigmentosa 3. Gene:*RPGR*. **Exons**: NM_001034853.1:2-15. **Variants(11)**: c.674_675delCC, c.179G>T, c.654_655delGA, c.823G>A, c.296C>A, c.389T>G, c.517G>C, c.173_174insA, c.846_847delAA, c.1433_1436delTGAC, c.703C>T

Retinitis Pigmentosa 35. Gene:*SEMA4A*. **Exons**: NM_022367.3:2-15. **Variants(2)**: c.1033G>C, c.1049T>G

Retinitis Pigmentosa 36. Gene:*PRCD*. **Exons**: NM_001077620.2:1-3. **Variants(2)**: c.5G>A, c.64C>T

Retinitis Pigmentosa 37. Gene:*NR2E3*. **Exons**: NM_014249.2:1-8. **Variants(1)**: c.1034_1038delTGCAG

Retinitis Pigmentosa 38. Gene:*MERTK*. **Exons**: NM_006343.2:1-19. **Variants(6)**: c.2323C>T, c.61+1G>A, c.2070_2074delAGGAC, c.2189+1G>T, c.1951C>T, c.1605-2A>G

Retinitis Pigmentosa 4. Gene:*RHO*. **Exons**: NM_000539.3:1-5. **Variants(2)**: c.448G>A, c.745G>T

Retinitis Pigmentosa 40. Gene:*PDE6B*. **Exons**: NM_000283.3:1-22. **Variants(4)**: c.1669C>T, c.1580T>C, c.1591C>T, c.892C>T

Retinitis Pigmentosa 41. Gene:*PROM1*. **Exons**: NM_006017.2:1-26. **Variants(2)**: c.1726C>T, c.1841delG

Retinitis Pigmentosa 43. Gene:*PDE6A*. **Exons**: NM_000440.2:1-22. **Variants(4)**: c.1683G>A, c.2053G>A, c.304C>A, c.1749C>G

Retinitis Pigmentosa 44. Gene:*RGR*. **Exons**: NM_001012720.1:1-7. **Variants(1)**: c.196A>C

Retinitis Pigmentosa 45. Gene:*CNGB1*. **Exons**: NM_001297.4:2-33. **Variants(2)**: c.2978G>T, c.3462+1G>A

Retinitis Pigmentosa 46. Gene:*IDH3B*. **Exons**: NM_006899.3:1-12. **Variants(2)**: c.589delA, c.395T>C

Retinitis Pigmentosa 49. Gene:*CNGA1*. **Exons**: NM_000087.3:4-11. **Variants(3)**: c.427A>T, c.959C>T, c.1972delA

Retinitis Pigmentosa 50. Gene:*BEST1*. **Exons**: NM_004183.3:2-11. **Variants(1)**: c.418C>G

Retinitis Pigmentosa 56. Gene:*IMPG2*. **Exons**: NM_016247.3:1-19. **Variants(3)**: c.2716C>T, c.635C>G, c.2890C>T

Retinitis Pigmentosa 57. Gene:*PDE6G*. **Exons**: NM_002602.3:2-4. **Variants(1)**: c.187+1G>T

Retinitis Pigmentosa 59. Gene:*DHDDS*. **Exons**: NM_024887.3:2-9. **Variants(1)**: c.124A>G

Retinitis Pigmentosa 61. Gene:*CLRN1*. **Exons**: NM_174878.2:1-3. **Variants(2)**: c.92C>T, c.461T>G

Retinitis Pigmentosa 62. Gene:*MAK*. **Exons**: NM_001242957.1:2-15. **Variants(4)**: c.497G>A, c.388A>C, c.718C>T, c.37G>A

Retinitis Pigmentosa 65. Gene:*CDHR1*. **Exons**: NM_033100.2:2-17. **Variants(3)**: c.1463delG, c.338delG, c.524dupA

Rhizomelic Chondrodysplasia Punctata Type 1. Gene:*PEX7*. **Exons**: NM_000288.3:2-10. **Variants(7)**: c.903+1G>C, c.649G>A, c.618G>A, c.875T>A, c.653C>T, c.854A>G, c.694C>T

Rhizomelic Chondrodysplasia Punctata Type 3. Gene:*AGPS*. **Exons**: NM_003659.3:1-20. **Variants(2)**: c.1703C>T, c.926C>T

Roberts Syndrome. Gene:*ESCO2*. **Exons**: NM_001017420.2:2-11. **Variants(10)**: c.876_879delCAGA, c.1597dupT, c.751dupG, c.1111_1112insG, c.308_309delAA, c.764_765delTT, c.745_746delGT, c.879_880delAG, c.1615T>G, c.505C>T

RYR1 -Related Congenital Fiber-Type Disproportion. Gene:*RYR1*. **Exons**: NM_000540.2:1-90,92-106. **Variants(9)**: c.6104A>T, c.9000+1G>T, c.13480G>T, c.10204T>G, c.10616G>A, c.1205T>C, c.5333C>A, c.738T>G, c.4405C>T

