FullGenomics

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HERES-GEN

DISEASES AND MUTATIONS LIST

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CYP11B1): (1): c.1343G>A (p.R448H)

17-Alpha-Hydroxylase Deficiency (CYP17A1): (20):c.1024C>A (p.P342T), c.1039C>T (p.R347C), c.1040G>A (p.R347H), c.1073G>A (p.R358Q),c.1084C>T (p.R362C), c.1216T>C (p.W406R), c.1226C>G (p.P409R), c.1250T>G (p.F417C),c.157_159delTTC (p.53delF), c.278T>G (p.F93C), c.286C>T (p.R96W), c.287G>A (p.R96Q),c.316T>C (p.S106P), c.340T>G (p.F114V), c.347A>T (p.D116V), c.51G>A (p.W17X), c.601T>A (p.Y201N), c.715C>T (p.R239X), c.81C>A (p.Y27X), c.985T>G (p.Y329D)

17-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD17B3): (8): c.166G>A (p.A56T), c.238C>T (p.R80W), c.239G>A (p.R80Q), c.389A>G (p.N130S), c.608C>T (p.A203V), c.695C>T (p.S232L), c.703A>G (p.M235V), c.803G>A (p.C268Y)

21-Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia (CYP21A2): (1): c.293-13C>G

21-Hydroxylase-Deficient Nonclassical Congenital Adrenal Hyperplasia (CYP21A2): (1): c.1360C>T (p.P454S)

3-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD3B2): (6): c.29C>A (p.A10E), c.424G>A (p.E142K), c.512G>A (p.W171X), c.664C>A (p.P222T), c.742_747delGTCCGAinsAACTA (p.V248NfsR249X), c.745C>T (p.R249X)

3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related (MCCC1): (2): c.1155A>C (p.R385S), c.1310T>C (p.L437P)

3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related (MCCC2): (8): c.1309A>G (p.I437V), c.295G>C (p.E99Q), c.464G>A (p.R155Q), c.499T>C (p.C167R), c.569A>G (p.H190R), c.803G>C (p.R268T), c.838G>T (p.D280Y), c.929C>G (p.P310R)

3-Methylglutaconic Aciduria: Type 3 (OPA3): (3): c.143-1G>C, c.320_337delAGCAGCGCCACAAGGAGG (p.Q108_E113del), c.415C>T (p.Q139X)

3-Phosphoglycerate Dehydrogenase Deficiency (PHGDH): (7): c.1117G>A (p.A373T), c.1129G>A (p.G377S), c.1273G>A (p.V425M), c.1468G>A (p.V490M), c.403C>T (p.R135W), c.712delG (p.G238fsX), c.781G>A (p.V261M)

5-Alpha Reductase Deficiency (SRD5A2): (10): c.164T>A (p.L55Q), c.344G>A (p.G115D), c.547G>A (p.G183S), c.586G>A (p.G196S), c.591G>T(p.E197D), c.635C>G (p.P212R), c.679C>T (p.R227X), c.682G>A (p.A228T), c.692A>G(p.H231R), c.736C>T (p.R246W)

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS): (6):c.155A>G (p.N52S), c.259C>T (p.P87S), c.286G>A (p.D96N), c.347A>G (p.D116G), c.46C>T (p.R16C), c.74G>A (p.R25Q)

ARSACS (SACS): (6): c.12973C>T (p.R4325X), c.3161T>C (p.F1054S), c.5836T>C (p.W1946R), c.7504C>T (p.R2502X), c.8844delT (p.I2949fs), c.9742T>C (p.W3248R)

Abetalipoproteinemia (MTTP): (2): c.2211delT, c.2593G>T (p.G865X)

Acrodermatitis Enteropathica (SLC39A4): (7): c.1120G>A (p.G374R), c.1223-1227delCCGGG, c.318C>A (p.N106K), c.599C>T (p.P200L), c.909G>C (p.Q303H), c.968-971delAGTC, c.989G>A (p.G330D)

Acute Infantile Liver Failure: TRMU Related (TRMU): (5): c.1102-3C>G, c.229T>C (p.Y77H), c.2T>A (p.M1K), c.815G>A (p.G272D), c.835G>A (p.V279M)

Acyl-CoA Oxidase I Deficiency (ACOX1): (5): c.372delCATGCCCGCCTGGAACTT, c.442C>T (p.R148X), c.532G>T (p.G178C), c.832A>G(p.M278V), c.926A>G (p.Q309R)

Adenosine Deaminase Deficiency (ADA): (22): c.220G>T(p.G74C), c.248C>A (p.A83D), c.301C>T (p.R101W), c.302G>A (p.R101Q), c.302G>T (p.R101L), c.320T>C (p.L107P), c.385G>A (p.V129M), c.419G>A (p.G140E), c.43C>G (p.H15D), c.445C>T (p.R149W), c.454C>A (p.L152M), c.466C>T (p.R156C), c.467G>A (p.R156H), c.529G>A (p.V177M), c.536C>A (p.A179D), c.58G>A (p.G20R), c.596A>C (p.Q199P), c.631C>T (p.R211C), c.632G>A (p.R211H), c.646G>A (p.G216R), c.872C>T(p.S291L), c.986C>T (p.A329V)

Adrenoleukodystrophy: X-Linked (ABCD1): (21): c.1165C>G (p.R389G), c.1202G>A (p.R401Q), c.1252C>T (p.R418W), c.1390C>T (p.R464X), c.1414_1415insC (p.Q472fs), c.1415_1416delAG, c.1429G>T (p.E477X), c.1451C>G (p.P484R),c.1534G>T (p.G512C), c.1544C>T (p.S515F), c.1552delC, c.311G>A (p.R104H), c.421G>A(p.A141T), c.443A>G (p.N148S), c.520T>G (p.Y174D), c.521A>G (p.Y174C), c.686T>C(p.L229P), c.796G>C (p.G266R), c.871G>A (p.E291K), c.871_873delGAG (p.291delE), c.901-1G>A (IVS1-1G>A)

Alkaptonuria (HGD): (14): c.1102A>G (p.M368V),c.1111_1112insC, c.1112A>G (p.H371R), c.140C>T (p.S47L), c.16-1G>A (IVS1-1G>A), c.174delA,c.342+1G>A (IVS5+1G>A), c.360T>G (p.C120W), c.457_458insG, c.481G>A (p.G161R), c.688C>T (p.P230S), c.808G>A (p.G270R), c.899T>G (p.V300G), c.990G>T (p.R330S)

Alpha Thalassemia (HBA2,HBA1): (8): SEA deletion, c.*+94A>G, c.207C>A (p.N69K), c.207C>G (p.N69K), c.223G>C (p.D75H), c.2T>C,c.377T>C (p.L126P), c.427T>C (p.X143Qext32)

Alpha-1-Antitrypsin Deficiency (SERPINA1): (4): c.1096G>A (p.E366K), c.1131A>T (p.L377F), c.187C>T (p.R63C), c.226_228delTTC (p.76delF)

Alpha-Mannosidosis (MAN2B1): (3): c.1830+1G>C (p.V549_E610del), c.2248C>T (p.R750W), c.2426T>C (p.L809P)

Alport Syndrome: COL4A3 Related (COL4A3): (3):c.4420_4424delCTTTT, c.4441C>T (p.R1481X), c.4571C>G (p.S1524X)

Alport Syndrome: COL4A4 Related (COL4A4): (4): c.3601G>A (p.G1201S), c.3713C>G (p.S1238X), c.4129C>T (p.R1377X), c.4923C>A (p.C1641X)

Alport Syndrome: X-linked (COL4A5): (3): c.4691G>C (p.C1564S), c.4946T>G (p.L1649R), c.5030G>A (p.R1677Q)

Amegakaryocytic Thrombocytopenia (MPL): (23): c.127C>T(p.R43X), c.1305G>C (p.W435C), c.1473G>A (p.W491X), c.1499delT (p.L500fs), c.1566-1G>T (IVS10-1G>T), c.1781T>G (p.L594W), c.1904C>T (p.P635L), c.213-1G>A (IVS2-1G>A), c.235_236delCT (p.L79fs), c.268C>T (p.R90X), c.304C>T (p.R102C), c.305G>C (p.R102P),c.311T>C (p.F104S), c.367C>T (p.R123X), c.376delT (F126Lfs), c.407C>A (p.P136H), c.407C>T(p.P136L), c.460T>C (p.W154R), c.556C>T (p.Q186X), c.769C>T (p.R257C), c.770G>T(p.R257L), c.79+2T>A (IVS1+2T>A), c.823C>A (p.P275T)

Andermann Syndrome (SLC12A6): (5): c.2023C>T (p.R675X), c.2436delG (p.T813fsX813), c.3031C>T (p.R1011X), c.619C>T (p.R207C), c.901delA

Androgen Insensitivity Syndrome: Complete (AR): (15): c.1739G>T (p.C580F), c.1748T>A (p.F583Y), c.1771A>T (p.K591X), c.178C>T (p.Q60X),c.2033T>C (p.L678P), c.2123T>G (p.L708R), c.2157G>A (p.W719X), c.2231G>A (p.G744E),c.2323C>T (p.R775C), c.2324G>A (p.R775H), c.2343G>T (p.M781I), c.2362A>G (p.M788V),c.2391G>A (p.W797X), c.2599G>A (p.V867M), c.2650A>T (p.K884X)

Antley-Bixler Syndrome (POR): (4): c.1370G>A (p.R457H), c.1475T>A (p.V492E), c.1615G>A (p.G539R), c.859G>C (p.A287P)

Argininemia (ARG1): (13): c.263_266delAGAA (p.K88fs), c.32T>C (p.I11T), c.365G>A (p.W122X), c.413G>T (p.G138V), c.466-2A>G, c.57+1G>A, c.61C>T (p.R21X), c.703G>A (p.G235R), c.703G>C (p.G235R), c.77delA (p.E26fs), c.844delC (p.L282fs), c.869C>G (p.T290S), c.871C>T (p.R291X)

Argininosuccinate Lyase Deficiency (ASL): (7): c.1060C>T (p.Q354X), c.1135C>T (p.R379C), c.1153C>T (p.R385C), c.283C>T (p.R95C), c.446+1G>A (IVS5+1G>A), c.532G>A (p.V178M), c.857A>G (p.Q286R)

Aromatase Deficiency (CYP19A1): (10): c.1094G>A (p.R365Q), c.1123C>T (p.R375C), c.1224delC (p.K409fs), c.1303C>T (p.R435C), c.1310G>A (p.C437Y), c.296+1G>A (IVS3+1G>A), c.468delC, c.628G>A (p.E210K), c.629-3C>A (IVS4-3C>A), c.743+2T>C (IVS6+2T>C)

Arthrogryposis, Mental Retardation, & Seizures (SLC35A3): (2): c.1012A>G (p.S338G), c.514C>T (p.Q172X)

Arts Syndrome (PRPS1): (2): c.398A>C (p.Q133P), c.455T>C (p.L152P)

Asparagine Synthetase Deficiency (ASNS): (1): c.1084T>G (p.F362V)
Aspartylglycosaminuria (AGA): (7): c.179G>A (p.G60D), c.200_201delAG, c.214T>C (p.S72P), c.302C>T (p.A101V), c.488G>C (p.C163S), c.904G>A(p.G302R), c.916T>C (p.C306R)

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Ataxia with Vitamin E Deficiency (TTPA): (14): c.175C>T,(p.R59W), c.205-1G>C, c.219_220insAT, c.303T>G (p.H101Q), c.306A>G (p.G102G), c.358G>A (p.A120T), c.400C>T (p.R134X), c.421G>A (p.E141K), c.486delT (p.W163Gfs),c.513_514insTT (p.T172fs), c.575G>A (p.R192H), c.661C>T (p.R221W), c.736G>C (p.G246R), c.744delA

Ataxia-Telangiectasia (ATM): (20): c.103C>T p.R35X),c.1564_1565delGA (p.E522fs), c.3245delATCinsTGAT (p.H1082fs), c.3576G>A (p.K1192K), c.3894insT, c.5712_5713insA (p.S1905fs), c.5762+1126A>G, c.5908C>T (p.Q1970X), c.5932G>T (p.E1978X), c.7268A>G (p.E2423G), c.7271T>G (p.V2424G), c.7327C>T(p.R2443X), c.7449G>A (p.W2483X), c.7517_7520delGAGA (p.R2506fs), c.7630-2A>C, c.7638_7646delTAGAATTTC (p.R2547_S2549delRIS), c.7876G>C (p.A2626P), c.7967T>C (p.L2656P), c.8030A>G (p.Y2677C), c.8480T>G (p.F2827C)

Autosomal Recessive Polycystic Kidney Disease (PKHD1): (39): c.10174C>T (p.Q3392X), c.10364delC (p.S3455fs), c.10402A>G (p.I3468V),c.10412T>G (p.V3471G), c.10505A>T (p.E3502V), c.10637delT (p.V3546fs), c.10658T>C (p.I3553T), c.107C>T (p.T36M), c.10856delA (p.K3619fs), c.10865G>A (p.C3622Y), c.11612G>A (p.W3871X), c.1486C>T (p.R496X), c.1529delG (p.G510fs), c.2269A>C (p.I757L), c.2414C>T (p.P805L), c.3229-2A>C (IVS28-2A>C), c.3747T>G (p.C1249W), c.3761_3762delCCinsG (p.A1254fs), c.383delC, c.4165C>A (p.P1389T), c.4220T>G (p.L1407R), c.4991C>T (p.S1664F), c.50C>T (p.A17V), c.5221G>A (p.V1741M), c.5381-9T>G (IVS33-9T>G), c.5513A>G (p.Y1838C), c.5750A>G (p.Q1917R), c.5895insA (p.L1966fsX1969), c.5984A>G (p.E1995G), c.657C>T (p.G219G), c.664A>G (p.I222V), c.6992T>A (p.I2331K), c.7350+653A>G (IVS46+653A>G), c.8011C>T (p.R2671X), c.8063G>T (p.C2688F), c.8870T>C (p.I2957T), c.9053C>T (p.S3018F), c.9530T>C (p.I3177T), c.9689delA (p.D3230fs)

