Clínica Victoria Rey

Listado de enfermedades que analiza el este Test de Compatibilidad o Matching Genético.

Cholestasis, progressive familial intrahepatic 2 - OMIM: 601847 - Gene: ABCB11 - Carrier frequency 1/159 - Coding region of NM_003742 (28)

Adrenoleukodystrophy - OMIM: 300100 - Gene: ABCD1 - Carrier frequency 1/10001 - Coding region of NM_000033 (10)

Methylmalonic aciduria and homocystinuria, cblJ type - OMIM: 614857 - Gene: ABCD4 - Carrier frequency 1/501 - Coding region of NM_005050 (19), NR_003256 (18)

Acyl-CoA dehydrogenase, medium chain, deficiency of - OMIM: 201450 - Gene: ACADM - Carrier frequency 1/62 - Coding region of NM_000016 (12), NM_001127328 (12), NM_001286042 (11), NM_001286043 (13), NM_001286044 (9)

Acyl-CoA dehydrogenase, short-chain, deficiency of - OMIM: 201470 - Gene: ACADS - Carrier frequency 1/113 - Coding region of NM_000017 (10), NM_001302554 (10)

2-methylbutyrylglycinuria - OMIM: 610006 - Gene: ACADSB - Carrier frequency 1/501 - Coding region of NM_001330174 (10), NM_001609 (11)

VLCAD deficiency - OMIM: 201475 - Gene: ACADVL - Carrier frequency 1/138 - Coding region of NM_000018 (20), NM_001033859 (19), NM_001270447 (21), NM_001270448 (19)

Alpha-methylacetoacetic aciduria - OMIM: 203750 - Gene: ACAT1 - Carrier frequency 1/216 - Coding region of NM_000019 (12)

Peroxisomal acyl-CoA oxidase deficiency - OMIM: 264470 - Gene: ACOX1 - Carrier frequency 1/501 - Coding region of NM_001185039 (14), NM_004035 (14), NM_007292 (14)

Combined malonic and methylmalonic acidemia - OMIM: 614265 - Gene: ACSF3 - Carrier frequency 1/501 - Coding region of NM_001127214 (10), NM_001243279 (11), NM_001284316 (9), NM_174917 (11), NR_045667 (5), NR_104293 (11)

Mental retardation, X-linked 63 - OMIM: 300387 - Gene: ACSL4 - Carrier frequency 1/45001* - Coding region of NM_001318509 (16), NM_001318510 (16), NM_004458 (16), NM_022977 (17)

Severe combined immunodeficiency due to ADA deficiency - OMIM: 102700 - Gene: ADA - Carrier frequency 1/388 - Coding region of NM_000022 (12), NM_001322050 (11), NM_001322051 (11), NR_136160 (11)

Hypermethioninemia due to adenosine kinase deficiency - OMIM: 614300 - Gene: ADK - Carrier frequency 1/501 - Coding region of NM_001123 (11), NM_001202449 (10), NM_001202450 (10), NM_006721 (11)

Mental retardation, X-linked, FRAXE type - OMIM: 309548 - Gene: AFF2 - Carrier frequency 1/62501 - Coding region of NM_001169122 (20), NM_001169123 (21), NM_001169124 (20), NM_001169125 (20), NM_001170628 (18), NM_002025 (21)

Aspartylglucosaminuria - OMIM: 208400 - Gene: AGA - Carrier frequency 1/1119 - Coding region of NM_000027 (9), NM_001171988 (9), NR_033655 (8)

Glycogen storage disease IIIa - OMIM: 232400 - Gene: AGL - Carrier frequency 1/159 - Coding region of NM_000028 (34), NM_000642 (34), NM_000643 (34), NM_000644 (34), NM_000646 (34)

Rhizomelic chondrodysplasia punctata, type 3 - OMIM: 600121 - Gene: AGPS - Carrier frequency 1/159 - Coding region of NM_003659 (20) Hyperoxaluria, primary, type 1 - OMIM: 259900 - Gene: AGXT - Carrier frequency 1/409 - Coding region of NM_000030 (11)

Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase - OMIM: 613752 - Gene: AHCY - Carrier frequency 1/708 - Coding region of NM_000687 (10), NM_001161766 (10), NM_001322084 (10), NM_001322085 (10), NM_001322086 (10)

Joubert syndrome 3 - OMIM: 608629 - Gene: AHI1 - Carrier frequency 1/501 - Coding region of NM_001134830 (27), NM_001134831 (29), NM_001134832 (23), NM_017651 (28)

Leber congenital amaurosis 4 - OMIM: 604393 - Gene: AIPL1 - Carrier frequency 1/417 - Coding region of NM_001033054 (5), NM_001033055 (5), NM_001285399 (6), NM_001285400 (6), NM_001285401 (6), NM_001285402 (6), NM_001285403 (5), NM_014336 (6)

Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia - OMIM: 240300 - Gene: AIRE - Carrier frequency 1/355 - Coding region of NM_-000383 (14)

Hyperprolinemia, type II - OMIM: 239510 - Gene: ALDH4A1 - Carrier frequency 1/501 - Coding region of NM_001161504 (15), NM_001319218 (14), NM_003748 (15), NM_170726 (16)

Fructose intolerance - OMIM: 229600 - Gene: ALDOB - Carrier frequency 1/72 - Coding region of NM_000035 (9)

Glycine encephalopathy - OMIM: 605899 - Gene: AMT - Carrier frequency 1/501 - Coding region of NM_000481 (9), NM_001164710 (8), NM_001164711 (8), NM_001164712 (10), NR_028435 (9)

Mental retardation, X-linked syndromic 5 - OMIM: 304340 - Gene: AP1S2 - Carrier frequency 1/500001 - Coding region of NM_001272071 (6), NM_003916 (5)

Androgen insensitivity - OMIM: 300068 - Gene: AR - Carrier frequency 1/20001 - Coding region of NM_000044 (8), NM_001011645 (8)

Argininemia - OMIM: 207800 - Gene: ARG1 - Carrier frequency 1/501 - Coding region of NM_000045 (8), NM_001244438 (8)

Joubert syndrome 8 - OMIM: 612291 - Gene: ARL13B - Carrier frequency 1/159 - Coding region of NM_001174150 (10), NM_001174151 (9), NM_001321328 (11), NM_144996 (8), NM_182896 (11), NR_033427 (9), NR_135621 (9)

Metachromatic leukodystrophy - OMIM: 250100 - Gene: ARSA - Carrier frequency 1/113 - Coding region of NM_000487 (8), NM_001085425 (9), NM_001085426 (9), NM_001085427 (9), NM_001085428 (8)

Mucopolysaccharidosis type VI (Maroteaux-Lamy) - OMIM: 253200 - Gene: ARSB - Carrier frequency 1/159 - Coding region of NM_000046 (8), NM_198709 (8)

Epileptic encephalopathy, early infantile, 1 - OMIM: 308350 - Gene: ARX - Carrier frequency 1/50001 - Coding region of NM_139058 (5)

Argininosuccinic aciduria - OMIM: 207900 - Gene: ASL - Carrier frequency 1/195 - Coding region of NM_000048 (17), NM_001024943 (16), NM_001024944 (15), NM_001024946 (15)

Canavan disease - OMIM: 271900 - Gene: ASPA - Carrier frequency 1/159 - Coding region of NM_000049 (6), NM_001128085 (7)

Citrullinemia - OMIM: 215700 - Gene: ASS1 - Carrier frequency 1/120 - Coding region of NM_000050 (16), NM_054012 (15)

Ataxia-telangiectasia - OMIM: 208900 - Gene: ATM - Carrier frequency 1/159 - Coding region of NM_000051 (63) Wilson disease - OMIM: 277900 - Gene: ATP7B - Carrier frequency 1/80 - Coding region of NM_000053 (21), NM_001005918 (17), NM_001243182 (22), NM_001330578 (20), NM_001330579 (19)