Salla Disease. Gene:*SLC17A5*. **Exons**: NM_012434.4:1-11. **Variants(2)**: c.406A>G, c.115C>T

Sandhoff Disease. Gene:*HEXB*. **Exons**: NM_000521.3:1-14. **Variants(4)**: c.1367A>C, c.1250C>T, c.965delT, c.850C>T

SC Phocomelia Syndrome. Gene:*ESCO2*. **Exons**: NM_001017420.2:2-11. **Variants(4)**: c.604C>T, c.760_761insA, c.760delA, c.1269G>A

Schindler Disease. Gene:*NAGA*. **Exons**: NM_000262.2:1-9. **Variants(1)**: c.973G>A

Schinzel phocomelia syndrome. Gene:*WNT7A*. **Exons**: NM_004625.3:1-4. **Variants(2)**: c.610G>A, c.874C>T

Schneckenbecken Dysplasia. Gene:*SLC35D1*. **Exons**: NM_015139.2:1-12. **Variants(3)**: c.932G>A, c.193A>C, c.319C>T

Schwartz-Jampel Syndrome Type 1. Gene:*HSPG2*. **Exons**: NM_005529.5:2-97. **Variants(2)**: c.4595G>A, c.8464+4A>G

SCNN1A-Related Autosomal Recessive Pseudohypoaldosteronism type 1. Gene:*SCNN1A*. **Exons**: NM_001038.5:2-13. **Variants(1)**: c.1522C>T

SCNN1B-Related Autosomal Recessive Pseudohypoaldosteronism type 1. Gene:*SCNN1B*. **Exons**: NM_000336.2:2-13. **Variants(1)**: c.109G>A

SCNN1G-Related Autosomal Recessive Pseudohypoaldosteronism type 1. Gene:*SCNN1G*. **Exons**: NM_001039.3:2-13. **Variants(2)**: c.1627delG, c.1570-1G>A

Seckel Syndrome 1. Gene:*ATR*. **Exons**: NM_001184.3:1-47. **Variants(1)**: c.5635G>T

Seckel syndrome 5. Gene:*CEP152*. **Exons**: NM_014985.3:2-26. **Variants(1)**: c.2034T>G

Senior-Loken Syndrome 4. Gene:*NPHP4*. **Exons**: NM_015102.3:2-30. **Variants(2)**: c.2335C>T, c.1972C>T

Senior-Loken Syndrome 5. Gene:*IQCB1*. **Exons**: NM_001023570.2:3-15. **Variants(4)**: c.1465C>T, c.1069C>T, c.1036G>T, c.1381C>T

SEPN1-related Congenital Muscular Dystrophy. Gene:*SEPN1*. **Exons**: NM_020451.2:2,4-13. **Variants(6)**: c.1385G>A, c.1358G>C, c.1384T>G, c.943G>A, c.1397G>A, c.818G>A

Septo-Optic Dysplasia. Gene:*HESX1*. **Exons**: NM_003865.2:1-4. **Variants(4)**: c.357+2T>C, c.478C>T, c.450_451delCA, c.77T>C

Severe combined immunodeficiency, Athabascan type. Gene:*DCLRE1C*. **Exons**: NM_001033855.1:1-7,9-14. **Variants(5)**: c.362+1G>T, c.597C>A, c.972+1G>C, c.917+1G>A, c.780+1delG

Severe Combined Immunodeficiency,T-negative/B-positive type. Gene:*JAK3*. **Exons**: NM_000215.3:2-24. **Variants(4)**: c.299A>G, c.1695C>A, c.1333C>T, c.1172_1173insG

Short-Rib Thoracic Dysplasia 2. Gene:*IFT80*. **Exons**: NM_020800.2:2-20. **Variants(3)**: c.2101G>C, c.1646_1648delTAT, c.315C>G

Short-Rib Thoracic Dysplasia 3. Gene:*DYNC2H1*. **Exons**: NM_001080463.1:1-90. **Variants(10)**: c.5959A>G, c.10063G>T, c.5971A>T, c.8512C>T, c.9044A>G, c.4610A>G, c.3719T>C, c.6614G>A, c.1759C>T, c.7382G>T

Shwachman-Diamond Syndrome. Gene:*SBDS*. **Exons**: NM_016038.2:1,3,5. **Variants(7)**: c.377G>C, c.652C>T, c.258+2T>C, c.258+1G>C, c.183_184delTAinsCT, c.120delG, c.505C>T

Sickle Cell Anemia. Gene:*HBB*. **Exons**: NM_000518.4:1-3. **Variants(5)**: c.19G>A, c.20A>T, c.79G>A, c.364G>A, c.364G>C

Simpson-Golabi-Behmel Syndrome Type 2. Gene:*OFD1*. **Exons**: NM_003611.2:1-23. **Variants(1)**: c.2126_2129dupAAAG

Sjogren-Larsson syndrome. Gene:*ALDH3A2*. **Exons**: NM_000382.2:1-10. **Variants(7)**: c.1157A>G, c.641G>A, c.521delT, c.809delG, c.943C>T, c.1297_1298delGA, c.1307_1311dupACAAA