Bardet-Biedl Syndrome: BBS1 Related (BBS1): (3): c.1169T>G (p.M390R), c.1645G>T (p.E549X), c.851delA

Bardet-Biedl Syndrome: BBS10 Related (BBS10): (3): c.101G>C (p.R34P), c.271_273ins1bp (p.C91fsX95), c.931T>G (p.S311A)

Bardet-Biedl Syndrome: BBS11 Related (TRIM32): (1): c.388C>T (p.P130S)

Bardet-Biedl Syndrome: BBS12 Related (BBS12): (5): c.1063C>T (p.R355X), c.1114_1115delTT (p.F372X), c.1483_1484delGA (p.E495fsX498), c.335_337delTAG, c.865G>C (p.A289P)

Bardet-Biedl Syndrome: BBS2 Related (BBS2): (8): c.1206_1207insA (p.R403fs), c.1895G>C (p.R632P), c.224T>G (p.V75G), c.311A>C (p.D104A), c.72C>G (p.Y24X), c.814C>T (p.R272X), c.823C>T (p.R275X), c.940delA

Bare Lymphocyte Syndrome: Type II (CIITA):(3): c.1141G>T(p.E381X), c.2888+1G>A (IVS13+1G>A), c.3317+1G>A (IVS18+1G>A)

Bartter Syndrome: Type 4A (BSND): (6): c.139G>A (p.G47R), c.1A>T, c.22C>T (p.R8W), c.23G>T (p.R8L), c.28G>A (p.G10S), c.3G>A (p.M1I)

Beta Thalassemia (HBB): (81): c.-136C>G, c.-137c>g, c.-137c>t, c.-138c>t, c.-140c>t, c.-142C>T, c.-151C>T, c.-29G>A, c.-50A>C, c.-78a>g, c.-79A>G, c.-80t>a, c.-81A>G, c.112delT, c.113G>A (p.W38X), c.114G>A (p.W38X), c.118C>T (p.Q40X), c.124_127delTTCT (p.F42Lfs), c.126delC, c.135delC (p.F46fs), c.154delC (p.P52fs), c.169G>C(p.G57R), c.17_18delCT, c.1A>G, c.203_204delTG (p.V68Afs), c.20delA (p.E7Gfs), c.217_218insA (p.S73Kfs), c.223+702_444+342del620insAAGTAGA, c.225delC, c.230delC,c.250delG, c.25_26delAA, c.271G>T (p.E91X), c.287_288insA (p.L97fs), c.295G>A (p.V99M),c.2T>C, c.2T>G, c.315+1G>A, c.315+2T>C, c.315+745C>G, c.316-146T>G, c.316-197C>T, c.316-1G>A, c.316-1G>C, c.316-1G>T, c.316-2A>C, c.316-2A>G, c.316-3C>A, c.316-3C>G,c.321_322insG (p.N109fs), c.36delT (p.T13fs), c.383_385delAGG (p.Q128_A129delQAinsP),c.415G>C (p.A139P), c.444+111A>G, c.444+113A>G, c.45_46insG (p.W16fs), c.46delT (p.W16Gfs), c.47G>A (p.W16X), c.48G>A (p.W16X), c.4delG (p.V2Cfs), c.51delC (p.K18Rfs), c.52A>T (p.K18X), c.59A>G (p.N20S), c.68_74delAAGTTGG, c.75T>A (p.G25G), c.84_85insC (p.L29fs), c.90C>T (p.G30G), c.92+1G>A, c.92+1G>T, c.92+2T>A, c.92+2T>C, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.92G>C (p.R31T), c.93-15T>G, c.93-1G>A, c.93-1G>C,c.93-1G>T, c.93-21G>A

Beta-Hexosaminidase Pseudodeficiency (HEXA): (2): c.739C>T (p.R247W), c.745C>T (p.R249W)

Beta-Ketothiolase Deficiency (ACAT1): (19): c.1006-1G>C,c.1006-2A>C, c.1083insA, c.1136G>T (p.G379V), c.1138G>A (p.A380T), c.149delC(p.T50Nfs), c.253_255delGAA (p.85delE), c.278A>G (p.N93S), c.2T>A (p.M1K), c.371A>G(p.K124R), c.380C>T (p.A127V), c.433C>G (p.Q145E), c.455G>C (p.G152A), c.547G>A(p.G183R), c.814C>T (p.Q272X), c.826+1G>T, c.935T>C (p.I312T), c.997G>C (p.A333P),c.99T>A (p.Y33X)

Biotinidase Deficiency (BTD): (21): c.100G>A (p.G34S),c.1049delC (p.A350fs), c.1052delC (p.T351fs), c.1207T>G (p.F403V), c.1239delC (p.Y414Ifs), c.1240_1251delTATCTCCACGTC (p.Y414_V417del), c.1330G>C (p.D444H), c.1368A>C(p.Q456H), c.1489C>T (p.P497S), c.1595C>T (p.T532M), c.1612C>T (p.R538C), c.235C>T (p.R79C), c.278A>G (p.Y93C), c.341G>T (p.G114V), c.393delC (p.F131Lfs), c.470G>A (p.R157H), c.511G>A (p.A171T), c.595G>A (p.V199M), c.755A>G (p.D252G), c.933delT (p.S311Rfs), c.98_104delGCGGCTGinsTCC (p.C33FfsX68)

Bloom Syndrome (BLM): (25): c.1284G>A (p.W428X), c.1642C>T (p.Q548X), c.1701G>A (p.W567X), c.1933C>T (p.Q645X), c.2074+2T>A, c.2193+1_2193+9del9, c.2207_2212delATCTGAinsTAGATTC (p.Y736Lfs), c.2343_2344dupGA (p.781EfsX), c.2407insT, c.2528C>T (p.T843I), c.2695C>T (p.R899X), c.2923delC (p.Q975K), c.3107G>T (p.C1036F), c.3143delA (p.1048NfsX), c.318_319insT (p.L107fs), c.3281C>A (p.S1094X), c.3558+1G>T, c.3564delC (p.1188Dfs), c.356_357delTA (p.C120Hfs), c.380delC (p.127Tfs), c.3875-2A>G, c.4008delG (p.1336Rfs), c.4076+1delG, c.557_559delCAA (p.S186X), c.947C>G (p.S316X)

Canavan Disease (ASPA): (8): c.2T>C (p.M1T), c.433-2A>G, c.654C>A (p.C218X), c.693C>A (p.Y231X), c.71A>G (p.E24G), c.79G>A (p.G27R), c.854A>C (p.E285A), c.914C>A (p.A305E)

Carnitine Palmitoyltransferase IA Deficiency (CPT1A): (10): c.1079A>G (p.E360G), c.1241C>T (p.A414V), c.1339C>T (p.R447X), c.1361A>G (p.D454G), c.1436C>T (p.P479L), c.1493A>G (p.Y498C), c.2126G>A (p.G709E), c.2129G>A (p.G710E), c.2156G>A (p.G719D), c.96T>G (p.Y32X)

Carnitine Palmitoyltransferase II Deficiency (CPT2): (19): c.109_110insGC, c.1148T>A (p.F383Y), c.1238_1239delAG, c.149C>A (p.P50H), c.1646G>A (p.G549D), c.1649A>G (p.Q550R), c.1737delC, c.1810C>T (p.P604S), c.1883A>C (p.Y628S),c.1891C>T (p.R631C), c.1923_1935delGAAGGCCTTAGAA, c.338C>T (p.S113L), c.359A>G (p.Y120C), c.370C>T (p.R124X), c.452G>A (p.R151Q), c.520G>A (p.E174K),c.534_558delGAACCCTGCAAAAAGTGACACTATCinsT, c.680C>T (p.P227L), c.983A>G (p.D328G)

Carnitine-Acylcarnitine Translocase Deficiency (SLC25A20): (7): c.106-2A>T, c.199-10T>G (IVS2-10T>G), c.496C>T (p.R166X), c.576G>A (p.W192X), c.713A>G (p.Q238R), c.84delT (p.H29Tfs), c.897_898insC (p.N300fs)

Carpenter Syndrome (RAB23): (2): c.408_409insT (p.136fsX), c.434T>A (p.L145X)

Cartilage-Hair Hypoplasia (RMRP): (2): c.263G>T, n.71A>G

Cerebrotendinous Xanthomatosis (CYP27A1): (14): c.1016C>T (p.T339M), c.1183C>A (p.R395S), c.1183C>T (p.R395C), c.1214G>A (p.R405Q),c.1263+1G>A, c.1420C>T (p.R474W), c.1421G>A (p.R474Q), c.1435C>T (p.R479C), c.379C>T (p.R127W), c.434G>A (p.G145E), c.583G>T (p.E195X), c.646G>C (p.A216P), c.819delT (p.D273fs), c.844+1G>A

Charcot-Marie-Tooth Disease with Deafness: X-Linked: GJB1 Related (GJB1): (22): c.123G>C (p.E41D), c.145T>C (p.S49P), c.164C>T (p.T55I), c.187G>A (p.V63I), c.194A>G (p.Y65C), c.223C>T (p.R75W), c.225delG (p.R75fs), c.254C>G (p.S85C), c.283G>A (p.V95M), c.304G>T (p.E102X), c.304delGAG (p.102delE), c.37G>T (p.V13L), c.397T>C (p.W133R), c.407T>C (p.V136A), c.415G>A (p.V139M), c.424C>T (p.R142W), c.43C>T (p.R15W), c.467T>G (p.L156R), c.514C>T (p.P172S), c.614A>G(p.N205S), c.658C>T (p.R220X), c.89T>A (p.I30N)

Charcot-Marie-Tooth Disease with Deafness: X-Linked: PRPS1 Related (PRPS1): (2): c.129A>C (p.E43D), c.344T>C (p.M115T)

Chediak-Higashi Syndrome (LYST): (4): c.118_119insG (p.A40fs), c.1902_1903insA (p.A635Sfs), c.3085C>T (p.Q1029X), c.9590delA (p.Y3197fs)

Cholesteryl Ester Storage Disease (LIPA): (4): c.1024G>A (p.G342R), c.652C>T (p.R218X), c.883C>T (p.H295Y), c.894G>A (p.Q298X)

Choreoacanthocytosis (VPS13A): (1): c.6058delC (p.P2020fs)

Choroideremia (CHM): (1): c.1609+2insT

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DISEASES AND MUTATIONS LIST

Chronic Granulomatous Disease: CYBA Related (CYBA): (12): c.171_172insG (p.K58fs), c.174delG (p.K58fs), c.244delC (p.P82fs), c.281A>G (p.H94R), c.354C>A (p.S118R), c.369+1G>A (IVS5+1G>A), c.373G>A (p.A125T), c.385_388delGAGC (p.E129SfsX61), c.467C>A (p.P156Q), c.70G>A (p.G24R), c.71G>A (p.G24E), c.7C>T (p.Q3X)

Chronic Granulomatous Disease: X-Linked (CYBB): (14): c.1166G>C (p.G389A), c.1244C>A (p.P415H), c.1499A>G (p.D500G), c.217C>T (p.R73X),c.252G>A (p.A84A), c.301C>T (p.H101Y), c.302A>G (p.H101R), c.337+5G>A (IVS3+5G>A),c.45+6T>C (IVS1+6T>C), c.466G>A (p.A156T), c.625C>T (p.H209Y), c.676C>T (p.R226X), c.907C>A (p.H303N), c.911C>G (p.P304R)

Citrin Deficiency (SLC25A13): (8): c.1180+1G>A, c.1180G>A (p.G394S), c.1314+1G>A, c.1663_1664insGAGATTACAGGTGGCTGCCCGGG (p.A555fs), c.1766G>A (p.R589Q), c.1802_1803insA (p.Y601fs), c.674C>A (p.S225X), c.851_854delGTAT (p.R284fs)

Citrullinemia: Type I (ASS1): (11): c.1085G>T (p.G362V), c.1168G>A (p.G390R), c.1194-1G>C, c.421-2A>G (IVS6-2A>G), c.470G>A (p.R157H), c.535T>C (p.W179R), c.539G>A (p.S180N), c.835C>T (p.R279X), c.928A>C (p.K310Q), c.970+5G>A, c.970G>A (p.G324S)

Classical Galactosemia (GALT): (18): c.-1039_753del3162,c.1138T>C (p.X380R), c.134_138delCAGCT, c.221T>C (p.L74P), c.253-2A>G, c.404C>G (p.S135W), c.404C>T (p.S135L), c.413C>T (p.T138M), c.425T>A (p.M142K), c.505C>A (p.Q169K), c.512T>C (p.F171S), c.563A>G (p.Q188R), c.584T>C (p.L195P), c.607G>A (p.E203K), c.626A>G (p.Y209C), c.820+51_*789del2294ins12, c.855G>T (p.K285N),c.997C>G (p.R333G)

Cockayne Syndrome: Type A (ERCC8): (3): c.37G>T (p.E13X), c.479C>T (p.A160V), c.966C>A (p.Y322X)

Cockayne Syndrome: Type B (ERCC6): (7): c.1034_1035insT (p.K345fs), c.1357C>T (p.R453X), c.1518delG (p.K506Nfs), c.1550G>A (p.W517X), c.1974_1975insTGTC (p.T659fs), c.2203C>T (p.R735X), c.972_973insA (p.E325Rfs)

Cohen Syndrome (VPS13B): (9): c.10888C>T (p.Q3630X), c.2911C>T (p.R971X), c.3348_3349delCT (p.C1117fx), c.4471G>T (p.E1491X), c.6578T>G (p.L2193R), c.7051C>T (p.R2351X), c.7934G>A (p.G2645D), c.8459T>C (p.I2820T), c.9259_9260insT (p.L3087fs)

