REFERENCE LABORATORY GENETICS

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CarrierRef®

Request No.:	123456	Code of client	
Patient Name:		Client	
Date of Birth:		Patient Ref. :	
Sample Type:	WHOLE BLOOD	Gender:	MALE
Analysis Code:	15048	Sample extraction date	
Sample Arrival Date:		Date of Result:	

REFERRAL REASON

We received a sample of an adult male patient to analyse his carrier status for heterozygous mutations in genes causing diseases with recessive inheritance pattern by using *CarrierRef®.*

SUMMARY OF RESULTS

The results obtained with *CarrierRef®* indicate that the patient **carries** a mutation in the gene involved in the disease detailed in the table below.

Condition/Pathology	Gene	Mutation
Cystic Fibrosis; Congenital bilateral absence of the vas deferens	**CFTR**	**c.1210-34TG(12)T(5) (NM_000492)**

- No other mutations were found with a possible marked phenotypic effect in the analysed regions (see in *Analysed Pathologies*).

POSITIVE RESULT: Cystic Fibrosis; Congenital bilateral absence of the vas deferens (OMIM 219700; 277180)

HIGH RISK	Pre-test Risk[1]	Risk of affected offspring[2]	Risk of carriers[3]
	<1/4.807	<1/140	1/2

Pre-test Risk[1] coincides with the incidence of pathology, **the risk of an affected offspring** [2] takes into account the status of the index case carrier and the probability of the spouse being a carrier. Finally, the **risk of generating offspring carriers**[3] of the mutation is obtained from the information on the status of the analysed parent.

RESULTS OBTAINED IN THE PATIENT:

The following mutation was identified:

	Identified Mutations	Carrier	Recommendations
Result:	CFTR : c.1210-34TG(12)T(5) (NM_000492)	Heterozygous	This variant has variable phenotypic consequences when accompanied by another pathogenic mutation in another allele. Specifically, the observed 5T-12TG allele accompanied by a second mild or classical CFTR mutation may provide a range of phenotypes between asymptomatic, non-classical CF and infertility due to the congenital absence of vas deferens [1].

What is Cystic Fibrosis; Congenital bilateral absence of the vas deferens?

Mutations in the *CFTR* gene may cause Cystic Fibrosis (CF) or other related disorders of lesser severity. CF is characterized by the production of sweat with high salt content and mucous secretions with abnormal viscosity. It is the most common genetic disorder among Caucasian children. A chronic and usually progressive disease that usually appears during early infancy or, rarely, at birth (meconium ileus). Any internal organ can be affected, although the major manifestations affect the respiratory system (chronic bronchitis), pancreas (pancreatic insufficiency, diabetes of the adolescent and occasionally pancreatitis) and, less frequently, intestine (intestinal obstruction) or liver (cirrhosis). Some mutations in the *CFTR* gene are related to less severe isolated phenotypes, known as *CFTR* related disorders, which include pancreatitis, recurrent infections and sterility due to the congenital absence of vas deferens or oligospermia.

Cystic Fibrosis; Can congenital bilateral absence of the vas deferens be treated?

Treatment of CF remains purely symptomatic through bronchial drainage, antibiotics for respiratory infections, pancreatic analysis and administration of vitamins and caloric supplements for digestive and nutritional problems. Treatment of pancreatitis focuses on pain relief, treatment of digestive disorders, diabetes, pseudocysts, and duodenal and bile duct obstruction.

What is the prognosis of this disease?

Thanks to the treatments, the prognosis of the CF has improved significantly in the last decades. Currently, the average life is over 35 years and the life expectancy is 40 years.
As for CFTR-related disorders, they are less severe and have a more favorable prognosis.

Additional information

CF is an autosomal recessive disease caused by mutations in the CFTR gene (chromosome 7). More than 1,250 mutations have been identified. Approximately 70% of the cases are caused by the F508 allele, while another 30 mutations would explain another 20% more. There is no clear correlation between genotype and phenotype. In addition to allelic heterogeneity and the occurrence of multiple mutations in the same gene, a wide range of factors may influence the phenotype, including the environment and disease-modifying genes.