SLC6A8-Related Creatine Transporter Deficiency. Gene:*SLC6A8*. **Exons**: NM_005629.3:2,5-13. **Variants(9)**: c.1631C>T, c.1011C>G, c.1540C>T, c.395G>T, c.1473C>G, c.1222_1224delTTC, c.1141G>C, c.321_323delCTT, c.1661C>T

Smith-Lemli-Opitz syndrome. Gene:*DHCR7*. **Exons**: NM_001360.2:3-9. **Variants(19)**: c.356A>T, c.724C>T, c.976G>T, c.907G>A, c.1210C>T, c.278C>T, c.744G>T, c.964-1G>C, c.1054C>T, c.453G>A, c.1337G>A, c.1055G>A, c.904T>C, c.725G>A, c.506C>T, c.730G>A, c.832-1G>C, c.1228G>A, c.1342G>A

Spastic paraplegia 5A, autosomal recessive. Gene:*CYP7B1*. **Exons**: NM_004820.3:2-6. **Variants(2)**: c.825T>A, c.187C>T

Spinal Muscular Atrophy.Gene:*SMN1*. **variants(1)**: EX7 del

Spondylocostal dysostosis 1. Gene:*DLL3*. **Exons**: NM_016941.3:1-3,5-6,8. **Variants(2)**: c.1511G>A, c.712C>T

Spondyloepimetaphyseal Dysplasia. Gene:*MATN3*. **Exons**: NM_002381.4:2-8. **Variants(1)**: c.910T>A

Stickler Syndrome Type IV. Gene:*COL9A1*. **Exons**: NM_001851.4:1-38. **Variants(1)**: c.883C>T

Stuve-Wiedemann Syndrome. Gene:*LIFR*. **Exons**: NM_002310.5:2-20. **Variants(4)**: c.2013_2014insT, c.653dupT, c.1789C>T, c.171_174delTAAC

Succinic Semialdehyde Dehydrogenase Deficiency. Gene:*ALDH5A1*. **Exons**: NM_001080.3:2-10. **Variants(3)**: c.1234C>T, c.612G>A, c.1226G>A

Sulfite Oxidase Deficiency. Gene:*SUOX*. **Exons**: NM_000456.2:4-6. **Variants(2)**: c.794C>A, c.650G>A

Tay-Sachs Disease. Gene:*HEXA*. **Exons**: NM_000520.4:1-14. **Variants(30)**: c.409C>T, c.1073+1G>A, c.1260G>C, c.1453T>C, c.116T>G, c.672+1G>A, c.1444G>A, c.1511G>A, c.915_917delCTT, c.1510delC, c.509G>A, c.540C>G, c.1177C>T, c.508C>T, c.1274_1277dupTATC, c.772G>C, c.78G>A, c.1510C>T, c.987G>A, c.805+1G>A, c.532C>T, c.1495C>T, c.1421+1G>C, c.1496G>A, c.533G>T, c.749G>A, c.1176G>A, c.533G>A, c.805G>A, c.1A>G

T-cell immunodeficiency, Congenital alopecia, and Nail dystrophy. Gene:*FOXN1*. **Exons**: NM_003593.2:1-8. **Variants(1)**: c.763C>T

Tetrahydrobiopterin (BH4)-deficient hyperphenylalaninemia (HPA). Gene:*PTS*. **Exons**: NM_000317.2:1-6. **Variants(1)**: c.286G>A

TMEM67-Related COACH Syndrome. Gene:*TMEM67*. **Exons**: NM_153704.5:1-28. **Variants(2)**: c.2498T>C, c.1769T>C

Tyrosinemia Type I. Gene:*FAH*. **Exons**: NM_000137.2:1-14. **Variants(13)**: c.1090G>T, c.47A>T, c.554-1G>T, c.1069G>T, c.786G>A, c.836A>G, c.1141A>G, c.782C>T, c.103G>A, c.1062+5G>A, c.1027G>T, c.698A>T, c.1009G>A

Tyrosinemia Type II. Gene:*TAT*. **Exons**: NM_000353.2:2-12. **Variants(5)**: c.169C>T, c.1085G>T, c.668C>G, c.236-5A>G, c.1249C>T

Tyrosinemia Type III. Gene:*HPD*. **Exons**: NM_002150.2:1-14. **Variants(3)**: c.1005C>G, c.774T>G, c.600C>G

Usher Syndrome Type 1B. Gene:*MYO7A*. **Exons**: NM_000260.3:2-49. **Variants(8)**: c.635G>A, c.448C>T, c.1884C>A, c.5227C>T, c.6025delG, c.634C>T, c.1996C>T, c.3504-1G>C

Usher Syndrome Type 1C. Gene:*USH1C*. **Exons**: NM_153676.3:1-27. **Variants(1)**: c.238_239insC