Alpha-thalassemia/mental retardation syndrome - OMIM: 301040 - Gene: ATRX - Carrier frequency 1/500001 - Coding region of NM_000489 (35), NM_138270 (34)

Bardet-Biedl syndrome 1 - OMIM: 209900 - Gene: BBS1 - Carrier frequency 1/195* - Coding region of NM_024649 (17)

Bardet-Biedl syndrome 10 - OMIM: 615987 - Gene: BBS10 - Carrier frequency 1/195* - Coding region of NM_024685 (2)

Bardet-Biedl syndrome 2 - OMIM: 615981 - Gene: BBS2 - Carrier frequency 1/195* - Coding region of NM_031885 (17)

Maple syrup urine disease, type Ia - OMIM: 248600 - Gene: BCKDHA - Carrier frequency 1/195 - Coding region of NM_000709 (9), NM_001164783 (9)

Maple syrup urine disease, type Ib - OMIM: 248600 - Gene: BCKDHB - Carrier frequency 1/195 - Coding region of NM_000056 (11), NM_001318975 (10), NM_183050 (10), NR_134945 (11)

Bjornstad syndrome - OMIM: 262000 - Gene: BCS1L - Carrier frequency 1/501 - Coding region of NM_001079866 (8), NM_001257342 (9), NM_001257343 (9), NM_001257344 (8), NM_001318836 (7), NM_001320717 (9), NM_004328 (9)

X-linked mental retardation (XLMR) associated with macrocephaly - OMIM: 300659 - Gene: BRWD3 - Carrier frequency 1/45001 - Coding region of NM_153252 (41) Bartter syndrome, type 4a - OMIM: 602522 - Gene: BSND - Carrier frequency 1/647 - Coding region of NM_057176 (4)

Biotinidase deficiency - OMIM: 253260 - Gene: BTD - Carrier frequency 1/113 - Coding region of NM_000060 (4), NM_001281723 (4), NM_001281724 (6), NM_001281725 (4), NM_001281726 (3), NM_001323582 (5)

Muscular dystrophy, limb-girdle, 2A - OMIM: 253600 - Gene: CAPN3 - Carrier frequency 1/159* - Coding region of NM_000070 (24), NM_024344 (23), NM_173087 (21), NM_173088 (13), NM_173089 (9), NM_173090 (10)

Mental retardation and microcephaly with pontine and cerebellar hypoplasia - OMIM: 300749 - Gene: CASK - Carrier frequency 1/500001 - Coding region of NM_001126054 (26), NM_001126055 (25), NM_003688 (27)

Ventricular tachycardia, catecholaminergic polymorphic, 2 - OMIM: 611938 - Gene: CASQ2 - Carrier frequency 1/51 - Coding region of NM_001232 (11)

Homocystinuria, B6-responsive and nonresponsive types - OMIM: 236200 - Gene: CBS - Carrier frequency 1/124 - Coding region of NM_000071 (17), NM_001178008 (17), NM_001178009 (18), NM_001320298 (18), NM_001321072 (14)

Immunodeficiency, X-linked, with hyper-IgM - OMIM: 308230 - Gene: CD40LG - Carrier frequency 1/250001 - Coding region of NM_000074 (5)

Deafness, autosomal recessive 12 - OMIM: 601386 - Gene: CDH23 - Carrier frequency 1/73 - Coding region of NM_001171930 (31), NM_001171931 (25), NM_001171932 (11), NM_001171933 (23), NM_001171934 (22), NM_001171935 (5), NM_001171936 (4), NM_022124 (68), NM_052836 (13)

Bardet-Biedl syndrome 14; Joubert syndrome 5; Meckel syndrome 4; Senior-Loken syndrome 6 - OMIM: 615991; 610188; 611134; 610189 - Gene: CEP290 - Carrier frequency 1/501 - Coding region of NM_025114 (54)

Retinitis pigmentosa 26 - OMIM: 608380 - Gene: CERKL - Carrier frequency 1/409 - Coding region of NM_001030311 (14), NM_001030312 (10), NM_001030313 (11), NM_001160277 (13), NM_201548 (13), NR_027689 (11), NR_027690 (12)

Cystic Fibrosis; Congenital bilateral absence of vas deferens - OMIM: 219700; 277180 - Gene: CFTR - Carrier frequency 1/36 - Coding region of NM_000492 (27)

Macular corneal dystrophy - OMIM: 217800 - Gene: CHST6 - Carrier frequency 1/72 - Coding region of NM_021615 (3)

Myotonia congenita, recessive (Becker disease); Myotonia congenita, dominant (Thomsen disease) - OMIM: 160800; 255700 - Gene: CLCN1 - Carrier frequency 1/159 - Coding region of NM_000083 (23), NR_046453 (22)

Ceroid lipofuscinosis, neuronal, 3 - OMIM: 204200 - Gene: CLN3 - Carrier frequency 1/409 - Coding region of NM_000086 (15), NM_001042432 (16), NM_001286104 (15), NM_001286105 (13), NM_001286109 (14), NM_001286110 (14)

Ceroid lipofuscinosis, neuronal, 5 - OMIM: 256731 - Gene: CLN5 - Carrier frequency 1/501* - Coding region of NM_006493 (4)

Ceroid lipofuscinosis, neuronal, 6 - OMIM: 601780 - Gene: CLN6 - Carrier frequency 1/501* - Coding region of NM_017882 (7)

Ceroid lipofuscinosis, neuronal, 8 - OMIM: 600143 - Gene: CLN8 - Carrier frequency 1/501* - Coding region of NM_018941 (3)

Usher syndrome, type 3A - OMIM: 276902 - Gene: CLRN1 - Carrier frequency 1/538 - Coding region of NM_001195794 (4), NM_001256819 (4), NM_052995 (4), NM_174878 (3), NR_046380 (5)

Retinitis pigmentosa 49 - OMIM: 613756 - Gene: CNGA1 - Carrier frequency 1/625 - Coding region of NM_000087 (11), NM_001142564 (10)

Retinitis pigmentosa 45 - OMIM: 613767 - Gene: CNGB1 - Carrier frequency 1/867 - Coding region of NM_001135639 (13), NM_001286130 (33), NM_001297 (33)

Achromatopsia 3 - OMIM: 262300 - Gene: CNGB3 - Carrier frequency 1/101 - Coding region of NM_019098 (18)

Alport syndrome, autosomal recessive - OMIM: 203780 - Gene: COL4A4 - Carrier frequency 1/113 - Coding region of NM_000092 (48)

Epidermolysis bullosa dystrophica, AR - OMIM: 226600 - Gene: COL7A1 - Carrier frequency 1/708 - Coding region of NM_000094 (118)

Carbamoylphosphate synthetase I deficiency - OMIM: 237300 - Gene: CPS1 - Carrier frequency 1/501 - Coding region of NM_001122633 (39), NM_001122634 (28), NM_001875 (38)

CPT deficiency, hepatic, type IA - OMIM: 255120 - Gene: CPT1A - Carrier frequency 1/708 - Coding region of NM_001031847 (19), NM_001876 (19)

CPT II deficiency, lethal neonatal - OMIM: 608836 - Gene: CPT2 - Carrier frequency 1/708 - Coding region of NM_000098 (5), NM_001330589 (5)

Leber congenital amaurosis 8 - OMIM: 613835 - Gene: CRB1 - Carrier frequency 1/832 - Coding region of NM_001193640 (10), NM_001257965 (15), NM_001257966 (10), NM_201253 (12), NR_047563 (13), NR_047564 (12)

Cystathioninuria - OMIM: 219500 - Gene: CTH - Carrier frequency 1/80 - Coding region of NM_001190463 (11), NM_001902 (12), NM_153742 (11)