Combined Pituitary Hormone Deficiency: PROP1 Related (PROP1): (11): c.109+1G>T, c.112_124delTCGAGTGCTCCAC (p.S38fsX), c.150delA (p.G50fsX), c.157delA (p.R53fsX), c.212G>A (p.R71H), c.217C>T (p.R73C), c.218G>A (p.R73H), c.2T>C, c.301delAG (p.S101fsX), c.358C>T (p.R120C), c.582G>A (p.W194X)

Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2): (5): c.338C>T (p.P113L), c.357C>A (p.F119L), c.422G>A (p.R141H), c.470T>C (p.F157S), c.691G>A (p.V231M)

Congenital Disorder of Glycosylation: Type 1B: MPI Related (MPI): (1): c.884G>A (p.R295H)

Congenital Disorder of Glycosylation: Type 1C: ALG6 Related (ALG6): (4): c.1432T>C (p.S478P), c.257+5G>A, c.895_897delATA, c.998C>T (p.A333V)

Congenital Ichthyosis: ABCA12 Related (ABCA12) (8): c.3535G>A (p.G1179R), c.4139A>G (p.N1380S), c.4142G>A (p.G1381E), c.4541G>A (p.R1514H), c.4615G>A (p.E1539K), c.4951G>A (p.G1651S), c.6610C>T (p.R2204X), c.7323delC (p.V2442Sfs)

Congenital Insensitivity to Pain with Anhidrosis (NTRK1): (12): c.1076A>G (p.Y359C), c.1550G>A (p.G517E), c.1660delC (p.R554fs), c.1729G>C (p.G577R), c.1759A>G (p.M587V), c.2046+3A>C, c.207_208delTG (p.E70Afs), c.2084C>T (p.P695L), c.2339G>C (p.R780P), c.25C>T (p.Q9X), c.429-1G>C, c.717+4A>T

Congenital Lipoid Adrenal Hyperplasia (STAR): (12): c.178+1_178+2insT (IVS2+3insT), c.201_202delCT, c.466-11T>A (IVS4-11T>A), c.545G>A (p.R182H), c.545G>T (p.R182L), c.559G>A (p.V187M), c.562C>T (p.R188C), c.64+1G>A, c.64+1G>T (IVS1+1G>T), c.650G>C (p.R217T), c.749G>A (p.W250X), c.772C>T (p.Q258X)

Congenital Myasthenic Syndrome: CHRNE Related (CHRNE): (12): c.1327delG (p.E443fs), c.1353_1354insG (p.N452Efs), c.250C>G (p.R84G), c.344+1G>A, c.37G>A (p.G13R), c.422C>T (p.P141L), c.500G>T (p.R167L), c.613_619delTGGGCCA (p.W205fs), c.850A>C (p.T284P), c.865C>T (p.L289F), c.911delT(p.L304fs), c.991C>T (p.R331W)

Congenital Myasthenic Syndrome: DOK7 Related (DOK7): (6): c.101-1G>T, c.1263_1264insC (p.S422fs), c.331+1G>T, c.539G>C (p.G180A), c.548_551delTCCT (p.F183fs), c.601C>T (p.R201X)

Congenital Myasthenic Syndrome: RAPSN Related (RAPSN): (11): c.-210A>G, c.133G>A (p.V45M), c.193-15C>A (IVS1-15C>A), c.264C>A (p.N88K), c.41T>C (p.L14P), c.46_47insC (p.L16fs), c.484G>A (p.E162K), c.490C>T (p.R164C), c.548_549insGTTCT (p.L183fs), c.807C>A (p.Y269X), c.848T>C (p.L283P)

Congenital Neutropenia: Recessive (HAX1): (6): c.121_125insG, c.130_131insA, c.256C>T (p.R86X), c.423_424insG, c.568C>T (p.Q190X), c.91delG

Copper Transport Disorders (ATP7A): (10): c.1707+1G>A (IVS6+1G>A), c.1707+1_1707+4delgtaa, c.1910C>T (p.S637L), c.1947-1_1947insATAAG, c.2938C>T (p.R980X), c.3056G>A (p.G1019D), c.3911A>G (p.N1304S), c.408delCAATCAGA (p.A136fs), c.4350delG (p.L1451fs), c.601C>T (p.R201X)

Corneal Dystrophy and Perceptive Deafness (SLC4A11): (8): c.1459_1462delTACGinsA (p.487_488delYAinsT), c.1463G>A (p.R488K), c.2313_2314insTATGACAC, c.2321+1G>A, c.2528T>C (p.L843P), c.2566A>G (p.M856V), c.554_561delGCTTCGCC (p.R185fs), c.637T>C (p.S213P)

Corticosterone Methyloxidase Deficiency (CYP11B2): (3): c.1382T>C (p.L461P), c.1492A>G (p.T498A), c.541C>T (p.R181W)

Crigler-Najjar Syndrome (UGT1A1): (11): c.1021C>T(p.R341X), c.1070A>G (p.Q357R), c.1124C>T (p.S375F), c.1198A>G (p.N400D), c.44T>G(p.L15R), c.508_513delTTC (p.170delF), c.524T>A (p.L175Q), c.840C>A (p.C280X), c.923G>A (p.G308E), c.991C>T (p.Q331X), c.992A>G (p.Q331R)

Cystic Fibrosis (CFTR): (149): c.1000C>T (p.R334W), c.1013C>T(p.T338I), c.1029delC, c.1040G>A (p.R347H), c.1040G>C (p.R347P), c.1055G>A (p.R352Q),c.1075C>A (p.Q359K), c.1079C>A (p.T360K), c.1090T>C (p.S364P), c.1116+1G>A, c.1153_1154insAT, c.1175T>G (p.V392G), c.11C>A (p.S4X), c.1364C>A (p.A455E), c.1408_1417delGTGATTATGG (p.V470fs), c.1438G>T (p.G480C), c.1477C>T (p.Q493X),c.1477delCA, c.14C>T (p.P5L), c.1519_1521delATC (p.507delI), c.1521_1523delCTT (p.508delF), c.1526delG (p.G509fs), c.1545_1546delTA (p.Y515Xfs), c.1558G>T (p.V520F),c.1572C>A (p.C524X), c.1585-1G>A, c.1585-8G>A, c.1610_1611delAC (p.D537fs), c.1624G>T (p.G542X), c.164+12T>C, c.1645A>C (p.S549R), c.1646G>A (p.S549N), c.1646G>T (p.S549I), c.1647T>G (p.S549R), c.1652G>A (p.G551D), c.1654C>T (p.Q552X), c.1657C>T (p.R553X), c.1675G>A (p.A559T), c.1679G>C (p.R560T), c.1680-1G>A, c.1680-886A>G, c.171G>A (p.W57X), c.1721C>A (p.P574H), c.1766+1G>A, c.1766+1G>T, c.1766+5G>T, c.178G>T (p.E60X), c.1818del84, c.1911delG, c.1923delCTCAAAACTinsA, c.1973delGAAATTCAATCCTinsAGAAA, c.1976delA (p.N659fs), c.1986_1989delAACT (p.T663R), c.19G>T (p.E7X), c.200C>T (p.P67L), c.2051_2052delAAinsG (p.K684SfsX38), c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2089_2090insA (p.R697Kfs), c.2125C>T (p.R709X), c.2128A>T (p.K710X), c.2174insA, c.2215delG (p.V739Y), c.223C>T (p.R75X), c.2290C>T (p.R764X), c.2538G>A (p.W846X), c.254G>A (p.G85E), c.261delTT, c.263T>G (p.L196X), c.2657+5G>A, c.2668C>T (p.Q890X), c.271G>A (p.G91R), c.273+1G>A,c.273+3A>C, c.2737_2738insG (p.Y913X), c.274-1G>A, c.274G>T (p.E92X),c.2908+1085_3367+260del7201, c.2909G>A (p.G970D), c.293A>G (p.Q98R), c.2988+1G>A, c.3022delG (p.V1008S), c.3039delC, c.3067_3072delATAGTG (p.I1023_V1024delT), c.3139_3139+1delGG, c.313delA (p.I105fs), c.3140-26A>G, c.3196C>T(p.R1066C), c.3209G>A (p.R1070Q), c.3254A>G (p.H1085R), c.325delTATinsG, c.3266G>A(p.W1089X), c.3276C>G (p.Y1092X), c.328G>C (p.D110H), c.3302T>A (p.M1101K), c.3368-2A>G, c.3454G>C (p.D1152H), c.3472C>T (p.R1158X), c.3484C>T (p.R1162X), c.349C>T(p.R117C), c.350G>A (p.R117H), c.3527delC, c.3535delACCA, c.3536_3539delCCAA(p.T1179fs), c.3587C>G (p.S1196X), c.3611G>A (p.W1204X), c.3659delC (p.T1220fs), c.366T>A (p.Y122X), c.3691delT, c.3700A>G (p.I1234V), c.3712C>T (p.Q1238X), c.3717+12191C>T, c.3717+4A>G (IVS22+4A>G), c.3731G>A (p.G1244E), c.3744delA,c.3752G>A (p.S1251N), c.3764C>A (p.S1255X), c.3767_3768insC (p.A1256fs),c.3773_3774insT (p.L1258fs), c.3846G>A (p.W1282X), c.3848G>T (p.R1283M),c.3878_3881delTATT (p.V1293fs), c.3908dupA (p.N1303Kfs), c.3909C>G (p.N1303K),c.4003C>T (p.L1335F), c.416A>T (p.H139L), c.4364C>G (p.S1455X), c.4426C>T (p.Q1476X), c.442delA, c.455T>G (p.M152R), c.489+1G>T, c.496A>G (p.K166E), c.531delT, c.532G>A(p.G178R), c.535C>A (p.Q179K), c.54-5940_273+10250del21080bp (p.S18fs), c.579+1G>T,c.579+5G>A (IVS4+5G>A), c.580-1G>T, c.613C>T (p.P205S), c.617T>G (p.L206W), c.653T>A (p.L218X), c.658C>T (p.Q220X), c.803delA (p.N268fs), c.805_806delAT (p.I269fs), c.868C>T(p.Q290X), c.933_935delCTT (p.311delF), c.946delT, c.988G>T (p.G330X)

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DISEASES AND MUTATIONS LIST

Cystinosis (CTNS): (14): c.-39155_848del57119, c.1015G>A (p.G339R), c.18_21delGACT, c.198_218delTATTACTATCCTTGAGCTCCC, c.199_219delATTACTATCCTTGAGCTCCCC (p.I67_P73del), c.283G>T (p.G95X), c.329G>T (p.G110V), c.414G>A (p.W138X), c.416C>T (p.S139F), c.473T>C (p.L158P), c.506G>A (p.G169D), c.589G>A (p.G197R), c.613G>A (p.D205N), c.969C>G (p.N323K)

Cystinuria: Non-Type I (SLC7A9): (15): c.131T>C (p.I44T),c.1445C>T (p.P482L), c.313G>A (p.G105R), c.368C>T (p.T123M), c.368_369delCG (p.T123fs),c.508G>A (p.V170M), c.544G>A (p.A182T), c.583G>A (p.G195R), c.604+2T>C, c.605-3C>A (IVS5-3C>A), c.614_615insA (p.K205fs), c.695A>G (p.Y232C), c.775G>A (p.G259R),c.782C>T (p.P261L), c.997C>T (p.R333W)

Cystinuria: Type I (SLC3A1): (10): c.1085G>A (p.R362H), c.1400T>C (p.M467T), c.1597T>A (p.Y533N), c.1843C>A (p.P615T), c.1955C>G (p.T652R), c.2033T>C (p.L678P), c.452A>G (p.Y151C), c.542G>A (p.R181Q), c.647C>T (p.T216M), c.808C>T (p.R270X)

D-Bifunctional Protein Deficiency (HSD17B4): (6): c.1369A>G (p.N457D), c.1369A>T (p.N457Y), c.422_423delAG, c.46G>A (p.G16S), c.63G>T (p.L21F), c.652G>T (p.V218L)

DMD-Related Muscular Dystrophies (DMD): (1): Deletion/Duplication Analysis

Diabetes: Recessive Permanent Neonatal (ABCC8): (2): c.1144G>A (p.E382K), c.215A>G (p.N72S)

Du Pan Syndrome (GDF5): (4): c.1133G>A (p.R378Q), c.1306C>A (p.P436T), c.1309delTTG, c.1322T>C (p.L441P)

Dyskeratosis Congenita: RTEL1 Related (RTEL1): (5): c.1548G>T (p.M516I), c.2216G>T (p.G763V), c.2869C>T (p.R981W), c.2920C>T (p.R974X), c.3791G>A (p.R1264H)

Dystrophic Epidermolysis Bullosa: Recessive (COL7A1): (11): c.8441-14_8435delGCTCTTGGCTCCAGGACCCCT, c.2470_2471insG, c.4039G>C (p.G1347R), c.425A>G (p.K142R), c.4783-1G>A, c.497_498insA (p.V168GfsX179), c.4991G>C (p.G1664A), c.5820G>A (p.P1940P), c.7344G>A (p.V2448X), c.8393T>A (p.M2798K), c.933C>A (p.Y311X)

Ehlers-Danlos Syndrome: Type VIIC (ADAMTS2): (2): c.2384G>A (p.W795X), c.673C>T (p.Q225X)

Ellis-van Creveld Syndrome: EVC Related (EVC): (10): c.1858_1879delTTGGGCCGACTGGGCGGCCTC (p.L620_L626del), c.1018C>T (p.R340X), c.1098+1G>A, c.1694delC (p.A565VfsX23), c.1868T>C (p.L623Q), c.1886+5G>T, c.2635C>T(p.Q879X), c.734delT (p.L245fs), c.910-911insA (p.R304fs), c.919T>C (p.S307P)

Ellis-van Creveld Syndrome: EVC2 Related (EVC,EVC2): (3): c. 1858_1879delTTGGGCCGACTGGGCGGCCTC (p.L620_L626del), c.1868T>C (p.L623Q), c.3025C>T (p.Q1009X)