Usher Syndrome Type 1D. Gene:*CDH23*. **Exons**: NM_022124.5:2-70. **Variants(8)**: c.7823G>A, c.193delC, c.6050-9G>A, c.4504C>T, c.288+1G>A, c.9565C>T, c.4488G>C, c.7224+5G>A

Usher Syndrome Type 1F. Gene:*PCDH15*. **Exons**: NM_033056.3:2-33. **Variants(5)**: c.3718-2A>G, c.733C>T, c.7C>T, c.1940C>G, c.1088delT

Usher Syndrome Type 1G. Gene:*USH1G*. **Exons**: NM_173477.2:1-3. **Variants(5)**: c.113G>A, c.143T>C, c.394_395insG, c.832_851delTCGGACGAGGACAGCGTCTC, c.186_187delCA

Usher Syndrome Type 2A. Gene:*USH2A*. **Exons**: NM_206933.2:2-72. **Variants(15)**: c.14803C>T, c.5975A>G, c.9799T>C, c.2209C>T, c.779T>G, c.2898delG, c.4338_4339delCT, c.12574C>T, c.9424G>T, c.2299delG, c.2276G>T, c.920_923dupGCCA, c.11864G>A, c.8981G>A, c.956G>A

Usher Syndrome Type 2C. Gene:*GPR98*. **Exons**: NM_032119.3:1-90. **Variants(8)**: c.17668_17669delAT, c.18131A>G, c.8790delC, c.8713_8716dupAACA, c.2258_2270delAAGTGCTGAAATC, c.5357_5358delAA, c.17137delG, c.6901C>T

Usher Syndrome Type 3A. Gene:*CLRN1*. **Exons**: NM_174878.2:1-3. **Variants(7)**: c.449T>C, c.118T>G, c.144T>G, c.528T>G, c.459_461delATT, c.189C>A, c.359T>A

Vitamin D-resistant Rickets Type IIA. Gene:*VDR*. **Exons**: NM_001017535.1:4-11. **Variants(7)**: c.915C>G, c.137G>A, c.1036G>A, c.88C>T, c.454C>T, c.239G>A, c.885C>A

VLDLR-Associated Cerebellar Hypoplasia. Gene:*VLDLR*. **Exons**: NM_003383.3:2-19. **Variants(3)**: c.1342C>T, c.2339delT, c.769C>T

Waardenburg Syndrome Type 3. Gene:*PAX3*. **Exons**: NM_181457.3:1-8. **Variants(2)**: c.251C>T, c.268T>C

Waardenburg Syndrome Type 4A. Gene:*EDNRB*. **Exons**: NM_000115.3:2-8. **Variants(2)**: c.548C>G, c.828G>T

Waardenburg Syndrome Type 4B. Gene:*EDN3*. **Exons**: NM_207034.1:2-5. **Variants(1)**: c.262_263delGCinsT

Warburg Micro Syndrome 1. Gene:*RAB3GAP1*. **Exons**: NM_012233.2:1-24. **Variants(5)**: c.1410C>A, c.2011C>T, c.1734G>A, c.748+1G>A, c.899+1G>A

Wilson Disease. Gene:*ATP7B*. **Exons**: NM_000053.3:1-21. **Variants(174)**: c.1772G>A, c.2131G>A, c.1369C>T, c.3646G>A, c.3359T>A, c.3903+1delG, c.2304_2305insG, c.3665A>T, c.2887C>T, c.1847G>A, c.3451C>T, c.1646T>C, c.2230T>C, c.2279C>T, c.3877G>A, c.2930C>T, c.2865+1G>A, c.3140delA, c.2828G>A, c.3818C>A, c.3505A>G, c.2827G>A, c.2009_2015delATATGCT, c.3713_3714delAA, c.314C>A, c.3436G>A, c.2662A>C, c.2293G>A, c.1639delC, c.3506T>C, c.1947-2A>G, c.3955C>T, c.397delT, c.2810delT, c.3402delC, c.4022-2A>G, c.1745_1746delTA, c.2122-8T>G, c.3412+1G>A, c.3091A>G, c.3244G>T, c.2463delC, c.2731-2A>G, c.3556G>A, c.3207C>A, c.2605G>A, c.2604delC, c.3659C>T, c.1924G>C, c.3182G>A, c.3699+1G>C, c.2519C>T, c.2335T>G, c.2297C>G, c.1708-5T>G, c.122A>G, c.2975C>A, c.2513delA, c.1958C>A, c.3517G>A, c.3895C>T, c.3061-12T>A, c.3147delC, c.2332C>G, c.2305A>G, c.2575+2T>C, c.2924C>A, c.3449delA, c.2132G>A, c.3840_3841insTAG, c.865C>T, c.4114C>T, c.813C>A, c.2862_2865delTCCT, c.1846C>T, c.3053C>T, c.3097A>G, c.3731delT, c.1475T>C, c.2356-2A>G, c.3104G>T, c.2071G>A, c.2899A>T, c.3191A>C, c.1340_1343delAAAC, c.3084_3085delGA, c.2755C>G, c.3317T>A, c.654_655delCC, c.3694A>C, c.3829C>T, c.448_452delGAGGG, c.1531C>T, c.343C>T, c.1470C>A, c.2447+2T>A, c.3800A>C, c.1436delC, c.3784G>T, c.3305T>C, c.1968C>A, c.4195delC, c.2804C>T, c.2223T>A, c.2659delG, c.2407G>A, c.2337G>A, c.2304delC, c.1934T>G, c.2695A>T, c.4021G>A, c.2866-6T>G, c.3904-2A>G, c.802_808delTGTAAGT, c.3008C>T, c.3128T>C, c.2333G>A, c.2108G>A, c.4088C>T, c.4112T>C, c.892delC, c.2871delC, c.3796G>A, c.213_214delAT, c.2294A>G, c.2333G>T, c.3263T>A, c.1568T>A, c.3700-2A>T, c.3060+5G>T, c.3190G>A, c.2129G>C, c.3086C>T, c.3532A>G, c.254G>T, c.2623G>A, c.3643G>T, c.3886G>A, c.1186G>T, c.1782delT, c.2762G>A, c.2998G>A, c.2807T>A, c.2532delA, c.2795C>A, c.2121+3A>G, c.2128G>A, c.2438_2440delTAAinsAT, c.2975C>T, c.2572A>G, c.3301G>A, c.3809A>G, c.2621C>T, c.328C>T, c.3443T>C, c.3122G>C, c.561T>A, c.3598C>T, c.2336G>A, c.453delC, c.3111delC, c.3548C>G, c.3007G>A, c.1995G>A, c.1630C>T, c.2123T>C, c.3295G>A, c.845delT, c.2906G>A, c.331C>T, c.2383C>T, c.3722C>T, c.2570T>C, c.3029A>C