Cystinosis, nephropathic - OMIM: 219800 - Gene: CTNS - Carrier frequency 1/225 - Coding region of NM_001031681 (13), NM_004937 (12)

Ceroid lipofuscinosis, neuronal, 10 - OMIM: 610127 - Gene: CTSD - Carrier frequency 1/501 - Coding region of NM_001909 (9)

Mental retardation, X-linked, syndromic 15 - OMIM: 300354 - Gene: CUL4B - Carrier frequency 1/500001 - Coding region of NM_001079872 (20), NM_001330624 (21), NM_003588 (22)

Adrenal hyperplasia, congenital, due to 17-alpha-hydroxylase deficiency - OMIM: 202110 - Gene: CYP17A1 - Carrier frequency 1/501 - Coding region of NM_000102 (8)

Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency - OMIM: 201910 - Gene: CYP21A2 - Carrier frequency 1/64 - NM_000500: Whole del; p.I173N; p.Q319X; c.293-13C>G; p.G293S; c.332_339delGAGACTAC

Cerebrotendinous xanthomatosis - OMIM: 213700 - Gene: CYP27A1 - Carrier frequency 1/113 - Coding region of NM_000784 (9)

Maple syrup urine disease, type II - OMIM: 248600 - Gene: DBT - Carrier frequency 1/216 - Coding region of NM_001918 (11)

Lissencephaly, X-linked - OMIM: 300067 - Gene: DCX - Carrier frequency 1/50001 - Coding region of NM_000555 (7), NM_001195553 (7), NM_178151 (7), NM_178152 (7), NM_178153 (7)

Smith-Lemli-Opitz syndrome - OMIM: 270400 - Gene: DHCR7 - Carrier frequency 1/88 - Coding region of NM_001163817 (9), NM_001360 (9)

Retinitis pigmentosa 59 - OMIM: 613861 - Gene: DHDDS - Carrier frequency 1/867 - Coding region of NM_001243564 (8), NM_001243565 (8), NM_001319959 (9), NM_024887 (9), NM_205861 (9)

Maple syrup urine disease, type III (Dihydrolipoamide dehydrogenase deficiency) - OMIM: 246900 - Gene: DLD - Carrier frequency 1/216 - Coding region of NM_000108 (14), NM_-001289750 (12), NM_001289751 (13), NM_001289752 (13)

Mental retardation, X-linked 90 - OMIM: 300850 - Gene: DLG3 - Carrier frequency 1/45001 - Coding region of NM_001166278 (12), NM_020730 (14), NM_021120 (19)

Duchenne muscular dystrophy; Becker muscular dystrophy - OMIM: 310200; 300376 - Gene: DMD - Carrier frequency 1/1651 - Coding region of NM_000109 (79), NM_004006 (79), NM_-004009 (79), NM_004010 (79), NM_004011 (51), NM_004012 (51), NM_004013 (36), NM_004014 (25), NM_004015 (18), NM_004016 (17), NM_004017 (17), NM_004018 (16), NM_004019 (9), NM_004020 (32), NM_004021 (35), NM_004022 (34), NM_004023 (31)

Primary ciliary dyskinesia - OMIM: 608644 - Gene: DNAH5 - Carrier frequency 1/62 - Coding region of NM_001369 (79)

Thryoid dyshormonogenesis 6 - OMIM: 607200 - Gene: DUOX2 - Carrier frequency 1/72* - Coding region of NM_014080 (34)

Thyroid dyshormonogenesis 5 - OMIM: 274900 - Gene: DUOXA2 - Carrier frequency 1/72* - Coding region of NM_207581 (6)

Limb-girdle muscular dystrophy type 2B - OMIM: 253601 - Gene: DYSF - Carrier frequency 1/501* - Coding region of NM_001130455 (55), NM_001130976 (54), NM_001130977 (55), NM_- 001130978 (56), NM_001130979 (56), NM_001130980 (55), NM_001130981 (56), NM_001130982 (56), NM_001130983 (56), NM_001130984 (55), NM_001130985 (55), NM_001130986 (54), NM_001130987 (56), NM_003494 (55)

Glutaric acidemia IIA - OMIM: 231680 - Gene: ETFA - Carrier frequency 1/708* - Coding region of NM_000126 (12), NM_001127716 (11) Glutaric acidemia IIB - OMIM: 231680 - Gene: ETFB - Carrier frequency 1/708* - Coding region of NM_001014763 (5), NM_001985 (6)

Glutaric acidemia IIC - OMIM: 231680 - Gene: ETFDH - Carrier frequency 1/708* - Coding region of NM_001281737 (12), NM_001281738 (11), NM_004453 (13)

Ethylmalonic encephalopathy - OMIM: 602473 - Gene: ETHE1 - Carrier frequency 1/970 - Coding region of NM_001320867 (7), NM_001320868 (6), NM_001320869 (5), NM_014297 (7)

Ellis-van Creveld syndrome - OMIM: 225500 - Gene: EVC2 - Carrier frequency 1/188* - Coding region of NM_001166136 (22), NM_147127 (22)

Retinitis pigmentosa 25 - OMIM: 602772 - Gene: EYS - Carrier frequency 1/140 - Coding region of NM_001142800 (43), NM_001142801 (12), NM_001292009 (44), NM_198283 (10)

Factor XI deficiency - OMIM: 612416 - Gene: F11 - Carrier frequency 1/501 - Coding region of NM_000128 (15)

Prothrombin deficiency, congenital - OMIM: 613679 - Gene: F2 - Carrier frequency 1/708 - Coding region of NM_000506 (14), NM_001311257 (14)

Factor V deficiency - OMIM: 227400 - Gene: F5 - Carrier frequency 1/501 - Coding region of NM_000130 (25) Hemophilia A - OMIM: 306700 - Gene: F8 - Carrier frequency 1/3001 - Coding region of NM_000132 (26), NM_019863 (5)

Hemophilia B - OMIM: 306900 - Gene: F9 - Carrier frequency 1/15001 - Coding region of NM_000133 (8), NM_001313913 (7)

Tyrosinemia, type I - OMIM: 276700 - Gene: FAH - Carrier frequency 1/167 - Coding region of NM_000137 (14)

Fanconi anemia - OMIM: 227650 - Gene: FANCA - Carrier frequency 1/249 - Coding region of NM_000135 (43), NM_001018112 (11), NM_001286167 (43)

Fanconi anemia, complementation group C - OMIM: 227645 - Gene: FANCC - Carrier frequency 1/535 - Coding region of NM_000136 (15), NM_001243743 (15), NM_001243744 (14)

Aarskog-Scott syndrome; Mental retardation, X-linked 16 - OMIM: 305400 - Gene: FGD1 - Carrier frequency 1/500001 - Coding region of NM_004463 (18)

Fumarase deficiency - OMIM: 606812 - Gene: FH - Carrier frequency 1/614 - Coding region of NM_000143 (10)

Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 - OMIM: 613153 - Gene: FKRP - Carrier frequency 1/708* - Coding region of NM_001039885 (4), NM_024301 (4)

Walker-Warburg syndrome, Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 - OMIM: 253800 - Gene: FKTN - Carrier frequency 1/355* - Coding region of NM_001079802 (11), NM_001198963 (12), NM_006731 (10)

Fragile X syndrome - OMIM: 300624 - Gene: FMR1 - Carrier frequency full mutation 1/2500; premutation 1/251 - NM_001185076: (CGG)n expansion

Glutamate formiminotransferase deficiency - OMIM: 229100 - Gene: FTCD - Carrier frequency 1/355 - Coding region of NM_001320412 (15), NM_006657 (15), NM_206965 (14)

Mental retardation, X-linked 9 - OMIM: 309549 - Gene: FTSJ1 - Carrier frequency 1/45001 - Coding region of NM_001282157 (9), NM_012280 (13), NM_177439 (14)