Emery-Dreifuss Myopathy: X-Linked (EMD): (3): c.130C>T (p.Q44X), c.1A>G, c.547C>A (p.P183T)

Enhanced S-Cone (NR2E3): (5): c.119-2A>C, c.226C>T (p.R76W), c.227G>A (p.R76Q), c.747+1G>C (IVS5+1G>C), c.932G>A (p.R311Q)

Ethylmalonic Aciduria (ETHE1): (4): c.3G>T (p.M1I), c.487C>T (p.R163W), c.488G>A (p.R163Q), c.505+1G>T

Fabry's Disease (GLA): (22): c.101A>G (p.N34S), c.1025G>A (p.R342Q), c.1081G>C (p.G361R), c.166T>G (p.C56G), c.194G>C (p.S65T), c.427G>C (p.A143P), c.436C>T (p.P146S), c.466G>A (p.A156T), c.484T>C (p.W162R), c.606T>G (p.C202W), c.644A>G (p.N215S), c.679C>T (p.R227X), c.680G>A (p.R227Q), c.791A>T (p.D264V), c.797A>T (p.D266V), c.806T>C (p.V269A), c.815A>G (p.N272S), c.888G>A (p.M296I), c.890C>T (p.S297F), c.979C>A (p.Q327K), c.982G>A (p.G328R), c.983G>C (p.G328A)

Factor IX Deficiency (F9): (7): c.1025C>T (p.T342M), c.128G>A(p.R43Q), c.1328T>C (p.I443T), c.217G>A (p.E73K), c.223C>T (p.R75X), c.316G>A (p.G106S), c.677G>A (p.R226Q)

Factor VIII Deficiency (F8): (32): c.1226A>G (p.E409G),c.1293G>T (p.L431F), c.1475A>G (p.Y492C), c.1648C>T (p.R550C), c.1660A>G (p.S554G), c.1750C>A (p.Q584K), c.1786T>C (p.S596P), c.1804C>T (p.R602X), c.1957G>A (p.V653M), c.2167G>A (p.A723T), c.2215G>A (p.E739K), c.43C>T (p.R15X), c.5096A>T (p.Y1699F), c.5122C>T (p.R1708C), c.5123G>A (p.R1708H), c.5143C>T (p.R1715X), c.5372T>C (p.M1791T), c.541G>A (p.V181M), c.5422C>T (p.L1808F), c.5822A>G (p.N1941S), c.6278A>G (p.D2093G), c.6360T>G (p.F2120L), c.6413C>A (p.S2138Y), c.6506G>A (p.R2169H), c.6532C>T (p.R2178C), c.6545G>A (p.R2182H), c.6683G>A (p.R2228Q), c.6744G>T (p.W2248C), c.6967C>T (p.R2323C), c.902G>T (p.R301L), c.935T>C (p.F312S), c.940A>G(p.T314A)

Familial Chloride Diarrhea (SLC26A3): (6): c.1386G>A (p.W462X), c.2023_2025dupATC (p.I675L), c.344delT (p.I115I), c.371A>T (p.H124L), c.559G>T(p.G187X), c.951delGGT (p.V318del)

Familial Dysautonomia (IKBKAP): (4): c.2087G>C (p.R696P), c.2128C>T (p.Q710X), c.2204+6T>C, c.2741C>T (p.P914L)

Familial Hyperinsulinism: Type 1: ABCC8 Related (ABCC8): (11): c.1333-1013A>G (IVS8-1013A>G), c.2147G>T (p.G716V), c.2506C>T (p.Q836X), c.3989-9G>A, c.4055G>C (p.R1352P), c.4159_4161delTTC (p.1387delF),c.4258C>T (p.R1420C), c.4477C>T (p.R1493W), c.4516G>A (p.E1506K), c.560T>A (p.V187D),c.579+2T>A

Familial Hyperinsulinism: Type 2: KCNJ11 Related (KCNJ11): (6): C.C761T (p.P254L), c.36C>A (p.Y12X), c.440T>C (p.L147P), c.776A>G (p.H259R), c.844G>A (p.E282K), c.G-134T

Familial Mediterranean Fever (MEFV): (10): c.1437C>G (p.F479L), c.2040G>A (p.M680I), c.2040G>C (p.M680I), c.2076_2078delAAT (p.692delI), c.2080A>G (p.M694V), c.2082G>A (p.M694I), c.2177T>C (p.V726A), c.2230G>T (p.A744S), c.2282G>A (p.R761H), c.800C>T (p.T267I)

Fanconi Anemia: Type A (FANCA): (10): c.1115_1118delTTGG, c.1606delT (p.S536fs), c.1615delG (p.D539fs), c.2172_2173insG (p.T724fs), c.295C>T (p.Q99X), c.3558_3559insG (p.R1187Efs), c.3720_3724delAAACA (p.E1240Dfs), c.4275delT (p.R1425fs), c.513G>A (p.W171X), c.890_893delGCTG (p.C297fs)

Fanconi Anemia: Type C (FANCC): (8): c.1642C>T (p.R548X), c.1661T>C (p.L554P), c.37C>T (p.Q13X), c.456+4A>T, c.553C>T (p.R185X), c.65G>A(p.W22X), c.66G>A (p.W22X), c.67delG

Fanconi Anemia: Type G (FANCG): (5): c.1480+1G>C, c.1794_1803delCTGGATCCGT (p.W599Pfs), c.307+1G>C, c.637_643delTACCGCC(p.Y213K+4X), c.925-2A>G

Fanconi Anemia: Type J (BRIP1): (1): c.2392C>T (p.R798X)

Fragile X Syndrome (FMR1): (1): c.-129CGG(>44)

Fumarase Deficiency (FH): (1): c.1433_1434insAAA

GM1-Gangliosidoses (GLB1): (17): c.1051C>T (p.R351X), c.1369C>T (p.R457X), c.1370G>A (p.R457Q), c.145C>T (p.R49C), c.1480-2A>G, c.152T>C(p.I51T), c.1577_1578insG, c.176G>A (p.R59H), c.1771T>A (p.Y591N), c.1772A>G (p.Y591C), c.202C>T (p.R68W), c.245C>T (p.T82M), c.367G>A (p.G123R), c.601C>T (p.R201C), c.622C>T (p.R208C), c.75+2_75+3insT, c.947A>G (p.Y316C)

GRACILE Syndrome (BCS1L): (12): c.103G>C (p.G35R), c.1057G>A (p.V353M), c.133C>T (p.R45C), c.148A>G (p.T50A), c.166C>T (p.R56X), c.232A>G (p.S78G), c.296C>T (p.P99L), c.464G>C (p.R155P), c.547C>T (p.R183C), c.548G>A (p.R183H), c.550C>T (p.R184C), c.830G>A (p.S277N)

Galactokinase Deficiency (GALK1): (7): c.1031C>T (p.T344M), c.1045G>A (p.G349S), c.1144C>T (p.Q382X), c.238G>T (p.E80X), c.593C>T (p.A198V), c.82C>A (p.P28T), c.94G>A (p.V32M)

Gaucher Disease (GBA): (6): c.1226A>G (p.N409S), c.1297G>T (p.V433L), c.1343A>T (p.D448V), c.1504C>T (p.R502C), c.1604G>A (p.R535H), c.84_85insG

Gitelman Syndrome (SLC12A3): (11): c.1046C>T (p.P348L), c.1180+1G>T (IVS9+1G>T), c.1670-191C>T, c.1763C>T (p.A588V), c.1868T>C (p.L623P), c.1889G>T (p.G629V), c.1926-1G>T, c.1961G>A (p.R654H), c.2548+253C>T, c.2883+1G>T, c.622C>T (p.R208W)

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DISEASES AND MUTATIONS LIST

Globoid Cell Leukodystrophy (GALC): (10): c.1153G>T (p.E385X), c.1161+6555_*9573del31670bp, c.1472delA (p.K491fs), c.1586C>T (p.T529M), c.1700A>C (p.Y567S), c.2002A>C (p.T668P), c.246A>G (p.I82M), c.683_694delATCTCTGGGAGTinsCTC (p.N228_S232del5insTP), c.857G>A (p.G286D), c.913A>G (p.I305V)

Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD): (7): c.1093G>A (p.A365T), c.1388G>A (p.R463H), c.1466G>C (p.R489P), c.1466G>T (p.R489L), c.577G>A (p.G163S), c.653C>T (p.S218F), c.961G>A (p.V291M)

Glutaric Acidemia: Type I (GCDH): (8): c.1083-2A>C (IVS10-2A>C), c.1093G>A (p.E365K), c.1198G>A (p.V400M), c.1204C>T (p.R402W), c.1262C>T(p.A421V), c.680G>C (p.R227P), c.743C>T (p.P248L), c.877G>A (p.A293T)

Glutaric Acidemia: Type IIA (ETFA): (5): c.346G>A (p.G116R), c.470T>G (p.V157G), c.797C>T (p.T266M), c.809_811delTAG (p.V270_A271delinsA), c.963+1delG

Glutaric Acidemia: Type IIB (ETFB): (2): c.655G>A (p.D219N), c.764G>A (p.R255Q)

Glutaric Acidemia: Type IIC (ETFDH): (8): c.1130T>C (p.L377P), c.1448C>T (p.P483L), c.250G>A (p.A84T), c.2T>C (p.M1T), c.36delA (p.A12fs), c.380T>A (p.L127H), c.524G>A (p.R175H), c.524G>T (p.R175L)

Glycine Encephalopathy: AMT Related (AMT): (6): c.125A>G (p.H42R), c.139G>A (p.G47R), c.574C>T (p.Q192X), c.826G>C (p.D276H), c.878-1G>A, c.959G>A (p.R320H)

Glycine Encephalopathy: GLDC Related (GLDC): (5): c.1545G>C (p.R515S), c.1691G>T (p.S564I), c.2266_2268delTTC (p.756delF), c.2284G>A (p.G762R), c.2T>C

Glycogen Storage Disease: Type IA (G6PC): (13): c.1039C>T (p.Q347X), c.113A>T (p.D38V), c.247C>T (p.R83C), c.248G>A (p.R83H), c.376_377insTA, c.562G>C (p.G188R), c.648G>T, c.724C>T (p.Q242X), c.724delC, c.79delC, c.809G>T (p.G270V), c.975delG (p.L326fs), c.979_981delTTC (p.327delF)

Glycogen Storage Disease: Type IB (SLC37A4): (5): c.1016G>A (p.G339D), c.1042_1043delCT, c.1099G>A (p.A367T), c.133T>C (p.W45R), c.796G>T (p.G266C)

Glycogen Storage Disease: Type II (GAA): (13): c.-32-13T>G (IVS1-13T>G), c.1561G>A (p.E521K), c.1585_1586delTCinsGT (p.S529V), c.1634C>T(p.P545L), c.1927G>A (p.G643R), c.1935C>A (p.D645E), c.2173C>T (p.R725W), c.2560C>T(p.R854X), c.2707_2709delK (p.903delK), c.525delT (p.E176Rfs), c.710C>T (p.A237V),c.896T>G (p.L299R), c.953T>C (p.M318T)

Glycogen Storage Disease: Type III (AGL): (15): c.1222C>T(p.R408X), c.1384delG (p.V462X), c.16C>T (p.Q6X), c.17_18delAG, c.2039G>A (p.W680X),c.2590C>T (p.R864X), c.2681+1G>A, c.2681+1G>T, c.3439A>G (p.R1147G), c.3682C>T (p.R1228X), c.3965delT (p.V1322AfsX27), c.3980G>A (p.W1327X), c.4260-12A>G (IVS32-12A>G), c.4342G>C (p.G1448R), c.4455delT (p.S1486fs)

Glycogen Storage Disease: Type IV (GBE1): (3): c.691+2T>C (IVS5+2T>C), c.986A>C (p.Y329S), c.986A>G (p.Y329C)

Glycogen Storage Disease: Type V (PYGM): (10): c.148C>T (p.R50X), c.1627A>T (p.K543X), c.1628A>C (p.K543T), c.1827G>A (p.K609K), c.2128_2130delTTC (p.710delF), c.2392T>C (p.W798R), c.255C>A (p.Y85X), c.613G>A(p.G205S), c.632delG (p.S211fs), c.808C>T (p.R270X)

Glycogen Storage Disease: Type VII (PFKM): (4): c.2214delC (p.P739Qfs), c.283C>T (p.R95X), c.329G>T (p.R110L), c.450+1G>A

Guanidinoacetate Methyltransferase Deficiency (GAMT): (4): c.148A>C (p.M50L), c.309_310insCCGGGACTGGGCC (p.L99_A103fs), c.327G>A, c.506G>A (p.C169Y)

HMG-CoA Lyase Deficiency (HMGCL): (7): c.109G>T(p.E37X), c.122G>A (p.R41Q), c.208G>C (p.V70L), c.561+1G>A, c.561+1G>T, c.835G>A(p.E279K), c.914_915delTT

Hemochromatosis: Type 2A: HFE2 Related (HFE2): (1): c.959G>T (p.G320V)

Hemochromatosis: Type 3: TFR2 Related (TFR2): (4): c.2069A>C (p.Q690P), c.515T>A (p.M172K), c.750C>G (p.Y250X), c.88_89insC (p.E60X)

Hemoglobinopathy: Hb C (HBB): (1): c.19G>A (p.E7K)

Hemoglobinopathy: Hb D (HBB): (1): c.364G>C (p.E122Q)
Hemoglobinopathy: Hb E (HBB): (1): c.79G>A (p.E27K)
Hemoglobinopathy: Hb O (HBB): (1): c.364G>A (p.E122K)

Hereditary Fructose Intolerance (ALDOB): (10): c.1005C>G (p.N335K), c.10C>T (p.R4X), c.178C>T (p.R60X), c.357_360delAAAC, c.442T>C (p.W148R),c.448G>C (p.A150P), c.524C>A (p.A175D), c.612T>G (p.Y204X), c.720C>A (p.C240X), c.865_867delCTT (p.289delL)