Wiskott-Aldrich Syndrome. Gene:*WAS*. **Exons**: NM_000377.2:1-12. **Variants(4)**: c.257G>T, c.257G>A, c.244T>C, c.100C>T

Wolcott-Rallison Syndrome. Gene:*EIF2AK3*. **Exons**: NM_004836.5:2-17. **Variants(2)**: c.1763G>A, c.994G>T

Wolfram Syndrome. Gene:*WFS1*. **Exons**: NM_006005.3:2-8. **Variants(6)**: c.676C>T, c.1511C>T, c.1944G>A, c.2084G>T, c.2171C>T, c.2455C>T

Xeroderma Pigmentosum Group A. Gene:*XPA*. **Exons**: NM_000380.3:2-6. **Variants(4)**: c.348T>A, c.323G>T, c.619C>T, c.682C>T

Xeroderma Pigmentosum Group B. Gene:*ERCC3*. **Exons**: NM_000122.1:2-15. **Variants(3)**: c.1273C>T, c.296T>C, c.1633C>T

Xeroderma Pigmentosum Group D. Gene:*ERCC2*. **Exons**: NM_000400.3:2-23. **Variants(4)**: c.1621A>C, c.2047C>T, c.1454T>C, c.2176C>T

Xeroderma Pigmentosum Group E. Gene:*DDB2*. **Exons**: NM_000107.2:1-10. **Variants(4)**: c.919G>T, c.937C>T, c.730A>G, c.818G>A

Xeroderma Pigmentosum Group F. Gene:*ERCC4*. **Exons**: NM_005236.2:1-11. **Variants(4)**: c.1730dupA, c.2281_2284delTTTG, c.706T>C, c.1765C>T

Xeroderma Pigmentosum Group G. Gene:*ERCC5*. **Exons**: NM_000123.3:1-15. **Variants(12)**: c.2620G>A, c.526C>T, c.2170delT, c.215C>A, c.2878G>T, c.2573T>C, c.2972delT, c.1494delA, c.2751delA, c.406C>T, c.787C>T, c.1115_1118delGGAA

X-Linked Adrenal Hypoplasia Congenita. Gene:*NR0B1*. **Exons**: NM_000475.4:1-2. **Variants(22)**: c.813C>G, c.591C>A, c.788T>A, c.704G>A, c.1319A>T, c.315G>C, c.273C>A, c.513G>A, c.1107G>A, c.1169delA, c.847C>T, c.1316T>G, c.1138T>G, c.388_389delTA, c.873G>C, c.1183C>T, c.800G>C, c.1142T>A, c.109C>T, c.890T>C, c.1146G>T, c.1197C>A

X-Linked Adrenoleukodystrophy. Gene:*ABCD1*. **Exons**: NM_000033.3:1-7,10. **Variants(12)**: c.443A>G, c.1390C>T, c.1451C>G, c.1202G>A, c.1552delC, c.1415_1416delAG, c.1165C>G, c.1252C>T, c.871G>A, c.1552C>T, c.1429G>T, c.796G>A