Friedreich ataxia with retained reflexes - OMIM: 229300 - Gene: FXN - Carrier frequency 1/75 - Coding region of NM_000144 (5), NM_001161706 (5), NM_181425 (5)

Glycogen storage disease Ia - OMIM: 232200 - Gene: G6PC - Carrier frequency 1/159 - Coding region of NM_000151 (5), NM_001270397 (5)

Hemolytic anemia, G6PD deficient (favism) - OMIM: 300908 - Gene: G6PD - Carrier frequency 1/51 - Coding region of NM_000402 (13), NM_001042351 (13)

Glycogen storage disease II (Pompe disease) - OMIM: 232300 - Gene: GAA - Carrier frequency 1/159 - Coding region of NM_000152 (20), NM_001079803 (21), NM_001079804 (20)

Krabbe disease - OMIM: 245200 - Gene: GALC - Carrier frequency 1/159 - Coding region of NM_000153 (17), NM_001201401 (16), NM_001201402 (17)

Galactose epimerase deficiency - OMIM: 230350 - Gene: GALE - Carrier frequency 1/159 - Coding region of NM_000403 (12), NM_001008216 (12), NM_001127621 (11)

Galactokinase deficiency with cataracts - OMIM: 230200 - Gene: GALK1 - Carrier frequency 1/225 - Coding region of NM_000154 (8)

Mucopolysaccharidosis IVA - OMIM: 253000 - Gene: GALNS - Carrier frequency 1/275 - Coding region of NM_000512 (14), NM_001323543 (13), NM_001323544 (15)

Galactosemia - OMIM: 230400 - Gene: GALT - Carrier frequency 1/113 - Coding region of NM_000155 (11), NM_001258332 (9)

Gaucher disease, perinatal lethal; Gaucher disease, type II; Gaucher disease, type III; Gaucher disease, type IIIC - OMIM: 608013; 230900; 231000; 231005 - Gene: GBA - Carrier frequency 1/159 - Coding region of NM_000157 (11), NM_001005741 (12), NM_001005742 (12), NM_001171811 (10), NM_001171812 (10)

Glycogen storage disease IV - OMIM: 232500 - Gene: GBE1 - Carrier frequency 1/501 - Coding region of NM_000158 (16)

Glutaric aciduria, type I - OMIM: 231670 - Gene: GCDH - Carrier frequency 1/95 - Coding region of NM_000159 (12), NM_013976 (12), NR_102316 (12), NR_102317 (11)

Charcot-Marie-Tooth Neuropathy Type 4A - OMIM: 214400 - Gene: GDAP1 - Carrier frequency 1/130 - Coding region of NM_001040875 (6), NM_018972 (6), NR_046346 (6)

Mental retardation, X-linked 41 - OMIM: 300849 - Gene: GDI1 - Carrier frequency 1/45001 - Coding region of NM_001493 (11)

Deafness, autosomal recessive 1A (DFNB1-A); Deafness, digenic (GJB2/GJB3, GJB2/GJB6) - OMIM: 220290 - Gene: GJB2 - Carrier frequency 1/43 - Coding region of NM_004004 (2)

Deafness, digenic, GJB2/GJB3 - OMIM: 220290 - Gene: GJB3 - Carrier frequency 1/159 - Coding region of NM_001005752 (2), NM_024009 (2)

Deafness, autosomal recessive 1B (DFNB1B); Deafness, digenic GJB2/GJB6 - OMIM: 612645; 220290 - Gene: GJB6 - Carrier frequency 1/39 - Coding region of NM_001110221 (3), NM_-001110220 (4), NM_001110219 (5), NM_006783 (3)

Fabry disease - OMIM: 301500 - Gene: GLA - Carrier frequency 1/1501 - Coding region of NM_000169 (7) GM1-gangliosidosis, type I; GM1-gangliosidosis, type II;

GM1-gangliosidosis, type III - OMIM: 230500; 230600; 230650 - Gene: GLB1 - Carrier frequency 1/195 - Coding region of NM_- 000404 (16), NM_001079811 (16), NM_001135602 (13), NM_001317040 (17)

Glycine encephalopathy - OMIM: 605899 - Gene: GLDC - Carrier frequency 1/501 - Coding region of NM_000170 (25)

Glycine N-methyltransferase deficiency - OMIM: 606664 - Gene: GNMT - Carrier frequency 1/501 - Coding region of NM_001318865 (6), NM_018960 (6), NR_134899 (5)

Mucolipidosis II , alpha/beta; Mucolipidosis III, alpha/beta - OMIM: 252500; 252600 - Gene: GNPTAB - Carrier frequency 1/251 - Coding region of NM_024312 (21) Hypogonadotropic hypogonadism 7 without anosmia - OMIM: 146110 - Gene: GNRHR - Carrier frequency 1/501 - Coding region of NM_000406 (3), NM_001012763 (3)

Mucopolysaccharidosis type IIID (Sanfilippo syndrome, type D) - OMIM: 252940 - Gene: GNS - Carrier frequency 1/501 - Coding region of NM_002076 (14)

Nystagmus 6, congenital, X-linked - OMIM: 300814 - Gene: GPR143 - Carrier frequency 1/30001 - Coding region of NM_000273 (9)

Hyperoxaluria, primary, type II - OMIM: 260000 - Gene: GRHPR - Carrier frequency 1/708 - Coding region of NM_012203 (9)

Mental retardation, X-linked 94 - OMIM: 300699 - Gene: GRIA3 - Carrier frequency 1/500001 - Coding region of NM_000828 (16), NM_001256743 (5), NM_007325 (16)

Leber congenital amaurosis 1 - OMIM: 204000 - Gene: GUCY2D - Carrier frequency 1/248 - Coding region of NM_000180 (20) LCHAD deficiency - OMIM: 609016 - Gene: HADHA - Carrier frequency 1/251 - Coding region of NM_000182 (20)

Trifunctional protein deficiency - OMIM: 609015 - Gene: HADHB - Carrier frequency 1/501 - Coding region of NM_000183 (16), NM_001281512 (15), NM_001281513 (17)

Histidinemia - OMIM: 235800 - Gene: HAL - Carrier frequency 1/72 - Coding region of NM_001258333 (20), NM_001258334 (20), NM_002108 (21)

Neutropenia, severe congenital 3, autosomal recessive - OMIM: 610738 - Gene: HAX1 - Carrier frequency 1/126 - Coding region of NM_001018837 (7), NM_006118 (7)

Thalassemias, alpha- - OMIM: 604131 - Gene: HBA1 - Carrier frequency 1/44 - Coding region of NM_000558 (3)

Thalassemias, alpha- - OMIM: 604131 - Gene: HBA2 - Carrier frequency 1/44 - Coding region of NM_000517 (3)

Beta thalassemia, Sickle-cell anemia and other hemoglobinopathies - OMIM: 613985 - Gene: HBB - Carrier frequency 1/23 - Coding region of NM_000518 (3)

Tay-Sachs (GM2-gangliosidosis, several forms) - OMIM: 272800 - Gene: HEXA - Carrier frequency 1/284 - Coding region of NM_000520 (14), NM_001318825 (14), NR_134869 (11)

Sandhoff disease, infantile, juvenile, and adult forms - OMIM: 268800 - Gene: HEXB - Carrier frequency 1/501 - Coding region of NM_000521 (14), NM_001292004 (14)

Hemochromatosis, type 2A (HFE2 Related) - OMIM: 602390 - Gene: HFE2 - Carrier frequency 1/1582 - Coding region of NM_001316767 (4), NM_145277 (3), NM_202004 (3), NM_213652 (2), NM_213653 (4)

Alkaptonuria - OMIM: 203500 - Gene: HGD - Carrier frequency 1/163 - Coding region of NM_000187 (14)

Mucopolysaccharidosis type IIIC (Sanfilippo C) - OMIM: 252930 - Gene: HGSNAT - Carrier frequency 1/501 - Coding region of NM_152419 (18)