Hereditary Spastic Paraplegia: TECPR2 Related (TECPR2): (1): c.3416delT (p.L1139fs)

Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3): (1): c.1981C>T (p.R661X)

Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3): (6): c.124C>T (p.R42X), c.1903C>T (p.R635X), c.3024delT, c.3247C>T (p.Q1083X), c.430C>T (p.R144X), c.727C>T (p.Q243X)

Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2): (1): c.283C>T (p.R95X)

Hermansky-Pudlak Syndrome: Type 1 (HPS1): (1): c.1472_1487dup16 (p.H497Qfs)

Hermansky-Pudlak Syndrome: Type 3 (HPS3): (4): c.1163+1G>A, c.1189C>T (p.R397W), c.1691+2T>G, c.2589+1G>C

Hermansky-Pudlak Syndrome: Type 4 (HPS4): (7): c.1876C>T (p.Q626X), c.2039delC (p.P680fs), c.397G>T (p.E133X), c.526C>T (p.Q176X), c.634C>T,(p.R212X), c.649G>T (p.E217X), c.957_958insGCTTGTCCAGATGGCAGGAAGGAG (p.E319_N320ins8)

Holocarboxylase Synthetase Deficiency (HLCS): (7): c.1513G>C (p.G505R), c.1522C>T (p.R508W), c.1648G>A (p.V550M), c.1795+5G>A (IVS10+5G>A), c.710T>C (p.L237P), c.772_781delACAAGCAAGG (p.T258fs), c.780delG

Homocystinuria Caused by CBS Deficiency (CBS): (8): c.1006C>T (p.R336C), c.341C>T (p.A114V), c.572C>T (p.T191M), c.797G>A (p.R266K), c.833T>C (p.I278T), c.919G>A (p.G307S), c.959T>C (p.V320A), c.969G>A (p.W324X)

Hunter Syndrome (IDS): (7): c.1264T>G (p.C422G), c.1327C>T (p.R443X), c.1402C>T (p.R468W), c.1403G>A (p.R468Q), c.1403G>T (p.R468L), c.404A>G (p.K135R), c.998C>T (p.S333L)

Hurler Syndrome (IDUA): (8): c.1037T>G (p.L346R), c.1205G>A (p.W402X), c.152G>A (p.G51D), c.1598C>G (p.P533R), c.1960T>G (p.X654G), c.208C>T (p.Q70X), c.266G>A (p.R89Q), c.979G>C (p.A327P)

Hypohidrotic Ectodermal Dysplasia: X-Linked (EDA): (5): c.1013C>T (p.T338M), c.1072C>G (p.Q358E), c.463C>T (p.R155C), c.466C>T (p.R156C), c.467G>A (p.R156H)

Hypophosphatasia (ALPL): (5): c.1001G>A (p.G334D), c.1133A>T (p.D378V), c.1559delT, c.571G>A (p.E191K), c.979T>C (p.F327L)

Inclusion Body Myopathy: Type 2 (GNE): (3): c.131G> C (p.C44S), c.1807G>C (p.V603L), c.2228T>C (p.M743T)

Infantile Cerebral and Cerebellar Atrophy (MED17): (1): c.1112T>C (p.L371P)

Isolated Microphthalmia: VSX2 Related (VSX2): (4): c.371-1G>A, c.599G>A (p.R200Q), c.599G>C (p.R200P), c.679C>T (p.R227W)

Isovaleric Acidemia (IVD): (1): c.941C>T (p.A314V)

Joubert Syndrome (TMEM216): (2): c.218G>A (p.R73H), c.218G>T (p.R73L)

Juvenile Retinoschisis: X-Linked (RS1): (15): c.208G>A(p.G70S), c.208G>C (p.G70R), c.214G>A (p.E72K), c.221G>T (p.G74V), c.286T>C (p.W96R),c.304C>T (p.R102W), c.305G>A (p.R102Q), c.325G>C (p.G109R), c.421C>T (p.R141C),c.460C>T (p.Q154X), c.461A>G (p.Q154R), c.53_78del (p.A18Gfs), c.574C>T (p.P192S),c.598C>T (p.R200C), c.637C>T (p.R213W)

Lamellar Ichthyosis: Type 1 (TGM1): (1): c.877-2A>G (IVS5-2A>G)

Laryngoonychocutaneous Syndrome (LAMA3): (1): c.151_152insG (p.V51GfsX3)

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DISEASES AND MUTATIONS LIST

Leber Congenital Amaurosis: CEP290 Related (CEP290): (1): c.2991+1655A>G (p.C998X)

Leber Congenital Amaurosis: GUCY2D Related (GUCY2D): (3): c.1694T>C (p.F565S), c.2943delG (p.G982V), c.387delC (p.P130Lfx)

Leber Congenital Amaurosis: LCA5 Related (LCA5): (3): c.1151delC, c.1476_1477insA (p.P493TfsX1), c.835C>T (p.Q279X)

Leber Congenital Amaurosis: RDH12 Related (RDH12): (6): c.146C>T (p.T49M), c.184C>T (p.R62X), c.295C>A (p.L99I), c.464C>T (p.T155I), c.565C>T (p.Q189X), c.677A>G (p.Y226C)

Leigh Syndrome: French-Canadian (LRPPRC): (1): c.1061C>T (p.A354V)

Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related (EIF2B5): (9): c.1157G>T (p.G386V), c.166T>G (p.F56V), c.167T>G(p.F56C), c.1882T>C (p.W628R), c.271A>G (p.T91A), c.338G>A (p.R113H), c.584G>A(p.R195H), c.925G>C (p.V309L), c.944G>A (p.R315H)

Leydig Cell Hypoplasia (Luteinizing Hormone Resistance) (LHCGR): (13): c.1027T>A (p.C343S), c.1060G>A (p.E354K), c.1505T>C (p.L502P), c.1627T>C(p.C543R), c.1635C>A (p.C545X), c.1660C>T (p.R554X), c.1777G>C (p.A593P), c.1822_1827delCTGGTT (p.608_609delLV), c.1847C>A (p.S616Y), c.391T>C (p.C131R),c.430G>T (p.V144F), c.455T>C (p.I152T), c.537-3C>A

Limb-Girdle Muscular Dystrophy: Type 2A (CAPN3): (6):c.1469G>A (p.R490Q), c.1525G>T (p.V509F), c.1715G>A (p.R572Q), c.2306G>A (p.R769Q), c.2362_2363delAGinsTCATCT (p.R788Sfs), c.550delA (p.T184fs)

Limb-Girdle Muscular Dystrophy: Type 2B (DYSF): (5): c.2271C>A (p.Y758X), c.2833delG (p.A945fs), c.4989_4993delGCCCGinsCCCC (p.E1663fs) ,c.5174+5G>A, c.5830C>T (p.R1944X)

Limb-Girdle Muscular Dystrophy: Type 2C (SGCG): (4): c.525delT (p.F175fsX), c.787G>A (p.E263K), c.848G>A (p.C283Y), c.87_88insT (p.G30fs)

Limb-Girdle Muscular Dystrophy: Type 2D (SGCA): (1): c.229C>T (p.R77C)

Limb-Girdle Muscular Dystrophy: Type 2E (SGCB): (6):c.272G>C (p.R91P), c.272G>T (p.R91L), c.299T>A (p.M100K), c.323T>G (p.L108R), c.341C>T (p.S114F), c.452C>G (p.T151R)

Limb-Girdle Muscular Dystrophy: Type 2F (SGCD): (5): c.391G>C (p.A131P), c.493C>T (p.R165X), c.653delC (p.A218fs), c.784G>A (p.E262K), c.89G>A (p.W30X)

Limb-Girdle Muscular Dystrophy: Type 2I (FKRP): (1): c.826C>A (p.L276I)

Lipoprotein Lipase Deficiency (LPL): (1): c.644G>A (p.G215E)

Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA): (2): c.1132C>T (p.Q378X), c.1528G>C (p.E510Q)

Lowe Oculocerebrorenal Syndrome (OCRL): (3): c.1499G>A (p.R500Q), c.1572C>G (p.H524Q), c.2530C>T (p.R844X)

Lysinuric Protein Intolerance (SLC7A7): (4): c.1228C>T (p.R410X), c.1384_1385insATCA (p.R462fs), c.726G>A (p.W242X), c.895-2A>T

Malonyl-CoA Decarboxylase Deficiency (MLYCD): (5): c.1064_1065delTT (p.F355fs), c.560C>G (p.S187X), c.638_641delGTGA (p.S213fs), c.8G>A (p.G3D), c.949-14A>G

Maple Syrup Urine Disease: Type 1A (BCKDHA):(4): c.1312T>A (p.Y438N), c.288+1G>A, c.860_867delGAGGCCCC, c.868G>A (p.G290R)

Maple Syrup Urine Disease: Type 1B (BCKDHB):(6): c.1114G>T (p.E372X), c.487G>T (p.E163X), c.548G>C (p.R183P), c.832G>A (p.G278S),c.853C>T (p.R285X), c.970C>T (p.R324X)

Maple Syrup Urine Disease: Type 2 (DBT): (15): c.1169A>G(p.D390G), c.1193T>C (p.L398P), c.1202T>C (p.I401T), c.1209+5G>C (IVS9+5G>C), c.1232C>A (p.P411Q), c.1355A>G (p.H452R), c.1448G>T (p.X483L), c.294C>G (p.I98M),c.363_364delCT (p.Y122Lfs), c.581C>G (p.S194X), c.670G>T (p.E224X), c.75_76delAT(p.C26Wfs), c.788T>G (p.M263R), c.901C>T (p.R301C), c.939G>C (p.K313N)

Maple Syrup Urine Disease: Type 3 (DLD): (8): c.104_105insA (p.Y35fs), c.1081A>G (p.M361V), c.1123G>A (p.E375K), c.1178T>C (p.I393T), c.1463C>T (p.P488L), c.1483A>G (p.R495G), c.214A>G (p.K72E), c.685G>T (p.G229C)

Maroteaux-Lamy Syndrome (ARSB): (6): c.1143-1G>C, c.1143-8T>G, c.1178A>C (p.H393P), c.284G>A (p.R95Q), c.629A>G (p.Y210C), c.944G>A (p.R315Q)

Meckel Syndrome: Type 1 (MKS1): (5): c.1024+1G>A (IVS11+1G>A), c.1408-35_1408-7del29 (p.G470fs), c.417G>A (p.E139X), c.50insCCGGG (p.D19AfsX), c.80+2T>C (IVS1+2T>C)

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (ACADM): (8): c.199T>C (p.Y67H), c.262C>T (p.L88F), c.362C>T (p.T121I), c.595G>A (p.G199R), c.616C>T (p.R206C), c.617G>A (p.C206H), c.811C>T (p.G267R), c.985A>G (p.K329E)

Megalencephalic Leukoencephalopathy (MLC1): (6): c.135_136insC (p.C46fsX), c.176G>A (p.G59E), c.178-10T>A, c.278C>T (p.S93L), c.880C>T (p.P294S), c.908_918delTGCTGCTGCTGinsGCA (p.V303GfsX96)

Metachromatic Leukodystrophy (ARSA): (18): c.1114C>T (p.R372W), c.1136C>T (p.P379L), c.1210+1G>A, c.1232C>T (p.T411I), c.1283C>T (p.P428L),c.257G>A (p.R86Q), c.263G>A (p.G88D), c.292_293delTCinsCT (p.S98L), c.293C>T (p.S98F), c.302G>A (p.G101D), c.302G>T (p.G101V), c.465+1G>A (IVS2+1G>A), c.542T>G (p.I181S),c.641C>T (p.A214V), c.739G>A (p.G247R), c.769G>C (p.D257H), c.827C>T (p.T276M),c.862A>C (p.T288P)

Methylmalonic Acidemia: MMAA Related (MMAA): (14): c.1076G>A (p.R359Q), c.161G>A (p.W54X), c.266T>C (p.L89P), c.283C>T (p.Q95X),c.358C>T (p.Q120X), c.397C>T (p.Q133X), c.433C>T (p.R145X), c.503delC (p.T168MfsX9),c.562G>C (p.G188R), c.64C>T (p.R22X), c.650T>A (p.L217X), c.653G>A (p.G218E),c.733+1G>A, c.988C>T (p.R330X)

Methylmalonic Acidemia: MMAB Related (MMAB): (11): c.197-1G>T, c.287T>C (p.I96T), c.291-1G>A, c.403G>A (p.A135T), c.556C>T (p.R186W), c.568C>T (p.R190C), c.569G>A (p.R190H), c.571C>T (p.R191W), c.572G>A (p.R191Q), c.656A>G (p.Y219C), c.700C>T (p.Q234X)

Methylmalonic Acidemia: MUT Related (MUT): (23): c.1097A>G (p.N366S), c.1105C>T (p.R369C), c.1106G>A (p.R369H), c.1280G>A (p.G427D),c.1867G>A (p.G623R), c.2054T>G (p.L685R), c.2080C>T (p.R694W), c.2099T>A (p.M700K), c.2150G>T (p.G717V), c.278G>A (p.R93H), c.281G>T (p.G94V), c.284C>G (p.P95R),c.299A>G (p.Y100C), c.313T>C (p.W105R), c.322C>T (p.R108C), c.521T>C (p.F174S),c.572C>A (p.A191E), c.607G>A (p.G203R), c.643G>A (p.G215S), c.643G>T (p.G215C),c.655A>T (p.N219Y), c.691T>A (p.Y231N), c.935G>T (p.G312V)

Methylmalonic Aciduria and Homocystinuria: Type cblC (MMACHC): (5): c.271_272insA (p.R91KfsX14), c.331C>T (p.R111X), c.394C>T (p.R132X), c.482G>A (p.R161Q), c.609G>A (p.W203X)

Mitochondrial Complex I Deficiency: NDUFS6 Related (NDUFS6): (1): c.344G>A (p.C115Y)