X-Linked Agammaglobulinemia. Gene:*BTK*. **Exons**: NM_000061.2:2-19. **Variants(28)**: c.1559G>A, c.1838G>A, c.1082A>G, c.1820C>A, c.1275C>A, c.1685G>C, c.83G>A, c.1223T>C, c.1684C>T, c.43C>T, c.1906G>T, c.1766A>G, c.862C>T, c.763C>T, c.1741T>C, c.1506C>A, c.37C>T, c.1773C>A, c.718G>T, c.755G>A, c.1574G>A, c.1558C>T, c.1288A>G, c.1001A>C, c.338T>A, c.97A>C, c.919A>G, c.2T>C

X-Linked Centronuclear Myopathy. Gene:*MTM1*. **Exons**: NM_000252.2:2-15. **Variants(4)**: c.205C>T, c.141_144delAGAA, c.721C>T, c.670C>T

X-linked Charcot-Marie-Tooth disease 5. Gene:*PRPS1*. **Exons**: NM_002764.3:1-7. **Variants(2)**: c.344T>C, c.129A>C

X-linked chondrodysplasia punctata 1. Gene:*ARSE*. **Exons**: NM_000047.2:2-11. **Variants(6)**: c.1743G>A, c.1442C>T, c.1732C>T, c.410G>T, c.119T>G, c.410G>C

X-linked Deafness 1. Gene:*PRPS1*. **Exons**: NM_002764.3:1-7. **Variants(4)**: c.259G>A, c.869T>C, c.916G>A, c.193G>A

X-linked Deafness 2. Gene:*POU3F4*. **Exons**: NM_000307.3:1. **Variants(7)**: c.967C>G, c.990A>T, c.1000A>G, c.499C>T, c.935C>T, c.604A>T, c.950T>G

X-linked Emery-Dreifuss Muscular Dystrophy 1. Gene:*EMD*. **Exons**: NM_000117.2:1-6. **Variants(2)**: c.631_635delCGTGC, c.547C>A

X-linked Emery-Dreifuss Muscular Dystrophy 6. Gene:*FHL1*. **Exons**: NM_001449.4:3-7. **Variants(4)**: c.310T>C, c.689-479G>A, c.672C>G, c.625T>C

X-Linked Hyper IgM Syndrome. Gene:*CD40LG*. **Exons**: NM_000074.2:1-5. **Variants(9)**: c.761C>T, c.680G>T, c.107T>G, c.418T>G, c.464T>C, c.632C>A, c.368C>A, c.419G>A, c.703G>C

X-Linked Hypohidrotic Ectodermal Dysplasia. Gene:*EDA*. **Exons**: NM_001399.4:1-2,4-8. **Variants(9)**: c.466C>T, c.206G>T, c.826C>T, c.183C>G, c.467G>A, c.463C>T, c.1045G>A, c.725delG, c.573_574insT

X-linked Infantile Spasm Syndrome 1. Gene:*ARX*. **Exons**: NM_139058.2:1,3,5. **Variants(2)**: c.1058C>T, c.81C>G

X-Linked Infantile Spinal Muscular Atrophy. Gene:*UBA1*. **Exons**: NM_003334.3:2-26. **Variants(3)**: c.1617G>T, c.1639A>G, c.1731C>T

X-Linked Juvenile Retinoschisis. Gene:*RS1*. **Exons**: NM_000330.3:1-6. **Variants(3)**: c.221G>T, c.214G>C, c.325G>C

X-Linked Leigh Syndrome. Gene:*PDHA1*. **Exons**: NM_000284.3:2-11. **Variants(2)**: c.773A>C, c.787C>G

X-Linked Lymphoproliferative syndrome 1. Gene:*SH2D1A*. **Exons**: NM_002351.4:1-4. **Variants(8)**: c.95G>C, c.302C>T, c.385T>A, c.172C>T, c.203C>T, c.163C>T, c.164G>T, c.3G>T

X-Linked Ocular Albinism. Gene:*GPR143*. **Exons**: NM_000273.2:2-9. **Variants(6)**: c.397T>A, c.695C>A, c.397T>C, c.455G>A, c.992_993insCG, c.816_829delGCAAACAGATATCA

X-Linked Severe Combined Immunodeficiency. Gene:*IL2RG*. **Exons**: NM_000206.2:1-8. **Variants(10)**: c.458T>A, c.452T>C, c.923C>A, c.454+1G>A, c.343T>C, c.854G>A, c.865C>T, c.186T>A, c.664C>T, c.355A>T

X-Linked Severe Congenital Neutropenia. Gene:*WAS*. **Exons**: NM_000377.2:1-12. **Variants(3)**: c.809T>C, c.814T>C, c.881T>C

X-linked Sideroblastic Anemia and Ataxia. Gene:*ABCB7*. **Exons**: NM_004299.3:1-16. **Variants(3)**: c.1203T>G, c.1234G>C, c.1300G>A

X-linked Sideroblastic Anemia. Gene:*ALAS2*. **Exons**: NM_000032.4:2-11. **Variants(4)**: c.1354C>T, c.569A>T, c.1427T>A, c.1163C>G