Holocarboxylase synthetase deficiency - OMIM: 253270 - Gene: HLCS - Carrier frequency 1/225 - Coding region of NM_000411 (12), NM_001242784 (12), NM_001242785 (12)

HMG-CoA lyase deficiency - OMIM: 246450 - Gene: HMGCL - Carrier frequency 1/159 - Coding region of NM_000191 (9), NM_001166059 (7)

Hyperoxaluria, primary, type III - OMIM: 613616 - Gene: HOGA1 - Carrier frequency 1/708 - Coding region of NM_001134670 (3), NM_138413 (7)

17-beta-hydroxysteroid dehydrogenase X deficiency - OMIM: 300438 - Gene: HSD17B10 - Carrier frequency 1/500001 - Coding region of NM_001037811 (6), NM_004493 (6)

Mental retardation, X-linked syndromic, Turner type - OMIM: 300706 - Gene: HUWE1 - Carrier frequency 1/500001 - Coding region of NM_031407 (84)

Retinitis pigmentosa 46 - OMIM: 612572 - Gene:IDH3B - Carrier frequency 1/501 - Coding region of NM_001258384 (12), NM_001330763 (13), NM_006899 (12), NM_174855 (12), NR_136344 (13)

Mucopolysaccharidosis II (Hunter syndrome) - OMIM: 309900 - Gene: IDS - Carrier frequency 1/51510 - Coding region of NM_000202 (9), NM_001166550 (9), NM_006123 (8), NR_104128 (9)

Mucopolysaccharidosis Ih (Hurler syndrome) - OMIM: 607014 - Gene: IDUA - Carrier frequency 1/210 - Coding region of NM_000203 (14), NR_110313 (14)

Mental retardation, X-linked 21/34 - OMIM: 300143 - Gene: IL1RAPL1 - Carrier frequency 1/45001 - Coding region of NM_014271 (11)

Severe combined immunodeficiency, X-linked - OMIM: 300400 - Gene: IL2RG - Carrier frequency 1/100001 - Coding region of NM_000206 (8)

Mental retardation, X-linked 1/78 - OMIM: 309530 - Gene: IQSEC2 - Carrier frequency 1/45001 - Coding region of NM_001111125 (15), NM_001243197 (3), NM_015075 (14)

Isovaleric acidemia - OMIM: 243500 - Gene: IVD - Carrier frequency 1/159 - Coding region of NM_001159508 (11), NM_002225 (12)

Thyroid dyshormonogenesis 4 - OMIM: 274800 - Gene: IYD - Carrier frequency 1/159 - Coding region of NM_001164694 (6), NM_001164695 (6), NM_001318495 (5), NM_203395 (5), NR_-134655 (6)

SCID, autosomal recessive, T-negative/B-positive type - OMIM: 600802 - Gene: JAK3 - Carrier frequency 1/159 - Coding region of NM_000215 (24)

Hyperinsulinemic hypoglycemia, familial, Type 2 - OMIM: 601820 - Gene: KCNJ11 - Carrier frequency 1/501 - Coding region of NM_000525 (1), NM_001166290 (2)

Mental retardation, X-linked, syndromic, Claes-Jensen type - OMIM: 300534 - Gene: KDM5C - Carrier frequency 1/500001 - Coding region of NM_001146702 (24), NM_001282622 (26), NM_004187 (26) MASA syndrome / CRASH syndrome - OMIM: 303350 - Gene: L1CAM - Carrier frequency 1/15001 - Coding region of NM_000425 (28), NM_001143963 (26), NM_001278116 (29), NM_024003 (27)

Epidermolysis bullosa, junctional, Herlitz type; Epidermolysis bullosa, junctional, non-Herlitz type - OMIM: 226700; 226650 - Gene: LAMB3 - Carrier frequency 1/902 - Coding region of NM_000228 (23), NM_001017402 (22), NM_001127641 (23)

Epidermolysis bullosa, junctional, Herlitz type; non-Herlitz type - OMIM: 226700; 226650 - Gene: LAMC2 - Carrier frequency 1/2501 - Coding region of NM_005562 (23), NM_018891 (22)

Hypercholesterolemia, familial, recessive - OMIM: 143890 - Gene: LDLR - Carrier frequency 1/240 - Coding region of NM_000527 (18), NM_001195798 (18), NM_001195799 (17), NM_-001195800 (16), NM_001195803 (16)

Hypercholesterolemia, familial, autosomal recessive - OMIM: 603813 - Gene: LDLRAP1 - Carrier frequency 1/501 - Coding region of NM_015627 (9)

Wolman disease (lysosomal acid lipase deficiency) - OMIM: 278000 - Gene: LIPA - Carrier frequency 1/501 - Coding region of NM_000235 (10), NM_001127605 (10), NM_001288979 (8)

Methylmalonic aciduria and homocystinuria, cblF type - OMIM: 277380 - Gene: LMBRD1 - Carrier frequency 1/501 - Coding region of NM_018368 (16)

Leigh syndrome, French-Canadian type - OMIM: 220111 - Gene: LRPPRC - Carrier frequency 1/708 - Coding region of NM_133259 (38)

Mannosidosis, alpha-, types I and II - OMIM: 248500 - Gene: MAN2B1 - Carrier frequency 1/501 - Coding region of NM_000528 (24), NM_001173498 (24)

Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency - OMIM: 250850 - Gene: MAT1A - Carrier frequency 1/708 - Coding region of NM_000429 (9)

3-Methylcrotonyl-CoA carboxylase 1 deficiency - OMIM: 210200 - Gene: MCCC1 - Carrier frequency 1/113 - Coding region of NM_001293273 (17), NM_020166 (19), NR_120639 (16), NR_- 120640 (19)

3-Methylcrotonyl-CoA carboxylase 2 deficiency - OMIM: 210210 - Gene: MCCC2 - Carrier frequency 1/140 - Coding region of NM_022132 (17)

Methylmalonyl-CoA epimerase deficiency - OMIM: 251120 - Gene: MCEE - Carrier frequency 1/501 - Coding region of NM_032601 (3)

Mucolipidosis IV - OMIM: 252650 - Gene: MCOLN1 - Carrier frequency 1/101 - Coding region of NM_020533 (14)

Mental retardation, X-linked, syndromic 13 - OMIM: 300055 - Gene: MECP2 - Carrier frequency 1/500001 - Coding region of NM_001110792 (3), NM_001316337 (5), NM_004992 (4)

Familial Mediterranean fever, AR - OMIM: 249100 - Gene: MEFV - Carrier frequency 1/36 - Coding region of NM_000243 (10), NM_001198536 (9)

Ceroid lipofuscinosis, neuronal, 7 - OMIM: 610951 - Gene: MFSD8 - Carrier frequency 1/501 - Coding region of NM_152778 (13) Meckel syndrome 1 - OMIM: 249000 - Gene: MKS1 - Carrier frequency 1/501 - Coding region of NM_001165927 (18), NM_001321268 (17), NM_001321269 (17), NM_001330397 (16), NM_- 017777 (18)

Malonyl-CoA decarboxylase deficiency - OMIM: 248360 - Gene: MLYCD - Carrier frequency 1/501 - Coding region of NM_012213 (5)

Methylmalonic aciduria, vitamin B12-responsive - OMIM: 251100 - Gene: MMAA - Carrier frequency 1/275 - Coding region of NM_172250 (7)

Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type - OMIM: 251110 - Gene: MMAB - Carrier frequency 1/396 - Coding region of NM_052845 (9), NR_038118 (10)

Methylmalonic aciduria and homocystinuria, cblC type - OMIM: 277400 - Gene: MMACHC - Carrier frequency 1/501 - Coding region of NM_001330540 (4), NM_015506 (4)