Mitochondrial DNA Depletion Syndrome: MNGIE Type (TYMP): (6): c.1425_1426insC (p.S476Lfs), c.433G>A (p.G145R), c.457G>A (p.G153S), c.516+2T>C (IVS4+2T>C), c.665A>G (p.K222R), c.866A>C (p.E289A)

Mitochondrial Myopathy and Sideroblastic Anemia (PUS1): (2): c.430C>T (p.R144W), c.658G>T (p.E220X)

Mitochondrial Trifunctional Protein Deficiency: HADHB Related (HADHB): (7): c.1175C>T (p.A392V), c.1331G>A (p.R444K), c.1364T>G (p.V455G), c.182G>A (p.R61H), c.740G>A (p.R247H), c.776_777insT (p.G259fs), c.788A>G (p.D263G)

Morquio Syndrome: Type A (GALNS): (6): c.1156C>T(p.R386C), c.178G>A (p.D60N), c.205T>G (p.F69V), c.337A>T (p.I113F), c.485C>T (p.S162F),c.901G>T (p.G301C)

Morquio Syndrome: Type B (GLB1): (8): c.1223A>C(p.Q408P), c.1313G>A (p.G438E), c.1444C>T (p.R482C), c.1445G>A (p.R482H), c.1498A>G(p.T500A), c.1527G>T (p.W509C), c.247T>C (p.Y83H), c.817_818delTGinsCT (p.W273L)

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DISEASES AND MUTATIONS LIST

Mucolipidosis: Type II/III (GNPTAB): (3): c.1120T>C (p.F374L), c.3503_3504delTC (p.L1168QfsX5), c.3565C>T (p.R1189X)

Mucolipidosis: Type IV (MCOLN1): (5): c.-1015_788del6433, c.1084G>T (p.D362Y), c.244delC (p.L82fsX), c.304C>T (p.R102X), c.406-2A>G

Multiple Pterygium Syndrome (CHRNG): (6): c.136C>T (p.R46X), c.13C>T (p.Q5X), c.1408C>T (p.R470X), c.320T>G (p.V107G), c.401_402delCT (p.P134fs), c.715C>T (p.R239C)

Multiple Sulfatase Deficiency (SUMF1): (1): c.463T>C (p.S155P)

Muscle-Eye-Brain Disease (POMGNT1): (3): c.1324C>T (p.R442C), c.1478C>G (p.P493R), c.1539+1G>A

Myotubular Myopathy: X-Linked (MTM1): (23): c.109C>T(p.R37X), c.1190A>G (p.Y415C), c.1261-10A>G, c.141_144delAGAA (p.E48Lfs), c.1420C>T(p.R474X), c.205C>T (p.R69C), c.208C>T (p.L70F), c.212_215delATTT (p.Y71X), c.260T>C (p.L87P), c.297_298insCG (p.T100RfsX11), c.397_398delAT (p.M133Vfs), c.469G>A (p.E157K),c.49G>T (p.E17X), c.566A>G (p.N189S), c.614C>T (p.P205L), c.649_650insG (p.A217GfsX4), c.664C>T (p.R222X), c.670C>T (p.R224X), c.679-1G>A (IVS8-1G>A), c.685T>C (p.S229P),c.709_710delAC (p.T237fsX), c.70C>T (p.R24X), c.721C>T (p.R241C)

Navajo Neurohepatopathy (MPV17): (1): c.149G>A (p.R50Q)

Nemaline Myopathy: NEB Related (NEB): (2): c.7434_7536del2502bp, c.8890-2A>G (IVS63-2A>G)

Nephrotic Syndrome: Type 1 (NPHS1): (5): c.121_122delCT (p.L41Dfs), c.1481delC, c.2335-1G>A, c.3325C>T (p.R1109X), c.3478C>T (p.R1160X)

Nephrotic Syndrome: Type 2 (NPHS2): (27): c.104_105insG (p.G35fsX69), c.274G>T (p.G92C), c.353C>T (p.P118L), c.412C>T (p.R138X), c.413G>A(p.R138Q), c.419delG (p.G140fsX180), c.467_468insT (p.L156fsX166), c.467delT(p.L156fsX180), c.479A>G (p.D160G), c.502C>A (p.R168S), c.502C>T (p.R168C), c.503G>A(p.R168H), c.538G>A (p.V180M), c.555delT (p.F185fsX186), c.622G>A (p.A208T), c.706_714delCTAGAGAGG (p.L236_R238del), c.714G>T (p.R238S), c.779T>A (p.V260E),c.851C>T (p.A284V), c.855_856delAA (p.Q285fsX302), c.85G>A (p.A29T), c.862G>A (p.A288T), c.868G>A (p.V290M), c.871C>T (p.R291W), c.948delT (p.A317L), c.964C>T(p.R322X), c.976_977insA (p.T326fsX345)

Neuronal Ceroid-Lipofuscinosis: CLN5 Related (CLN5): (7): c.1054G>T (p.E352X), c.1121A>G (p.Y374C), c.1175_1176delAT (p.Y392X), c.225G>A (p.W75X), c.335G>A (p.R112H), c.377G>A (p.C126Y), c.835G>A (p.D279N)

Neuronal Ceroid-Lipofuscinosis: CLN6 Related (CLN6): (8): c.139C>T (p.L47F), c.17G>C (p.R6T), c.200T>C (p.L67P), c.214G>T (p.E72X), c.308G>A (p.R103Q), c.368G>A (p.G123D), c.460_462delATC (p.I154del), c.663C>G (p.Y221X)

Neuronal Ceroid-Lipofuscinosis: CLN8 Related (CLN8): (4): c.610C>T (p.R204C), c.70C>G (p.R24G), c.789G>C (p.W263C), c.88G>C (p.A30P)

Neuronal Ceroid-Lipofuscinosis: MFSD8 Related (MFSD8): (2): c.754+2T>A, c.881C>A (p.T294K)

Neuronal Ceroid-Lipofuscinosis: PPT1 Related (PPT1): (8): c.134G>A (p.C45Y), c.223A>C (p.T75P), c.236A>G (p.D79G), c.29T>A (p.L10X), c.322G>C (p.G108R), c.364A>T (p.R122W), c.451C>T (p.R151X), c.656T>A (p.L219Q)

Neuronal Ceroid-Lipofuscinosis: TPP1 Related (TPP1): (9): c.1093T>C (p.C365R), c.1094G>A (p.C365Y), c.1340G>A (p.R477H), c.509-1G>A, c.509-1G>C, c.616C>T (p.R206C), c.622C>T (p.R208X), c.851G>T (p.G284V), c.857A>G (p.N286S)

Niemann-Pick Disease: Type A (SMPD1): (6): c.1267C>T (p.H423Y), c.1493G>A (p.R498H), c.1493G>T (p.R498L), c.1734G>C (p.K578N), c.911T>C (p.L304P), c.996delC

Niemann-Pick Disease: Type B (SMPD1): (3): c.1280A>G (p.H427R), c.1829_1831delGCC (p.610delR), c.880C>A (p.Q294K)

Niemann-Pick Disease: Type C1 (NPC1): (14): c.1133T>C(p.V378A), c.2324A>C (p.Q775P), c.2665G>A (p.V889M), c.2783A>C (p.Q928P),c.2848G>A (p.V950M), c.2932C>T (p.R978C), c.2974G>C (p.G992R), c.2974G>T(p.G992W), c.3107C>T (p.T1036M), c.3182T>C (p.I1061T), c.3263A>G (p.Y1088C), c.337T>C(p.C113R), c.3467A>G (p.N1156S), c.530G>A (p.C177Y)

Niemann-Pick Disease: Type C2 (NPC2): (11): c.115G>A (p.V39M), c.133C>T (p.Q45X), c.141C>A (p.C47X), c.190+5G>A, c.199T>C (p.S67P),c.295T>C (p.C99R), c.332delA (p.N111Ifs), c.352G>T (p.E118X), c.358C>T (p.P120S), c.436C>T(p.Q146X), c.58G>T (p.E20X)

Nijmegen Breakage Syndrome (NBN): (1): c.657_661delACAAA (p.K219fs)

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): (28): c.-23+1G>A, c.-259C>T, c.109G>A (p.V37I), c.134G>A (p.G45E), c.139G>T (p.E47X), c.167delT, c.229T>C (p.W77R), c.231G>A (p.W77X), c.235delC, c.250G>C (p.V84L),c.269T>C (p.L90P), c.283G>A (p.V95M), c.290_291insA (p.Y97fs), c.299_300delAT(p.H100Rfs), c.313_326delAAGTTCATCAAGGG, c.334_335delAA (p.K112fs), c.358delGAG(p.120delE), c.35G>T (p.G12V), c.35delG (p.G12fs), c.370C>T (p.Q124X), c.427C>T(p.R143W), c.439G>A (p.E147K), c.44A>C (p.K15T), c.516G>A (p.W172X), c.550C>T(p.R184W), c.551G>C (p.R184P), c.617A>G (p.N206S), c.71G>A (p.W24X)

Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related (LOXHD1): (2): c.2008C>T (p.R670X), c.4714C>T (p.R1572X)

Nonsyndromic Hearing Loss and Deafness: MYO15A Related (MYO15A): (10): c.3313G>T (p.E1105X), c.3334delG (p.G1112fs), c.3685C>T (p.Q1229X), c.3866+1G>A, c.3866+1G>T, c.453_455delCGAinsTGGACGCCTGGTCGGGCAGTGG (p.E152GfsX81), c.6331A>T (p.N2111Y), c.6337A>T (p.I2113F), c.7801A>T (p.K2601X), c.8148G>T (p.Q2716H)

Oculocutaneous Albinism: Type 1 (TYR): (27): c.1064C>T(p.A355V), c.1090A>C (p.N364H), c.1118C>A (p.T373K),c.1138_1158delTCTGCCAACGATCCTATCTTC (p.S380_F386del), c.1150C>G (p.P384A),c.1184+1G>A, c.1309G>A (p.D437N), c.133_134insC (p.P45fs), c.140G>A (p.G47D),c.1467_1468insT (p.A490Cfs), c.1469C>A (p.A490D), c.149C>T (p.S50L), c.1A>G (p.M1V),c.229C>T (p.R77W), c.242C>T (p.P81L), c.265T>C (p.C89R), c.272G>A (p.C91Y), c.325G>A (p.G109R), c.32G>A (p.W11X), c.568delG (p.G191Dfs), c.707G>A (p.W236X), c.710delA (p.D237fs), c.820-2A>G, c.823G>T (p.V275F), c.832C>T (p.R278X), c.892C>T (p.R298W), c.978delA (p.Q326fs)

Oculocutaneous Albinism: Type 3 (TYRP1): (6): c.1057_1060delAACA (p.N353fs), c.1067G>A (p.R356Q), c.107delT, c.1103delA (p.K368fs), c.1120C>T (p.R374X), c.497C>G (p.S166X)

Oculocutaneous Albinism: Type 4 (SLC45A2): (2): c.469G>A (p.D157N), c.563G>T (p.G188V)

Omenn Syndrome: DCLRE1C Related (DCLRE1C): (1): c.597C>A (p.Y199X)

Omenn Syndrome: RAG2 Related (RAG2): (1) c.685C>T (p.R229W)

Ornithine Transcarbamylase Deficiency (OTC): (11): c.118C>T (p.R40C), c.119G>A (p.R40H), c.134T>C (p.L45P), c.148G>T (p.G50X), c.236G>A (p.G79E), c.259G>A (p.E87K), c.274C>T (p.R92X), c.281G>C (p.R94T), c.533C>T (p.T178M), c.77G>A(p.R26Q), c.829C>T (p.R277W)

Ornithine Translocase Deficiency (SLC25A15): (3): c.535C>T (p.R179X), c.562_564delTTC (p.188delF), c.95C>G (p.T32R)

Osteopetrosis: TCIRG1 Related (TCIRG1): (6): c.117+4A>T, c.1213G>A (p.G405R), c.1331G>T (p.R444L), c.1392C>A (p.C464X), c.1674-1G>A, c.922delC (p.Q308fs)

POLG Related Disorders: Autosomal Recessive (POLG): (16): c.1399G>A (p.A467T), c.1491G>C (p.Q497H), c.1760C>T (p.P587L), c.2243G>C (p.W748S), c.2542G>A (p.G848S), c.2591A>G (p.N864S), c.2617G>T (p.E873X), c.2794C>T(p.H932Y), c.3151G>C (p.G1051R), c.3218C>T (p.P1073L), c.3488T>G (p.M1163R), c.679C>T(p.R227W), c.695G>A (p.R232H), c.752C>T (p.T251I), c.8G>C (p.R3P), c.911T>G (p.L304R)

Papillon-Lefevre Syndrome (CTSC): (11): c.1047delA (p.G350Vfs), c.1056delT (p.Y352fs), c.1287G>C (p.W429C), c.380A>C (p.H127P), c.628C>T(p.R210X), c.755A>T (p.Q252L), c.815G>A (p.R272H), c.856C>T (p.Q286X), c.857A>G(p.Q286R), c.890-1G>A, c.96T>G (p.Y32X)

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DISEASES AND MUTATIONS LIST

Pendred Syndrome (SLC26A4): (7): c.1001+1G>A, c.1151A>G (p.E384G), c.1246A>C (p.T416P), c.2168A>G (p.H723R), c.707T>C (p.L236P), c.716T>A (p.V239D), c.919-2A>G

Persistent Mullerian Duct Syndrome: Type I (AMH): (6): c.1144G>T (p.E382X), c.1518C>G (p.H506Q), c.1574G>A (p.C525Y), c.17_18delTC, c.283C>T (p.R95X), c.571C>T (p.R191X)

Persistent Mullerian Duct Syndrome: Type II (AMHR2): (14): c.118G>T (p.G40X), c.1217G>A (p.R406Q), c.1277A>G (p.D426G), c.1330_1356delCTGGGCAATACCCCTACCTCTGATGAG, c.1373T>C (p.V458A), c.1471G>C(p.D491H), c.1510C>T (p.R504C), c.160C>T (p.R54C), c.232+1G>A, c.289C>T (p.R97X),c.425G>T (p.G142V), c.596delA, c.742G>A (p.E248K), c.846T>G (p.H282Q)