X-Linked Spastic Paraplegia 2. Gene:*PLP1*. **Exons**: NM_000533.3:1-7. **Variants(1)**: c.409C>T

X-linked Thrombocytopenia. Gene:*WAS*. **Exons**: NM_000377.2:1-12. **Variants(4)**: c.1442T>A, c.173C>G, c.167C>T, c.134C>T

Yunis-Varon syndrome. Gene:*FIG4*. **Exons**: NM_014845.5:1-23. **Variants(3)**: c.1260_1261delGT, c.311G>A, c.831_838delTAAATTTG

2）Variants of group B

Acyl-CoA Dehydrogenase Deficiency,Short-Chain. Gene:*ACADS*. **Exons**: NM_000017.2:2-10. **Variants(1)**: c.319C>T

Alpha1-Antitrypsin Deficiency. Gene:*SERPINA1*. **Exons**: NM_000295.4:2-5. **Variants(9)**: c.839A>T, c.347T>A, c.272G>A, c.230C>T, c.194T>C, c.863A>T, c.1096G>A, c.187C>T, c.415G>A

Ataxia With Vitamin E Deficiency. Gene:*TTPA*. **Exons**: NM_000370.3:2-5. **Variants(1)**: c.575G>A

Autosomal Recessive Deafness 1A. Gene:GJB2**. Exons:** NM_004004.5:2**. Variants(4):** c.520T>C, c.535G>C, c.551G>A, c.101T>C

Autosomal Recessive Polycystic Kidney Disease. Gene:PKHD1**. Exons:** NM_138694.3:2-67. **Variants(3):** c.10926G>A, c.5125C>T, c.8581A>G

Beta-thalassemia. Gene:HBB**. Exons:** NM_000518.4:1-3**. Variants(17)**:c.-50-u101C>T, c.-50-u88C>T, c.102_104delGGT, c.182T>A, c.295G>A, c.315+4_315+5delAG, c.315delG, c.316-125A>G, c.316-238C>T, c.316-8T>G, c.344T>C, c.364G>T, c.378_379insCCA, c.382C>T, c.383A>C, c.86T>G, c.92_94dupGGC

Canavan Disease. Gene:*ASPA*. **Exons**: NM_000049.2:1-6. **Variants(2)**: c.212G>A, c.863A>G

Choroideremia. Gene:*CHM*. **Exons**: NM_000390.2:1-15. **Variants(2)**: c.1609+1_1609+2insT, c.1609+2_1609+3insT

Duchenne Muscular Dystrophy. Gene:DMD**. Exons:** NM_004006.2:1-79**. Variants(1):** c.1934A>G

Factor V Leiden thrombophilia. Gene:*F5*. **Exons**: NM_000130.4:1-25. **Variants(1)**: c.1601G>A

Factor XI Deficiency. Gene:*F11*. **Exons**: NM_000128.3:2-15. **Variants(6)**: c.166T>C, c.809A>T, c.901T>C, c.438C>A, c.1716+1G>A, c.403G>T

Familial Mediterranean Fever. Gene:*MEFV*. **Exons**: NM_000243.2:1-10. **Variants(1)**: c.2084A>G

GLDC-Related Glycine Encephalopathy. Gene:*GLDC*. **Exons**: NM_000170.2:2-25. **Variants(1)**: c.2405C>T

Glycogen Storage Disease Type II. Gene:GAA**. Exons:** NM_000152.3:2-20. **Variants(1):** c.-32-13T>G

Glycogen Storage Disease Type V. Gene:*PYGM*. **Exons**: NM_005609.2:1-20. **Variants(13)**: c.613G>A, c.2128_2130delTTC, c.1628A>C, c.1621G>T, c.2392T>C, c.1827G>A, c.1094C>T, c.1963G>A, c.255C>A, c.1722T>G, c.148C>T, c.1726C>T, c.1A>G

Hemophilia A. Gene:F8**. Exons:** NM_000132.3:1-26**. Variants(80)**: c.1660A>G, c.5944T>A, c.6658_6660delGCC, c.6301C>G, c.2059C>T, c.289G>C, c.6658G>C, c.1748A>G, c.5302C>T, c.2225A>G, c.6104T>C, c.2138A>T, c.5422C>T, c.5123G>A, c.5918A>T, c.6533G>A, c.6955C>T, c.1649G>A, c.6790A>G, c.1910A>G, c.979C>G, c.5218A>G, c.6278A>G, c.6113A>G, c.842C>A, c.908C>A, c.5305G>C, c.6021G>A, c.6067G>A, c.5938C>G, c.6532C>T, c.1018G>A, c.1688C>G, c.6920A>C, c.5329C>T, c.1589A>G, c.655G>A, c.6685C>T, c.1679G>C, c.896A>T, c.1992G>C, c.6622C>G, c.1733T>C, c.1280A>T, c.2043G>A, c.6371A>G, c.5093T>C, c.1700T>C, c.5303G>A, c.5305G>A, c.6575G>T, c.669A>T, c.2215G>A, c.1094A>G, c.871G>A, c.5180A>T, c.1736A>C, c.6214C>T, c.5900G>A, c.1930T>G, c.6089G>A, c.1730C>T, c.5954G>A, c.396A>C, c.5558C>T, c.1834C>T, c.2128G>T, c.6956C>T, c.446C>G, c.338A>G, c.1372C>T, c.1309C>T, c.361G>A, c.6932C>A, c.5143C>G, c.6119G>A, c.1648C>T, c.5096A>T, c.5501A>G, c.6787_6788insTTG