Other Links

· Cystic Fibrosis mutation database: www.genet.sickkids.on.ca
· CFTR John Hopkins mutation database: www.cftr2.org
· Gene Reviews: www.ncbi.nlm.nih.gov/books/NBK1250

MATERIALS, METHODS AND LIMITATIONS OF THE TEST

CarrierRef® uses a **massive sequencing** to detect the presence of genetic variants related to human recessive diseases. Genome regions of interest are captured, amplified, and sequenced with high coverage (minimum coverage of 20x) to ensure reliability in the detection of sequence variants. Only high quality readings are used with an average error rate of less than 1 in 1000. The ability of this technique to detect Single Nucleotide Variants (SNVs) and small insertions and deletions (in/dels) of up to 9 nucleotides with high sensitivity and specificity (99%) is well known. Likewise, it is recognized that there is less reliability in the detection of other types of genetic variation (changes in the number of copies of medium/large size, translocations, inversions and triplet expansions); In this sense, the presence of segmental duplications, pseudogenes and other repetitive sequences may interfere with the correct detection of variants. Structural type variants affecting a single complete exon in genes in which only the coding region is sequenced, as well as any kind of inversion, may not be detected. The detection rate for copy number variations covering more than one exon, is 95%. The *CarrierRef®* can detect variants in: **1) Coding regions in autosomes**. It includes (exons), along with intron/exon boundaries, genes that cause prevalent diseases, and congenital metabolic errors (amino acids, carbohydrates and fatty acids) which, although rare, can have serious consequences if not detected and controlled in time. (A total of 70 genes and 1115 exons are studied). **2) Point mutations in autosomes**, for the study of genes in which only a small number of mutations are known as the cause of the genetic origin of the disease. (A total of 1381 mutations are studied in 120 genes). **3) X-linked variants.** The particular pattern of inheritance of these diseases deserves a differentiated attention, so that in the X chromosome, the study of complete coding regions (exons) is combined with the directed analysis of mutations.

CarrierRef® allows to identify the genetic variants, detailed in the final table of this report. In this report, only those that cause disease (**pathogenic**) are described, according to current knowledge. Besides that, it is quite likely that other variants are also detected, for which no role has been described in the literature in relation to the disease. From these, only the ones that are classified as *potentially pathogenic* will be reported, based on their theoretical potential to cause a negative impact on gene function, whether due to the type of change they generate in the protein, or in response to a combination of this information together with its frequency in cases and controls, functional studies, segregation studies, ... that may exist in the scientific literature. The known genetic susceptibility variants, **benign** genetic variants, variant of **uncertain clinical significance** or other incidental variants will also be reported, upon request of the requesting physician.

Copy number analysis. The copy number of the regions of interest is determined by comparing the depth of coverage relative to other genomic "locus" using specific analytical algorithms [5]. The presence of other types of mutations in these "locus" may interfere with this measure. In the specific case of the copy number of exon 7 of the *SMN1* gene, related to SMA, it should be remembered that the existence of deletion carriers was reported in one allele combined with the duplication of the same *SMN1* in the other allele, with which they would appear as individuals with normal dose in our analysis. On the other hand, in a small percentage of the cases, the SMA is originated by other mutational mechanisms different from the copy number variations. Therefore, a 'normal' *CarrierRef®* result compatible with the presence of two copies of the *SMN1* gene, significantly reduces the probability of having a deletion allele, but there is still a residual risk of having a progeny with a number of copies changed. In addition, it should be remembered that some cases of SMA appear "de novo", with which it is not possible to detect them using *CarrierRef®.*

Known technical limitations. There are different situations that can interfere in the detection of mutations and can result in the non-identification of variants that are actually present in the patient's sample. On the one hand, the presence of certain types of adjacent genetic variants could mask the identification of certain mutations; In addition, a bone marrow transplant or blood transfusion can also lead to ambiguity and misinterpretation of the genotype. Although rigorous controls are performed to minimize this type of error, it is also not impossible for a technical or human error to occur in the form of errors in the identification of samples, the existence of cross-contamination of DNA and other technical problems. The *CarrierRef®* uses massive sequencing technology after capturing target regions. It is known that in the library capture and preparation process as well as sequencing, there may be regions of interest for which no sufficient sequence quantity is available to determine the presence of variants. With the controls used, we can ensure that > 95% of target regions are sufficiently covered (> 30x).

Range of the test. The *CarrierRef®* looks at only part of the spectrum of known mutations that are the cause of certain genetic diseases related to intellectual disability and other birth defects, but there are many other diseases of genetic origin that are not analysed by this test. Not all genetic forms of the diseases studied can be detected by this test. Ask your geneticist or genetic counselor if there is a genetic disease in your family, which may require other types of laboratory tests, genetics or not.