Methylmalonic aciduria and homocystinuria, cblD type, variant 1, variant 2 - OMIM: 277410 - Gene: MMADHC - Carrier frequency 1/501 - Coding region of NM_015702 (8)

Thrombocytopenia, congenital amegakaryocytic - OMIM: 604498 - Gene: MPL - Carrier frequency 1/914 - Coding region of NM_005373 (12)

Homocystinuria due to MTHFR deficiency - OMIM: 236250 - Gene: MTHFR - Carrier frequency 1/1119 - Coding region of NM_001330358 (12), NM_005957 (12)

Charcot-Marie-Tooth disease, type 4B1 - OMIM: 601382 - Gene: MTMR2 - Carrier frequency 1/501 - Coding region of NM_001243571 (18), NM_016156 (15), NM_201278 (17), NM_201281 (16)

Homocystinuria-megaloblastic anemia, cbl E type - OMIM: 236270 - Gene: MTRR - Carrier frequency 1/501 - Coding region of NM_002454 (15), NM_024010 (15), NR_134480 (15), NR_134481 (14), NR_134482 (14)

Methylmalonic aciduria, mut(0) type - OMIM: 251000 - Gene: MUT - Carrier frequency 1/178 - Coding region of NM_000255 (13)

Mevalonic aciduria; Hyper-IgD syndrome - OMIM: 610377; 260920 - Gene: MVK - Carrier frequency 1/230 - Coding region of NM_000431 (11), NM_001114185 (11), NM_001301182 (10)

Usher syndrome, type 1B; Deafness, autosomal recessive 2 (DFNB2); Deafness, autosomal dominant 11 (DFNA11) - OMIM: 276900; 600060; 601317 - Gene: MYO7A - Carrier frequency 1/92 - Coding region of NM_000260 (49), NM_001127179 (27), NM_001127180 (49)

Mucopolysaccharidosis type IIIB (Sanfilippo B) - OMIM: 252920 - Gene: NAGLU - Carrier frequency 1/501 - Coding region of NM_000263 (6)

Nijmegen Breakage Syndrome (Ataxia telangectasia, type 1) - OMIM: 251260 - Gene: NBN - Carrier frequency 1/159 - Coding region of NM_001024688 (17), NM_002485 (16) Norrie disease - OMIM: 310600 - Gene: NDP - Carrier frequency 1/500001 - Coding region of NM_000266 (3)

Charcot-Marie-Tooth disease, type 4D - OMIM: 601455 - Gene: NDRG1 - Carrier frequency 1/501 - Coding region of NM_001135242 (16), NM_001258432 (14), NM_001258433 (15), NM_006096 (16)

Nemaline myopathy 2, autosomal recessive - OMIM: 256030 - Gene: NEB - Carrier frequency 1/159 - Coding region of NM_001164507 (182), NM_001164508 (182), NM_001271208 (183), NM_004543 (149)

Mental retardation, X-linked, Asperger syndrome susceptibility, X-linked - OMIM: 300427 - Gene: NLGN4X - Carrier frequency 1/45001 - Coding region of NM_001282145 (7), NM_001282146 (6), NM_020742 (6), NM_181332 (6)

Niemann-Pick disease, type C1 - OMIM: 257220 - Gene: NPC1 - Carrier frequency 1/181 - Coding region of NM_000271 (25)

Niemann-pick disease, type C2 - OMIM: 607625 - Gene: NPC2 - Carrier frequency 1/867 - Coding region of NM_006432 (5)

Joubert syndrome 4 - OMIM: 609583 - Gene: NPHP1 - Carrier frequency 1/159 - Coding region of NM_000272 (20), NM_001128178 (20), NM_001128179 (18), NM_207181 (20)

Nephrotic syndrome, type 1 - OMIM: 256300 - Gene: NPHS1 - Carrier frequency 1/501 - Coding region of NM_004646 (29)

Enhanced S-cone syndrome (Goldmann-Favre syndrome) - OMIM: 268100 - Gene: NR2E3 - Carrier frequency 1/1582 - Coding region of NM_014249 (9), NM_016346 (8)

Dent disease 2 - OMIM: 300555 - Gene: OCRL - Carrier frequency 1/5001 - Coding region of NM_000276 (24), NM_001318784 (24), NM_001587 (23) Mental retardation, X-linked - OMIM: 300486 - Gene: OPHN1 - Carrier frequency 1/500001 - Coding region of NM_002547 (25)

Ornithine transcarbamylase deficiency - OMIM: 311250 - Gene: OTC - Carrier frequency 1/50001 - Coding region of NM_000531 (10)

Auditory neuropathy, autosomal recessive, 1 - OMIM: 601071 - Gene: OTOF - Carrier frequency 1/102 - Coding region of NM_001287489 (46), NM_004802 (30), NM_194248 (47), NM_194322 (29), NM_194323 (29)

Osteogenesis imperfecta, type VIII - OMIM: 610915 - Gene: P3H1 - Carrier frequency 1/159 - Coding region of NM_001146289 (15), NM_001243246 (14), NM_022356 (15)

Phenylketonuria - OMIM: 261600 - Gene: PAH - Carrier frequency 1/49 - Coding region of NM_000277 (13)

Mental retardation, X-linked 30/47 - OMIM: 300558 - Gene: PAK3 - Carrier frequency 1/45001 - Coding region of NM_001128166 (16), NM_001128167 (15), NM_001128168 (20), NM_-001128172 (15), NM_001128173 (19), NM_001324325 (16), NM_001324326 (16), NM_001324327 (18), NM_001324328 (17), NM_001324329 (19), NM_001324330 (16), NM_001324331 (17), NM_001324332 (18), NM_001324333 (18), NM_001324334 (17), NM_002578 (18), NR_136740 (3), NR_136741 (4), NR_136742 (5), NR_136743 (4), NR_136744 (5), NR_136745 (6), NR_136746 (6), NR_136747 (18), NR_136748 (16)

HARP syndrome - OMIM: 607236 - Gene: PANK2 - Carrier frequency 1/501 - Coding region of NM_001324191 (8), NM_001324192 (2), NM_001324193 (7), NM_024960 (7), NM_153638 (7), NM_153640 (7), NR_136715 (7)

Pyruvate carboxylase deficiency - OMIM: 266150 - Gene: PC - Carrier frequency 1/251 - Coding region of NM_000920 (22), NM_001040716 (23), NM_022172 (21)

Hyperphenylalaninemia, BH4-deficient, D - OMIM: 264070 - Gene: PCBD1 - Carrier frequency 1/501 - Coding region of NM_000281 (4), NM_001289797 (4), NM_001323004 (4)

Propionic acidemia - OMIM: 606054 - Gene: PCCA - Carrier frequency 1/708* - Coding region of NM_000282 (24), NM_001127692 (23), NM_001178004 (23)

Propionic acidemia - OMIM: 606054 - Gene: PCCB - Carrier frequency 1/708* - Coding region of NM_000532 (15), NM_001178014 (16)

Deafness, autosomal recessive 23; Usher syndrome, type 1F; Usher syndrome, type 1D/F digenic - OMIM: 609533; 602083; 601067 - Gene: PCDH15 - Carrier frequency 1/80 - Coding region of NM_001142763 (35), NM_001142764 (34), NM_001142765 (32), NM_001142766 (32), NM_001142767 (31), NM_001142768 (33), NM_001142769 (36), NM_001142770 (35), NM_001142771 (35), NM_001142772 (34), NM_001142773 (32), NM_033056 (33)

Retinitis pigmentosa 43 - OMIM: 613810 - Gene: PDE6A - Carrier frequency 1/501 - Coding region of NM_000440 (22)

Pyruvate dehydrogenase E1-beta deficiency - OMIM: 614111 - Gene: PDHB - Carrier frequency 1/251 - Coding region of NM_000925 (10), NM_001173468 (11), NM_001315536 (9), NR_-033384 (9)