Hemophilia B. Gene:F9**. Exons:** NM_000133.3:1-8**. Variants(14)**: c.391+7A>G, c.1159A>G, c.271T>A, c.1291T>A, c.19A>T, c.793G>A, c.1061G>A, c.1001T>C, c.1298A>C, c.797C>T, c.1265C>A, c.1345C>T, c.1015A>G, c.1009G>A

HFE-Related Hereditary Hemochromatosis . Gene:*HFE*. **Exons**: NM_000410.3:1-6. **Variants(3)**: c.845G>A, c.187C>G, c.848A>C

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency. Gene:*CBS*. **Exons**: NM_000071.2:3-17. **Variants(1)**: c.1105C>T

Macular degeneration, juvenile. Gene:*CNGB3*. **Exons**: NM_019098.4:1-18. **Variants(2)**: c.1208G>A, c.1148delC

MTHFR deficiency. Gene:*MTHFR*. **Exons**: NM_005957.4:2-12. **Variants(1)**: c.665C>T

Nephropathic Cystinosis. Gene:*CTNS*. **Exons**: NM_004937.2:3-12. **Variants(1)**: c.124G>A

Oculocutaneous Albinism Type 2. Gene:*OCA2*. **Exons**: NM_000275.2:2-24. **Variants(1)**: c.1441G>A

Odontoonychodermal Dysplasia. Gene:*WNT10A*. **Exons**: NM_025216.2:1-3. **Variants(1)**: c.682T>A

Phenylketonuria. Gene:PAH**. Exons:** NM_000277.1:1-13**. Variants(24)**: c.799C>G, c.1256A>G, c.965C>G, c.1241A>G, c.175G>T, c.205C>T, c.307G>A, c.428A>G, c.839A>C, c.365C>A, c.1194A>C, c.1162G>A, c.1117G>A, c.877T>A, c.155T>C, c.512G>C, c.937G>A, c.434A>T, c.1223G>A, c.1139C>T, c.386A>G, c.281T>G, c.1208C>T, c.158G>A

Protein C deficiency. Gene:*PROC*. **Exons**: NM_000312.3:2-9. **Variants(6)**: c.629C>T, c.1335C>G, c.866C>T, c.1000G>A, c.793C>T, c.902C>T

Protein S deficiency. Gene:*PROS1*. **Exons**: NM_000313.3:1-5,8-10,12,14. **Variants(2)**: c.701A>G, c.835C>T

Pseudocholinesterase deficiency. Gene:*BCHE*. **Exons**: NM_000055.2:2-4. **Variants(1)**: c.293A>G

Smith-Lemli-Opitz syndrome. Gene:*DHCR7*. **Exons**: NM_001360.2:3-9. **Variants(1)**: c.1A>G

Stargardt Disease 1. Gene:*ABCA4*. **Exons**: NM_000350.2:1-50. **Variants(27)**: c.5693G>A, c.5912T>G, c.6320G>A, c.3113C>T, c.6088C>T, c.3322C>T, c.1018T>G, c.1225delA, c.5882G>A, c.5338C>G, c.5908C>T, c.6079C>T, c.2588G>C, c.634C>T, c.3210_3211dupGT, c.5819T>C, c.52C>T, c.3083C>T, c.3106G>A, c.4139C>T, c.1715G>A, c.3364G>A, c.571-2A>G, c.2565G>A, c.3386G>T, c.6148G>C, c.2791G>A

Tay-Sachs pseudodeficiency Disease. Gene:*HEXA*. **Exons**: NM_000520.4:1-14. **Variants(2)**: c.745C>T, c.739C>T

TFR2-Related Hereditary Hemochromatosis. Gene:*TFR2*. **Exons**: NM_003227.3:1-18. **Variants(14)**: c.1330G>A, c.750C>G, c.949C>T, c.1469T>G, c.1665delC, c.1861_1872delGCCGTGGCCCAG, c.1186C>T, c.2137-1G>A, c.88_89insC, c.515T>A, c.2069A>C, c.1235_1237delACA, c.2374G>A, c.313C>T

Usher Syndrome Type 1D. Gene:*CDH23*. **Exons**: NM_022124.5:2-70. **Variants(1)**: c.5237G>A

Venous Thromboembolism. Gene:*F2*. **Exons**: NM_000506.3:1-14. **Variants(1)**: c.*97G>A

Xeroderma Pigmentosum Group G. Gene:*ERCC5*. **Exons**: NM_000123.3:1-15. **Variants(1)**: c.2375C>T