Phenylalanine Hydroxylase Deficiency (PAH): (61): c.1042C>G (p.L348V), c.1045T>C (p.S349P), c.1066-11G>A (IVS10-11G>A), c.1068C>G (p.Y356X), c.1139C>T (p.T380M), c.1157A>G (p.Y386C), c.1169A>G (p.E390G), c.117C>G(p.F39L), c.1222C>T (p.R408W), c.1223G>A (p.R408Q), c.1238G>C (p.R413P), c.1241A>G (p.Y414C), c.1301C>A (p.A434D), c.1315+1G>A (IVS12+1G>A), c.136G>A (p.G46S), c.143T>C (p.L48S), c.194T>C (p.I65T), c.199T>C (p.S67P), c.1A>G (p.M1V), c.241_256delACCCATTTGGATAAAC (p.T81fs), c.331C>T (p.R111X), c.3G>A (p.M1I), c.442-1G>A (IVS4-1G>A), c.463_464insTGTGTACC (p.R155fs), c.473G>A (p.R158Q), c.533A>G(p.E178G), c.569T>G (p.V190G), c.581T>C (p.L194P), c.611A>G (p.Y204C), c.682G>T(p.E228X), c.721C>T (p.R241C), c.722G>A (p.R241H), c.722G>T (p.R241L), c.727C>T(p.R243X), c.728G>A (p.R243Q), c.734T>C (p.V245A), c.745C>T (p.L249F), c.754C>T (p.R252W), c.755G>A (p.R252Q), c.764T>C (p.L255S), c.770G>T (p.G257V), c.781C>T(p.R261X), c.782G>A (p.R261Q), c.800A>G (p.Q267R), c.814G>T (p.G272X), c.818C>T(p.S273F), c.829T>G (p.Y277D), c.838G>A (p.E280K), c.842+2T>A (IVS7+2T>A), c.842+5G>A(IVS7+5G>A), c.842C>T (p.P281L), c.856G>A (p.E286K), c.896T>G (p.F299C), c.898G>T (p.A300S), c.899C>T (p.A300V), c.904delT (p.F302fs), c.913-7A>G (IVS8-7A>G), c.926C>A (p.A309D), c.926C>T (p.A309V), c.935G>T (p.G312V), c.997C>T (p.L333F)

Polyglandular Autoimmune Syndrome: Type I (AIRE): (5): c.1163_1164insA (p.M388IfsX36), c.254A>G (p.Y85C), c.415C>T (p.R139X), c.769C>T (p.R257X), c.967_979delCTGTCCCCTCCGC (p.L323SfsX51)

Pontocerebellar Hypoplasia: EXOSC3 Related (EXOSC3): (4): c.238G>T (p.V80F), c.294_303delTGTTTACTGG (p.V99Wfs), c.395A>C (p.D132A), c.92G>C (p.G31A)

Pontocerebellar Hypoplasia: RARS2 Related (RARS2): (3): c.1024A>G (p.M342V), c.110+5A>G, c.35A>G (p.Q12R)

Pontocerebellar Hypoplasia: SEPSECS Related (SEPSECS): (1): c.1001A>G (p.Y334C)

Pontocerebellar Hypoplasia: TSEN54 Related (TSEN54): (3): c.1027C>T (p.Q343X), c.736C>T (p.Q246X), c.919G>T (p.A307S)

Pontocerebellar Hypoplasia: VPS53 Related (VPS53): (2): c.1556+5G>A, c.2084A>G (p.Q695R)

Pontocerebellar Hypoplasia: VRK1 Related (VRK1): (2): c.1072C>T (p.R358X), c.397C>T (p.R133C)

Primary Carnitine Deficiency (SLC22A5): (12): c.1195C>T (p.R399W), c.1196G>A (p.R399Q), c.1202_1203insA (p.Y401fsX), c.1324_1325delGCinsAT (p.A442I), c.1433C>T (p.P478L), c.396G>A (p.W132X), c.43G>T (p.G15W), c.505C>T (p.R169W), c.506G>A (p.R169Q), c.632A>G (p.Y211C), c.844C>T (p.R282X), c.95A>G(p.N32S)

Primary Ciliary Dyskinesia: DNAI1 Related (DNAI1): (5): c.1490G>A (p.G497D), c.1543G>A (p.G515S), c.1658_1669delCCAAGGTCTTCA (p.Thr553_Phe556del), c.282_283insAATA (p.G95Nfs), c.48+2_48+3insT

Primary Ciliary Dyskinesia: DNAI2 Related (DNAI2): (4): c.1304G>A (p.W435X), c.1494+1G>A, c.346-3T>G, c.787C>T (p.R263X)

Primary Congenital Glaucoma (CYP1B1): (9): c.1064_1076delGAGTGCAGGCAGA (p.R355Hfs), c.1093G>T (p.G365W), c.1199_1200insTCATGCCACC, c.1405C>T (p.R469W), c.1410_1422delCATTGGCGAAGAA (p.C470fs), c.155C>T (p.P52L), c.182G>A (p.G61E), c.535delG (p.A179fs), c.862_863insC

Primary Hyperoxaluria: Type 1 (AGXT): (11): c.121G>A (p.G41R), c.198C>G (p.Y66X), c.245G>A (p.G82E), c.454T>A (p.F152I), c.466G>A (p.G156R), c.508G>A (p.G170R), c.613T>C (p.S205P), c.697C>T (p.R233C), c.698G>A (p.R233H), c.731T>C (p.I244T), c.738G>A (p.W246X)

Primary Hyperoxaluria: Type 2 (GRHPR): (3): c.103delG, c.295C>T (p.R99X), c.404+3delAAGT

Primary Hyperoxaluria: Type 3 (HOGA1): (2): c.860G>T (p.G287V), c.944_946delAGG (p.315delE)

Progressive Familial Intrahepatic Cholestasis: Type 2 (ABCB11): (5): c.1295G>C (p.R432T), c.1723C>T (p.R575X), c.3169C>T (p.R1057X), c.3767_3768insC, c.890A>G (p.E297G)

Propionic Acidemia: PCCA Related (PCCA): (13): 916_917insT, c.1192T>C (p.C398R), c.1196G>A (p.R399Q), c.1268C>T (p.P423L),c.1643+1G>A (IVS18+1G>A), c.1644-6C>G (IVS18-6C>G), c.1685C>G (p.S562X),c.1746G>A (p.S582S), c.229C>T (p.R77W), c.590G>A (p.G197E), c.862A>G (p.R288G),c.890A>G (p.Q297R), c.937C>T (p.R313X)

Propionic Acidemia: PCCB Related (PCCB): (13): c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA (p.G407fs), c.1228C>T (p.R410W), c.1283C>T (p.T428I), c.1304A>G (p.Y435C), c.1495C>T (p.R499X), c.1534C>T (p.R512C), c.1539_1540insCCC (p.R514PfsX38), c.1556T>C (p.L519P), c.1606A>G (p.N536D), c.280G>T(p.G94X), c.335G>A (p.G112D), c.457G>C (p.A153P), c.502G>A (p.E168K)

Pseudocholinesterase Deficiency (BCHE): (1): c.293A>G (p.D98G)

Pycnodysostosis (CTSK): (2): c.926T>C (p.L309P), c.990A>G (p.X330W)

Pyruvate Carboxylase Deficiency (PC): (15): c.1351C>T(p.R451C), c.1748G>T (p.R583L), c.1828G>A (p.A610T), c.1828G>T (p.A610S), c.184C>T(p.R62C), c.1892G>A (p.R631Q), c.2229G>T (p.M743I), c.2473+2_2473+5delTAGG, c.2491_2492delGT (p.V831fs), c.2493_2494delGT (p.F832Xfs), c.2540C>T (p.A847V), c.2876_2877insT (p.F959fs), c.3409_3410delCT (p.L1137fs), c.434T>C (p.V145A), c.467G>A (p.R156Q)

Pyruvate Dehydrogenase Deficiency (PDHB): (2): c.1030C>T (p.P344S), c.395A>G (p.Y132C)

Pyruvate Dehydrogenase Deficiency: X-Linked (PDHA1): (4): c.1133G>A (p.R378H), c.1145_1146insATCA, c.648A>C (p.L216F), c.901C>G (p.R301G)

Renal Tubular Acidosis and Deafness (ATP6V1B1): (7): c.1037C>G (p.P346R), c.1155_1156insC (p.I386fs), c.1248+1G>C, c.232G>A (p.G78R), c.242T>C (p.L81P), c.497delC (p.T166fs), c.585+1G>A

Retinal Dystrophies: RLBP1 Related (RLBP1): (3): c.141+2T>C, c.141G>A (p.K47=), c.700C>T (p.R234W)

Retinal Dystrophies: RPE65 Related (RPE65): (12): c.1022T>C (p.L341S), c.1067delA (p.N356fs), c.1087C>A (p.P363T), c.11+5G>A, c.1102T>C (p.Y368H), c.1292A>G (p.Y431C), c.1355T>G (p.V452G), c.1543C>T (p.R515W), c.271C>T (p.R91W), c.700C>T (p.R234X), c.907A>T (p.K303X), c.95-2A>T (IVS2-2A>T)

Retinitis Pigmentosa: CERKL Related (CERKL): (5): c.238+1G>A (IVS1+1G>A), c.420delT (p.I141Lfs), c.598A>T (p.K200X), c.769C>T (p.R257X), c.780delT (p.P261Lfs)

Retinitis Pigmentosa: DHDDS Related (DHDDS): (1): c.124A>G (p.K42E)

Retinitis Pigmentosa: FAM161A Related (FAM161A): (5): c.1309A>T, c.1355_1356delCA (p.T452fs), c.1567C>T (p.R523X), c.1786C>T (p.R596X), c.685C>T (p.R229X)

Rhizomelic Chondrodysplasia Punctata: Type I (PEX7): (8): c.120C>G (p.Y40X), c.345T>G (p.Y115X), c.40A>C (p.T14P), c.45_52insGGGACGCC (p.H18RfsX35), c.649G>A (p.G217R), c.653C>T (p.A218V), c.875T>A (p.L292X), c.903+1G>C

SCID: X-Linked (IL2RG): (12): c.186T>A (p.C62X), c.341G>A (p.G114D), c.343T>C (p.C115R), c.355A>T (p.K119X), c.454+1G>A, c.458T>A (p.I153N), c.515T>C (p.L172P), c.664C>T (p.R222C), c.854G>A (p.R285Q), c.865C>T (p.R289X), c.878T>A (p.L293Q), c.923C>A (p.S308X)

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DISEASES AND MUTATIONS LIST

Salla Disease (SLC17A5): (5): c.1001C>G (p.P334R), c.115C>T (p.R39C), c.406A>G (p.K136E), c.548A>G (p.H183R), c.802_816delTCATCATTAAGAAAT (p.L336fsX13)

Sandhoff Disease (HEXB): (14): c.1082+5G>A, c.1250C>T (p.P417L), c.1303_1304delAG (p.R435fs), c.1509-26G>A, c.1514G>A (p.R505Q), c.1597C>T (p.R533C), c.1615C>T (p.R539C), c.445+1G>A, c.508C>T (p.R170X), c.76delA, c.796T>G (p.Y266D), c.800_816delCACCAAATGATGTCCGT (p.T267fs), c.845G>A (p.G282E), c.850C>T(p.R284X)

Sanfilippo Syndrome: Type A (SGSH): (11): c.1080delC (p.T360fs), c.1105G>A (p.E369K), c.1298G>A (p.R433Q), c.1339G>A (p.E447K), c.197C>G(p.S66W), c.220C>T (p.R74C), c.383C>T (p.P128L), c.449G>A (p.R150Q), c.617G>C(p.R206P), c.734G>A (p.R245H), c.892T>C (p.S298P)

Sanfilippo Syndrome: Type B (NAGLU): (10): c.1444C>T (p.R482W), c.1562C>T (p.P521L), c.1693C>T (p.R565W), c.1694G>C (p.R565P), c.1876C>T (p.R626X), c.1927C>T (p.R643C), c.1928G>A (p.R643H), c.2021G>A (p.R674H), c.700C>T (p.R234C), c.889C>T (p.R297X)

Sanfilippo Syndrome: Type C (HGSNAT): (13): c.1030C>T(p.R344C), c.1150C>T (p.R384X), c.1345insG (p.D449fsX), c.1529T>A (p.M510K), c.1553C>T(p.S518F), c.1622C>T (p.S541L), c.234+1G>A (IVS2+1G>A), c.372-2A>G (IVS3-A>G),c.493+1G>A (IVS4+1G>A), c.525_526insT (p.A175fsX), c.848C>T (p.P283L), c.852-1G>A,c.962T>G (p.L321X)

Sanfilippo Syndrome: Type D (GNS): (5): c.1063C>T (p.R355X), c.1138insGTCCT (p.D380fsX), c.1168C>T (p.Q390X), c.1169delA (p.Q390fsX), c.1226insG (p.R409fsX)

Short-Chain Acyl-CoA Dehydrogenase Deficiency (ACADS): (5): c.1058C>T (p.S353L), c.1138C>T (p.R380W), c.1147C>T (p.R383C), c.319C>T (p.R107C), c.575C>T (p.A192V)

Sickle-Cell Anemia (HBB): (1): c.20A>T (p.E7V)

Sjogren-Larsson Syndrome (ALDH3A2): (2): c.1297_1298delGA (p.E433fs), c.943C>T (p.P315S)

Sly Syndrome (GUSB): (5): c.1222C>T (p.P408S), c.1244C>T (p.P415L), c.1429C>T (p.R477W), c.1856C>T (p.A629V), c.526C>T (p.L176F)