INSUFFICIENTLY COVERED REGIONS

EXONS with incomplete sequence or coverage	15X

There are no uncovered exons

ANALYSED PATHOLOGIES

Diseases, Genes and Variants Tested

3-Methylcrotonyl-CoA carboxylase 1 deficiency-210200 - Gene:MCCC1 - Carrier frequency 1/113 - Coding Region of NM_020166 (+7 variants)
3-Methylcrotonyl-CoA carboxylase 2 deficiency-210210 - Gene:MCCC2 - Carrier frequency 1/140 - Coding Region of NM_022132 (+8 variants)
Acyl-CoA dehydrogenase, medium chain, deficiency of-201450 - Gene:ACADM - Carrier frequency 1/62 - Coding Region of NM_000016 (+10 variants)
Acyl-CoA dehydrogenase, short-chain, deficiency of-201470 - Gene:ACADS - Carrier frequency 1/133 - Coding Region of NM_000017 (+16 variants)
Alkaptonuria-203500 - Gene:HGD - Carrier frequency 1/163 - Coding Region of NM_000187 (+13 variants)
Alpha-methylacetoacetic aciduria-203750 - Gene:ACAT1 - Carrier frequency 1/708 - Coding Region of NM_000019 (+7 variants)
Argininemia-207800 - Gene:ARG1 - Carrier frequency 1/501 - Coding Region of NM_000045 (+7 variants)
Argininosuccinic aciduria-207900 - Gene:ASL - Carrier frequency 1/195 - Coding Region of NM_001024944 (+8 variants)
Biotinidase deficiency-253260 - Gene:BTD - Carrier frequency 1/124 - Coding Region of NM_000060 (+19 variants)
CPT I (Carnitine Palmitoyltransferase IA) deficiency, hepatic, type IA-255120 - Gene:CPT1A - Carrier frequency 1/708 - Coding Region of NM_001031847 (+28 variants)
CPT II (Carnitine Palmitoyltransferase) deficiency, myopathy due to -608836 - Gene:CPT2 - Carrier frequency 1/708 - Coding Region of NM_000098 (+21 variants)
Carnitine deficiency, systemic primary-212140 - Gene:SLC22A5 - Carrier frequency 1/159 - Coding Region of NM_003060 (+95 variants)
Citrullinemia-215700 - Gene:ASS1 - Carrier frequency 1/159 - Coding Region of NM_000050 (+16 variants)
Cystathioninuria-219500 - Gene:CTH - Carrier frequency 1/80 - Coding Region of NM_001190463 (+2 variants)
Cystic fibrosis-219700 - Gene:CFTR - Carrier frequency 1/36 - Coding Region of NM_000492 (+932 variants)
Cystinuria-220100 - Gene:SLC3A1 - Carrier frequency 1/43 - Coding Region of NM_000341 (+7 variants)
Cystinuria-220100 - Gene:SLC7A9 - Carrier frequency 1/43 - Coding Region of NM_001126335 (+11 variants)
Deafness, autosomal recessive 1A (DFNB1-related)-220290 - Gene:GJB2 - Carrier frequency 1/17 - Coding Region of NM_004004 (+72 variants)
Diabetes mellitus, neonatal, with congenital hypothyroidism-610199 - Gene:GLIS3 - Carrier frequency 1/708 - Coding Region of NM_001042413 (+1 variants)
Familial Mediterranean fever, autosomal recessive, including mild form-249100 - Gene:MEFV - Carrier frequency 1/36 - Coding Region of NM_000243 (+165 variants)
Galactokinase deficiency with cataracts-230200 - Gene:GALK1 - Carrier frequency 1/225 - Coding Region of NM_000154 (+5 variants)
Galactose epimerase deficiency-230350 - Gene:GALE - Carrier frequency 1/159 - Coding Region of NM_001127621 (+11 variants)
Galactosemia-230400 - Gene:GALT - Carrier frequency 1/113 - Coding Region of NM_001258332 (+238 variants)
Glutaric acidemia IIA-231680 - Gene:ETFA - Carrier frequency 1/708 - Coding Region of NM_001127716 (+3 variants)
Glutaric acidemia IIB-231680 - Gene:ETFB - Carrier frequency 1/708 - Coding Region of NM_001014763 (+2 variants)
Glutaric acidemia IIC-231680 - Gene:ETFDH - Carrier frequency 1/708 - Coding Region of NM_004453 (+5 variants)
Glutaric aciduria, type I-231670 - Gene:GCDH - Carrier frequency 1/95 - Coding Region of NM_000159 (+8 variants)
HMG-CoA lyase deficiency-246450 - Gene:HMGCL - Carrier frequency 1/159 - Coding Region of NM_000191 (+5 variants)
Histidinemia-235800 - Gene:HAL - Carrier frequency 1/72 - Coding Region of NM_001258334 (+5 variants)
Homocystinuria, B6-responsive and nonresponsive types-236200 - Gene:CBS - Carrier frequency 1/124 - Coding Region of NM_000071 (+15 variants)
Hypermethioninemia due to Glycine N-methyltransferase deficiency-606664 - Gene:GNMT - Carrier frequency 1/501 - Coding Region of NM_018960 (+2 variants)
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase-613752 - Gene:AHCY - Carrier frequency 1/708 - Coding Region of NM_001161766 (+3 variants)