Peroxisome biogenesis disorder 1A (Zellweger) - OMIM: 214100 - Gene: PEX1 - Carrier frequency 1/138 - Coding region of NM_000466 (24), NM_001282677 (23), NM_001282678 (24)

Peroxisome biogenesis disorder 6A (Zellweger) - OMIM: 614870 - Gene: PEX10 - Carrier frequency 1/138 - Coding region of NM_002617 (6), NM_153818 (6)

Heimler syndrome, type 2 - OMIM: 616617 - Gene: PEX6 - Carrier frequency 1/501 - Coding region of NM_000287 (17), NM_001316313 (17), NR_133009 (15)

Rhizomelic chondroplasia punctata, type I - OMIM: 215100 - Gene: PEX7 - Carrier frequency 1/159 - Coding region of NM_000288 (10) Phosphoglycerate kinase 1 deficiency - OMIM: 300653 - Gene: PGK1 - Carrier frequency 1/500001 - Coding region of NM_000291 (11)

Mental retardation syndrome, X-linked, Siderius type - OMIM: 300263 - Gene: PHF8 - Carrier frequency 1/500001 - Coding region of NM_001184896 (22), NM_001184897 (22), NM_-001184898 (22), NM_015107 (22)

Autosomal Recessive Polycystic Kidney Disease - OMIM: 263200 - Gene: PKHD1 - Carrier frequency 1/101 - Coding region of NM_138694 (67), NM_170724 (61)

Ehlers-Danlos syndrome, type VI - OMIM: 225400 - Gene: PLOD1 - Carrier frequency 1/317 - Coding region of NM_000302 (19), NM_001316320 (20)

Pelizaeus-Merzbacher disease - OMIM: 312080 - Gene: PLP1 - Carrier frequency 1/500001 - Coding region of NM_000533 (7), NM_001128834 (8), NM_001305004 (7), NM_199478 (7)

Congenital disorder of glycosylation, type Ia - OMIM: 212065 - Gene: PMM2 - Carrier frequency 1/355 - Coding region of NM_000303 (8)

Treacher Collins syndrome 3 - OMIM: 248390 - Gene: POLR1C - Carrier frequency 1/159 - Coding region of NM_001318876 (9), NM_203290 (9)

Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3 - OMIM: 253280 - Gene: POMGNT1 - Carrier frequency 1/708 - Coding region of NM_-001243766 (23), NM_001290129 (22), NM_001290130 (22), NM_017739 (22)

Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 - OMIM: 236670 - Gene: POMT1 - Carrier frequency 1/159 - Coding region of NM_001077365 (20), NM_001077366 (19), NM_001136113 (20), NM_001136114 (18), NM_007171 (20)

Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2 - OMIM: 613150 - Gene: POMT2 - Carrier frequency 1/159 - Coding region of NM_013382 (21)

Deafness, X-linked 2 - OMIM: 304400 - Gene: POU3F4 - Carrier frequency 1/97501 - Coding region of NM_000307 (1) Ceroid lipofuscinosis, neuronal, type 1 - OMIM: 256730 - Gene: PPT1 - Carrier frequency 1/251 - Coding region of NM_000310 (9), NM_001142604 (6)

Renpenning syndrome - OMIM: 309500 - Gene: PQBP1 - Carrier frequency 1/500001 - Coding region of NM_001032381 (7), NM_001032382 (7), NM_001032383 (7), NM_001032384 (7), NM_001167989 (7), NM_001167990 (7), NM_001167992 (6), NM_005710 (6), NM_144495 (6) Pituitary hormone deficiency, combined, 2 - OMIM: 262600 - Gene: PROP1 - Carrier frequency 1/46 - Coding region of NM_006261 (3)

Arts syndrome; Charcot-Marie-Tooth disease, X-linked recessive, 5; Deafness, X-linked 1; Gout, PRPS-related (Phosphoribosylpyrophosphate synthetase superactivity) - OMIM: 301835; 311070; 304500; 300661 - Gene: PRPS1 - Carrier frequency 1/250001 - Coding region of NM_001204402 (4), NM_002764 (7)

Hyperphenylalaninemia, BH4-deficient, A - OMIM: 261640 - Gene: PTS - Carrier frequency 1/501 - Coding region of NM_000317 (6)

McArdle disease (Glycogen Storage Disease, type V) - OMIM: 232600 - Gene: PYGM - Carrier frequency 1/225 - Coding region of NM_001164716 (18), NM_005609 (20)

Hyperphenylalaninemia, BH4-deficient, C - OMIM: 261630 - Gene: QDPR - Carrier frequency 1/501 - Coding region of NM_000320 (7), NM_001306140 (6)

Microphthalmia, isolated 3 - OMIM: 611038 - Gene: RAX - Carrier frequency 1/159 - Coding region of NM_013435 (3)

Leber congenital amaurosis 13 - OMIM: 612712 - Gene: RDH12 - Carrier frequency 1/501 - Coding region of NM_152443 (9)

Anauxetic dysplasia; Cartilage-hair hypoplasia - OMIM: 607095; 250250 - Gene: RMRP - Carrier frequency 1/225 - Coding region of NR_003051 (1)

Retinitis pigmentosa 2 - OMIM: 312600 - Gene: RP2 - Carrier frequency 1/5001 - Coding region of NM_006915 (5)

Leber congenital amaurosis 2 - OMIM: 204100 - Gene: RPE65 - Carrier frequency 1/263 - Coding region of NM_000329 (14)

Cone-rod dystrophy, X-linked, 1 - OMIM: 304020 - Gene: RPGR - Carrier frequency 1/50001 - Coding region of NM_000328 (19), NM_001034853 (15)

Coffin-Lowry syndrome - OMIM: 303600 - Gene: RPS6KA3 - Carrier frequency 1/50001 - Coding region of NM_004586 (22) Retinoschisis, X-linked - OMIM: 312700 - Gene: RS1 - Carrier frequency 1/6251 - Coding region of NM_000330 (6)

Spastic ataxia, Charlevoix-Saguenay type (ARSACS) - OMIM: 270550 - Gene: SACS - Carrier frequency 1/501 - Coding region of NM_001278055 (8), NM_014363 (10)

Emphysema due to Alpha1 Anti-Trypsin deficiency - OMIM: 613490 - Gene: SERPINA1 - Carrier frequency 1/20 - Coding region of NM_000295 (5), NM_001002235 (5), NM_001002236 (7), NM_001127700 (5), NM_001127701 (7), NM_001127702 (6), NM_001127703 (7), NM_001127704 (7), NM_001127705 (7), NM_001127706 (6), NM_001127707 (6)

Muscular dystrophy, limb-girdle, type 2D - OMIM: 608099 - Gene: SGCA - Carrier frequency 1/293 - Coding region of NM_000023 (10), NM_001135697 (8), NR_135553 (9)

Muscular dystrophy, limb-girdle, type 2E - OMIM: 604286 - Gene: SGCB - Carrier frequency 1/525 - Coding region of NM_000232 (6)

Mucopolysaccharidisis type IIIA (Sanfilippo A) - OMIM: 252900 - Gene: SGSH - Carrier frequency 1/501 - Coding region of NM_000199 (8)

Charcot-Marie-Tooth disease, type 4C - OMIM: 601596 - Gene: SH3TC2 - Carrier frequency 1/72 - Coding region of NM_024577 (17)

Allan-Herndon-Dudley syndrome - OMIM: 300523 - Gene: SLC16A2 - Carrier frequency 1/500001 - Coding region of NM_006517 (6)

Carnitine deficiency, systemic primary - OMIM: 212140 - Gene: SLC22A5 - Carrier frequency 1/159 - Coding region of NM_001308122 (11), NM_003060 (10)

Citrullinemia, neonatal-onset, type II - OMIM: 605814 - Gene: SLC25A13 - Carrier frequency 1/241 - Coding region of NM_001160210 (18), NM_014251 (18), NR_027662 (17)