Smith-Lemli-Opitz Syndrome (DHCR7):(50): c.1039G>A(p.G347S), c.1054C>T (p.R352W), c.1055G>A (p.R352Q), c.1079T>C (p.L360P), c.111G>A(p.W37X), c.1139G>A (p.C380Y), c.1190C>T (p.S397L), c.1210C>T (p.R404C), c.1228G>A (p.G410S), c.1295A>G (p.Y432C), c.1327C>T (p.R443C), c.1337G>A (p.R446Q), c.1342G>A(p.E448K), c.1351T>C (p.C451R), c.1384T>C (p.Y462H), c.1406G>C (p.R469P), c.1424T>C(p.F475S), c.151C>T (p.P51S), c.1A>G, c.203T>C (p.L68P), c.278C>T (p.T93M), c.292C>T(p.Q98X), c.296T>C (p.L99P), c.326T>C (p.L109P), c.356A>T (p.H119L), c.443T>G (p.L148R),c.452G>A (p.W151X), c.453G>A (p.W151X), c.470T>C (p.L157P), c.502T>A (p.F168I),c.506C>T (p.S169L), c.523G>C (p.D175H), c.532A>T (p.I178F), c.536C>T (p.P179L), c.545G>T(p.W182L), c.575C>T (p.S192F), c.670G>A (p.E224K), c.682C>T (p.R228W), c.724C>T(p.R242C), c.725G>A (p.R242H), c.728C>G (p.P243R), c.744G>T (p.W248C), c.818T>G(p.V273G), c.852C>A (p.F284L), c.853_855delTTC (p.285delF), c.861C>A (p.N287K),c.906C>G (p.F302L), c.964-1G>C, c.970T>C (p.Y324H), c.976G>T (p.V326L)

Spinal Muscular Atrophy: SMN1 Linked (SMN1): (19): c.22_23insA, c.305G>A (p.W102X), c.400G>A (p.E134K), c.439_443delGAAGT, c.43C>T(p.Q15X), c.558delA, c.585_586insT, c.683T>A (p.L228X), c.734C>T p.P245L),c.768_778dupTGCTGATGCTT, c.815A>G (p.Y272C), c.821C>T (p.T274I), c.823G>A (p.G275S),c.834+2T>G, c.835-18_835-12delCCTTTAT, c.835G>T, c.836G>T, c.91_92insT (19): DEL EXON 7

Stargardt Disease (ABCA4): (16): c.1018T>G (p.Y340D) ,c.1622T>C (p.L541P), c.1938-1G>A, c.2461T>A (p.W821R), c.2565G>A (p.W855X),c.2588G>C (p.G863A), c.3083C>T (p.A1028V), c.3106G>A (p.E1036K), c.3113C>T(p.A1038V), c.3210_3211insGT (p.S1071Vfs), c.3364G>A (p.E1122K), c.52C>T (p.R18W), c.5338C>G (p.P1780A), c.571-2A>G, c.6079C>T (p.L2027F), c.634C>T (p.R212C)

Stuve-Wiedemann Syndrome (LIFR): (9): c.1601-2A>G, c.1620_1621insA, c.170delC, c.1789C>T (pR597X), c.2274_2275insT, c.2434C>T (p.R812X), c.2472_2476delTATGT, c.653_654insT, c.756_757insT (p.K253X)

Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2): (7): c.-26+2T>C, c.1018_1020delGTT (p.340delV), c.1957T>A (p.C653S), c.398C>T (p.A133V), c.532C>T (p.R178X), c.764G>A (p.G255E), c.835C>T (p.R279W)

Tay-Sachs Disease (HEXA): (78): c.1003A>T (p.I335F), c.1008G>T (p.Q336H), c.1043_1046delTCAA (p.F348fs), c.1061_1063delTCT (p.F354_Y355delinsX), c.1073+1G>A, c.1121A>G (p.Q374R), c.1123delG (p.E375fs),c.1141delG (p.V381fs), c.1146+1G>A, c.116T>G (p.L39R), c.1177C>T (p.R393X), c.1178G>C(p.R393P), c.1211_1212delTG (p.L404fs), c.1277_1278insTATC, c.1292G>A (p.W431X), c.1302C>G (p.F434L), c.1307_1308delTA (p.I436fs), c.1351C>G (p.L451V), c.1385A>T (p.E462V), c.1421+1G>C, c.1422-2A>G, c.1426A>T (p.R476X), c.1432G>A (p.G478R), c.1451T>C (p.L484P), c.1495C>T (p.R499C), c.1496G>A (p.R499H), c.1510C>T (p.R504C),c.1510delC (p.R504fs), c.1511G>A (p.R504H), c.1511G>T (p.R504L), c.1537C>T (p.Q513X),c.155C>A (p.S52X), c.1A>G (p.M1V), c.2T>C (p.M1T), c.340G>A (p.E114K), c.346+1G>C, c.380T>G (p.L127R), c.409C>T (p.R137X), c.413-2A>G, c.426delT (p.F142fs), c.459+5G>A(IVS4+5G>A), c.508C>T (p.R170W), c.509G>A (p.R170Q), c.532C>T (p.R178C), c.533G>A(p.R178H), c.533G>T (p.R178L), c.535C>T (p.H179Y), c.536A>G (p.H179R), c.538T>C(p.Y180H), c.540C>G (p.Y180X), c.570+3A>G, c.571-1G>T, c.571-2A>G (IVS5-2A>G), c.571-8A>G, c.590A>C (p.K197T), c.598G>A (p.V200M), c.607T>G (p.W203G), c.611A>G(p.H204R), c.613delC, c.615delG (p.L205fs), c.621T>G (p.D207E), c.623A>T (p.D208V),c.624_627delTCCT (p.D208fs), c.629C>T (p.S210F), c.632T>C (p.F211S), c.736G>A (p.A246T),c.749G>A (p.G250D), c.778C>T (p.P260S), c.78G>A (p.W26X), c.796T>G (p.W266G), c.805+1G>A, c.805+1G>C, c.805+2T>C, c.805G>A (p.G269S), c.910_912delTTC (p.305delF), c.947_948insA (p.Y316fs), c.964G>A (p.D322N), c.964G>T (p.D322Y)

Trichohepatoenteric Syndrome: Type 1 (TTC37): (9): c.2578-7delTTTTT, c.1632+1delG, c.2251C>T (p.Q751X), c.2515+1G>C, c.2808G>A p.W936X), c.3847G>A (p.D1283N), c.439C>T (p.Q147X), c.4620+1G>C, c.751G>A (p.G251R)

Tyrosine Hydroxylase Deficiency (TH): (1): c.698G>A (p.R233H)

Tyrosinemia: Type I (FAH): (10): c.1009G>A (p.G337S), c.1062+5G>A, c.1069G>T (p.E357X), c.192G>T (p.Q64H), c.554-1G>T, c.607-6T>G, c.698A>T (p.D233V), c.707-1G>C, c.782C>T (p.P261L), c.786G>A (p.W262X)

Tyrosinemia: Type II (TAT): (5): c.1085G>T (p.G362V), c.1249C>T (p.R417X), c.169C>T (p.R57X), c.236-5A>G, c.668C>G (p.S223X)

Usher Syndrome: Type 1B (MYO7A): (13): c.1190C>A(p.A397D), c.1797G>A (p.M599I), c.1996C>T (p.R666X), c.2476G>A (p.A826T), c.3719G>A(p.R1240Q), c.448C>T (p.R150X), c.5581C>T (p.R1861X), c.6025delG (p.A2009fs), c.634C>T (p.R212C), c.635G>A (p.R212H), c.640G>A (p.G214R), c.700C>T (p.Q234X), c.93C>A (p.C31X)

Usher Syndrome: Type 1C (USH1C): (6): c.216G>A (p.V72fs), c.238_239insC, c.36+1G>T, c.496+1G>A, c.496+1G>T, c.91C>T (p.R31X)

Usher Syndrome: Type 1D (CDH23): (14): c.172C>T (p.Q58X), c.3367C>T (p.Q1123X), c.3617C>G (p.P1206R), c.3713_3714delCT (p.S1238fs), c.3880C>T(p.Q1294X), c.4069C>T (p.Q1357X), c.4488G>C (p.Q1496H), c.4504C>T (p.R1502X), c.5237G>A (p.R1746Q), c.5985C>A (p.Y1995X), c.6307G>T (p.E2103X), c.7549A>G (p.S2517G), c.8230G>A (p.G2744S), c.8497C>G (p.R2833G)

Usher Syndrome: Type 1F (PCDH15): (7): c.1101delT (p.A367fsX), c.1942C>T (p.R648X), c.2067C>A (p.Y684X), c.2800C>T (p.R934X), c.4272delA (p.L1425fs), c.733C>T (p.R245X), c.7C>T (p.R3X)

Usher Syndrome: Type 2A (USH2A): (23): c.1000C>T(p.R334W), c.11328T>A (p.Y3776X), c.11328T>G (p.Y3776X), c.12067-2A>G, c.1256G>T(p.C419F), c.12708T>A (p.C4236X), c.13576C>T (p.R4526X), c.14020A>G (p.R4674G), c.14403C>G (p.Y4801X), c.1840+1G>A, c.1876C>T (p.R626X), c.2209C>T (p.R737X), c.2276G>T (p.C759F), c.2299delG (p.E767SfsX21), c.3788G>A (p.W1263X), c.4338_4339delCT (p.C1447fs), c.5329C>T (p.R1777W), c.6235A>T (p.K2079X), c.7123delG(p.G2375fs), c.9165_9168delCTAT (p.I3055MfsX2), c.923_924insGCCA (p.H308fs),c.9469C>T (p.Q3157X), c.9492_9498delTGATGAG (p.D3165fs)

Usher Syndrome: Type 3 (CLRN1): (5): c.131T>A (p.M120K), c.144T>G (p.N48K), c.221T>C (p.L74P), c.567T>G (p.Y189X), c.634C>T (p.Q212X)

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL): (29): c.1144A>C (p.K382Q), c.1226C>T (p.T409M), c.1246G>A (p.A416T), c.1322G>A(p.G441D), c.1349G>A (p.R450H), c.1358G>A (p.R453Q), c.1372T>C (p.F458L), c.1405C>T

(p.R469W), c.1512G>T (p.E504D), c.1531C>T (p.R511W), c.1606_1609delGCAG (p.A536fs),c.1837C>T (p.R613W), c.265C>T (p.P89S), c.272C>A (p.P91Q), c.364A>G (p.N122D), c.37C>T(p.Q13X), c.388_391delGAGA (p.E130fs), c.520G>A (p.V174M), c.553G>A (p.G185S),c.577G>C (p.G193R), c.664G>A (p.G222R), c.685C>T (p.R229X), c.739A>C (p.K247Q),c.753-2A>C (IVS8-2A>C), c.779C>T (p.T260M), c.790A>G (p.K264E), c.848T>C (p.V283A),c.856A>G (p.R286G), c.881G>A (p.G294E)

Walker-Warburg Syndrome (FKTN): (5): c.1167insA (p.F390fs), c.139C>T (p.R47X), c.515A>G (p.H172R), c.648-1243G>T (IVS5-1243G>T), c.748T>G (p.C250G)

Werner Syndrome (WRN): (8): c.1336C>T (p.R368X), c.1730A>T (p.K577M), c.2089-3024A>G, c.3139-1G>C (IVS25-1G>C), c.3493C>T (p.Q1165X), c.3686A>T (p.Q1229L), c.3913C>T (p.R1305X), c.3915_3916insA (p.R1306fs)

Wilson Disease (ATP7B): (17): c.-370_-394delTGGCCGAGACCGCGG, c.1340_1343delAAAC, c.1934T>G (p.M645R), c.2123T>C (p.L708P), c.2293G>A (p.D765N), c.2304delC (p.M769Cfs), c.2332C>G (p.R778G), c.2333G>T (p.R778L), c.2336G>A (p.W779X), c.2337G>A (p.W779X), c.2906G>A(p.R969Q), c.3191A>C (p.E1064A), c.3207C>A (p.H1069Q), c.3683G>C (p.R1228T),c.3809A>G (p.N1270S), c.3817C>T (p.P1273S), c.845delT (p.L282Pfs)

Wiskott-Aldrich Syndrome (WAS): (6): c.100C>T (p.R34X), c.1097delG (p.G366fs), c.177delT (p.P59fs), c.257G>T (p.R86L), c.505+2T>G, c.560-1G>A

Wolcott-Rallison Syndrome (EIF2AK3): (5): c.1047_1060delAGTCATTCCCATCA (p.V350Sfs), c.1262delA (p.N421fs), c.1409C>G (p.S470X), c.1570delGAAA (p.E524fsX), c.478delG (p.A160fs)

Wolman Disease (LIPA): (3): c.260G>T (p.G87V), c.419G>A (p.W140X), c.964C>T (p.Q322X)

Xeroderma Pigmentosum: Group A (XPA): (7): c.172+2T>G, c.323G>T (p.C108F), c.348T>A (p.Y116X), c.374delC (p.T125fs), c.390-1G>C, c.619C>T (p.R207X), c.682C>T (p.R228X)

Xeroderma Pigmentosum: Group C (XPC): (5): c.1643_1644delTG (p.V548fs), c.1735C>T (p.R579X), c.413-24A>G, c.413-9T>A, c.566_567delAT (p.Y189fs)

Zellweger Spectrum Disorders: PEX1 Related (PEX1): (3): c.2097insT (p.I700fs), c.2528G>A (p.G843D), c.2916delA (p.G973fs)

Zellweger Spectrum Disorders: PEX10 Related (PEX10): (2) c.764_765insA, c.874_875delCT

Zellweger Spectrum Disorders: PEX2 Related (PEX2): (1): c.355C>T (p.R119X)

Zellweger Spectrum Disorders: PEX6 Related (PEX6): (8): c.1130+1G>A (IVS3+1G>A), c.1301delC (p.S434Ffs), c.1601T>C (p.L534P), c.1688+1G>A (IVS7+1G>A), c.1715C>T (p.T572I), c.1962-1G>A (p.L655fsX3), c.511insT (p.G171Wfs), c.802_815delGACGGACTGGCGCT (p.D268Cfs)