Hypermethioninemia, persistent, due to MAT1 deficiency-250850 - Gene:MAT1A - Carrier frequency 1/708 - Coding Region of NM_000429 (+7 variants)
Hyperprolinemia, type II-239510 - Gene:ALDH4A1 - Carrier frequency 1/708 - Coding Region of NM_001161504 (+1 variants)
Hypothryoidism, congenital, nongoitrous 4-275100 - Gene:TSHB - Carrier frequency 1/33* - Coding Region of NM_000549 (+3 variants)
Hypothyroidism, congenital, nongoitrous 1-275200 - Gene:TSHR - Carrier frequency 1/33 - Coding Region of NM_000369 (+30 variants)
Isovaleric acidemia-243500 - Gene:IVD - Carrier frequency 1/159 - Coding Region of NM_001159508 (+4 variants)
LCHAD deficiency-609016 - Gene:HADHA - Carrier frequency 1/126 - Coding Region of NM_000182 (+7 variants)
Maple syrup urine disease, type II-248600 - Gene:DBT - Carrier frequency 1/216 - Coding Region of NM_001918 (+4 variants)
Maple syrup urine disease, type III-248600 - Gene:DLD - Carrier frequency 1/216 - Coding Region of NM_000108 (+6 variants)
Maple syrup urine disease, type Ia-248600 - Gene:BCKDHA - Carrier frequency 1/216 - Coding Region of NM_000709 (+10 variants)
Maple syrup urine disease, type Ib-248600 - Gene:BCKDHB - Carrier frequency 1/216 - Coding Region of NM_000056 (+4 variants)
Methylmalonic aciduria and homocystinuria, cblC type-277400 - Gene:MMACHC - Carrier frequency 1/111 - Coding Region of NM_015506 (+4 variants)
Methylmalonic aciduria and homocystinuria, cblD type-277410 - Gene:MMADHC - Carrier frequency 1/111 - Coding Region of NM_015702 (+7 variants)
Methylmalonic aciduria, vitamin B12-responsive, cblB type-251110 - Gene:MMAB - Carrier frequency 1/111 - Coding Region of NM_052845 (+1 variants)
Methylmalonic aciduria, vitamin B12-responsive, cblA type-251100 - Gene:MMAA - Carrier frequency 1/111 - Coding Region of NM_172250 (+3 variants)
Methylmalonic aciduria, mut(0) type-251000 - Gene:MUT - Carrier frequency 1/138 - Coding Region of NM_000255 (+10 variants)
Methylmalonyl-CoA epimerase deficiency-251120 - Gene:MCEE - Carrier frequency 1/1119 - Coding Region of NM_032601 (+1 variants)
Mevalonic aciduria-610377 - Gene:MVK - Carrier frequency 1/230 - Coding Region of NM_000431 (+90 variants)
Pendred syndrome-274600 - Gene:SLC26A4 - Carrier frequency 1/51 - Coding Region of NM_000441 (+65 variants)
Phenylketonuria-261600 - Gene:PAH - Carrier frequency 1/49 - Coding Region of NM_000277 (+496 variants)
Propionic acidemia-606054 - Gene:PCCA - Carrier frequency 1/708 - Coding Region of NM_001127692 (+5 variants)
Propionic acidemia-606054 - Gene:PCCB - Carrier frequency 1/708 - Coding Region of NM_000532 (+15 variants)
Spinal muscle atrophy (several types)-253300,253550,253400,271150 - Gene:SMN1 - Carrier frequency 1/40 - NM_022874: Exon 7 deletion
Thalassemia, alpha-604131 - Gene:HBA2 - Carrier frequency 1/44 - Coding Region of NM_000517 (+34 variants)
Thalassemia, alpha-604131 - Gene:HBA1 - Carrier frequency 1/44 - Coding Region of NM_000558 (+122 variants)
Thalassemias, beta- (Sickle Cell Anemia)-613985 - Gene:HBB - Carrier frequency 1/23 - Coding Region of NM_000518 (+541 variants)
Thryoid dyshormonogenesis 6-607200 - Gene:DUOX2 - Carrier frequency 1/72 - Coding Region of NM_014080 (+6 variants)
Thyroid dyshormonogenesis 1-274400 - Gene:SLC5A5 - Carrier frequency 1/72 - Coding Region of NM_000453 (+9 variants)
Thyroid dyshormonogenesis 2A-274500 - Gene:TPO - Carrier frequency 1/72 - Coding Region of NM_175722 (+11 variants)
Thyroid dyshormonogenesis 3-274700 - Gene:TG - Carrier frequency 1/159 - Coding Region of NM_003235 (+16 variants)