Carnitine-acylcarnitine translocase deficiency - OMIM: 212138 - Gene: SLC25A20 - Carrier frequency 1/501 - Coding region of NM_000387 (9)

Achondrogenesis Ib - OMIM: 600972 - Gene: SLC26A2 - Carrier frequency 1/140 - Coding region of NM_000112 (3)

Pendred syndrome - OMIM: 274600 - Gene: SLC26A4 - Carrier frequency 1/51 - Coding region of NM_000441 (21)

Glycogen storage disease Ib - OMIM: 232220 - Gene: SLC37A4 - Carrier frequency 1/159 - Coding region of NM_001164277 (11), NM_001164278 (12), NM_001164279 (11), NM_001164280 (9), NM_001467 (10)

Cystinuria - OMIM: 220100 - Gene: SLC3A1 - Carrier frequency 1/43 - Coding region of NM_000341 (10)

Folate malabsorption, hereditary - OMIM: 229050 - Gene: SLC46A1 - Carrier frequency 1/501 - Coding region of NM_001242366 (5), NM_080669 (6)

Corneal endothelial dystrophy and sensorineural deafness (CDPD) - OMIM: 217400 - Gene: SLC4A11 - Carrier frequency 1/501 - Coding region of NM_001174089 (20), NM_001174090 (20), NM_032034 (19), NR_135000 (20)

Thyroid dyshormonogenesis 1 - OMIM: 274400 - Gene: SLC5A5 - Carrier frequency 1/72 - Coding region of NM_000453 (15)

Hartnup disorder - OMIM: 234500 - Gene: SLC6A19 - Carrier frequency 1/159 - Coding region of NM_001003841 (12) Cystinuria - OMIM: 220100 - Gene: SLC7A9 - Carrier frequency 1/43 - Coding region of NM_001126335 (13), NM_001243036 (13), NM_014270 (13)

Spinal muscular atrophy, type I - OMIM: 253300 - Gene: SMN1 - Carrier frequency 1/40 - NM_022874: exon 7 deletion

Niemann-Pick disease, type A; Niemann-Pick disease, type B - OMIM: 257200; 607616 - Gene: SMPD1 - Carrier frequency 1/251 - Coding region of NM_000543 (6), NM_001007593 (6), NM_001318087 (6), NM_001318088 (6), NR_027400 (5), NR_134502 (6)

Spastic paraplegia 11, autosomal recessive - OMIM: 604360 - Gene: SPG11 - Carrier frequency 1/159 - Coding region of NM_001160227 (38), NM_025137 (40)

Spastic paraplegia 7, autosomal recessive - OMIM: 607259 - Gene: SPG7 - Carrier frequency 1/113 - Coding region of NM_003119 (17), NM_199367 (10)

Lipoid adrenal hyperplasia - OMIM: 201710 - Gene: STAR - Carrier frequency 1/159 - Coding region of NM_000349 (7)

Leigh syndrome, due to COX deficiency - OMIM: 256000 - Gene: SURF1 - Carrier frequency 1/96 - Coding region of NM_001280787 (8), NM_003172 (9)

Epilepsy, X-linked, with variable learning disabilities and behavior disorders - OMIM: 300491 - Gene: SYN1 - Carrier frequency 1/45001 - Coding region of NM_006950 (13), NM_133499 (13)

Tyrosinemia, type II - OMIM: 276600 - Gene: TAT - Carrier frequency 1/501 - Coding region of NM_000353 (12)

Hemochromatosis, type 3 - OMIM: 604250 - Gene: TFR2 - Carrier frequency 1/867 - Coding region of NM_001206855 (15), NM_003227 (18)

Thyroid dyshormonogenesis 3 - OMIM: 274700 - Gene: TG - Carrier frequency 1/159 - Coding region of NM_003235 (48)

Ichthyosis, congenital, autosomal recessive 1 - OMIM: 242300 - Gene: TGM1 - Carrier frequency 1/501 - Coding region of NM_000359 (15)

Segawa syndrome, recessive - OMIM: 605407 - Gene: TH - Carrier frequency 1/708 - Coding region of NM_000360 (13), NM_199292 (14), NM_199293 (14)

Mental retardation, X-linked 12/35 - OMIM: 300957 - Gene: THOC2 - Carrier frequency 1/500001 - Coding region of NM_001081550 (39)

Thyroid hormone resistance - OMIM: 274700 - Gene: THRB - Carrier frequency 1/101 - Coding region of NM_000461 (10), NM_001128176 (11), NM_001128177 (11), NM_001252634 (12)

Joubert syndrome 2 - OMIM: 608091 - Gene: TMEM216 - Carrier frequency 1/159 - Coding region of NM_001173990 (5), NM_001173991 (5), NM_001330285 (5), NM_016499 (5)

Thyroid dyshormonogenesis 2A - OMIM: 274500 - Gene: TPO - Carrier frequency 1/72* - Coding region of NM_000547 (17), NM_001206744 (17), NM_001206745 (16), NM_175719 (16), NM_175721 (15), NM_175722 (15)

Ceroid lipofuscinosis, neuronal, 2 - OMIM: 204500 - Gene: TPP1 - Carrier frequency 1/159 - Coding region of NM_000391 (13)

Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness - OMIM: 615441 - Gene: TRDN - Carrier frequency 1/51 - Coding region of NM_001251987 (21), NM_001256020 (9), NM_001256021 (8), NM_001256022 (6), NM_006073 (41)

Hypothryoidism, congenital, nongoitrous 4 - OMIM: 275100 - Gene: TSHB - Carrier frequency 1/33 - Coding region of NM_000549 (3), NM_001277991 (1)

Hypothyroidism, congenital, nongoitrous, 1 - OMIM: 275200 - Gene: TSHR - Carrier frequency 1/33 - Coding region of NM_000369 (10), NM_001018036 (9), NM_001142626 (9)

Mental retardation, X-linked 58 - OMIM: 300210 - Gene: TSPAN7 - Carrier frequency 1/45001 - Coding region of NM_004615 (8)

Ataxia with isolated vitamin E deficiency - OMIM: 277460 - Gene: TTPA - Carrier frequency 1/275 - Coding region of NM_000370 (5)

Mental retardation, X-linked, syndromic 14 - OMIM: 300676 - Gene: UPF3B - Carrier frequency 1/45001 - Coding region of NM_023010 (10), NM_080632 (11)

Deafness, autosomal recessive 18A - OMIM: 602092 - Gene: USH1C - Carrier frequency 1/159 - Coding region of NM_001297764 (20), NM_005709 (21), NM_153676 (27), NR_123738 (20)

Usher syndrome, type 1G - OMIM: 606943 - Gene: USH1G - Carrier frequency 1/315 - Coding region of NM_001282489 (3), NM_173477 (3)

Retinitis pigmentosa 39; Usher syndrome 2A - OMIM: 613809 - Gene: USH2A - Carrier frequency 1/88 - Coding region of NM_007123 (21), NM_206933 (72)

Mental retardation, X-linked 99 - OMIM: 300919 - Gene: USP9X - Carrier frequency 1/45001 - Coding region of NM_001039590 (45), NM_001039591 (45)

Usher syndrome, type 2D / Deafness, autosomal recessive 31 - OMIM: 611383 - Gene: WHRN - Carrier frequency 1/54 - Coding region of NM_001083885 (12), NM_001173425 (12), NM_-015404 (12)

Mental retardation, X-linked syndromic, Raymond type - OMIM: 300799 - Gene: ZDHHC9 - Carrier frequency 1/45001 - Coding region of NM_001008222 (10), NM_016032 (11)

Mental retardation, X-linked 97 - OMIM: 300803 - Gene: ZNF711 - Carrier frequency 1/45001 - Coding region of NM_001330574 (11), NM_021998 (9)