Thyroid dyshormonogenesis 4-274800 - Gene:IYD - Carrier frequency 1/72 - Coding Region of NM_001164694 (+3 variants)

Thyroid dyshormonogenesis 5-274900 - Gene:DUOXA2 - Carrier frequency 1/72 - Coding Region of NM_207581 (+1 variants)

Tyrosinemia, type I-276700 - Gene:FAH - Carrier frequency 1/167 - Coding Region of NM_000137 (+16 variants)

VLCAD deficiency-201475 - Gene:ACADVL - Carrier frequency 1/138 - Coding Region of NM_001033859 (+20 variants)

Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency-201910 - Gene:CYP21A2 - Carrier frequency 1/64 - Variants in NM_000500 (11) Whole del; p.I173N; p.Q319X; c.293-13C>G; p.G293S; c.332_339delGAGACTAC

Abetalipoproteinemia-200100 - Gene:MTTP - Carrier frequency 1/708 - Variants in NM_000253 (2) c.2212delT; c.G2593T:p.G865X

Achromatopsia: CNGB3 Related-262300 - Gene:CNGB3 - Carrier frequency 1/101 - Variants in NM_019098 (2) c.1148delC:p.T383fs; c.817_824delCCCAGACT

Acrodermatitis enteropathica-201100 - Gene:SLC39A4 - Carrier frequency 1/225 - Variants in NM_017767 (10) c.G1501A:p.G501R; c.1223-1227delCCGGG:p.W411RfsX7; c.1017ins53; c.G1045A:p.G349R; c.G914A:p.G305D; c.968-971delAGTC; c.G834C:p.Q278H; c.C524T:p.P175L; c.C243A:p.N81K; c.C208T:p.R70C

Alport syndrome, autosomal recessive, COL4A3 related-203780 - Gene:COL4A3 - Carrier frequency 1/113 - Variants in NM_000091 (3) c.4415-4419delCTTTT; c.C4441T:p.R1481X; c.C4571G:p.S1524X

Alport syndrome, autosomal recessive, COL4A4 related-203780 - Gene:COL4A4 - Carrier frequency 1/113 - Variants in NM_000092 (5) c.C4923A:p.C1641X; c.C4715T:p.P1572L; c.C4129T:p.R1377X; c.C3713A:p.S1238X; c.G3601A:p.G1201S

Andermann Syndrome / Agenesis of the corpus callosum with peripheral neuropathy-218000 - Gene:SLC12A6 - Carrier frequency 1/708 - Variants in NM_001042497 (5)

c.C2986T:p.R996X; c.2436delG:p.T813fsX813; c.C1978T:p.R660X; c.901delA; c.C574T:p.R192C

Aromatase deficiency-613546 - Gene:CYP19A1 - Carrier frequency 1/1370 - Variants in NM_000103 (8) c.G1310A:p.C437Y; c.C1303T:p.R435C; c.1222-1224delC; c.C1123T:p.R375C; c.G1094A:p.R365Q; c.858+2T>C; c.G628A:p.E210K; c.468delC

Aspartylglucosaminuria-208400 - Gene:AGA - Carrier frequency 1/1119 - Variants in NM_000027 (10) c.G503A:p.W168X; c.G488C:p.C163S; c.G482A:p.R161Q; c.T439C:p.S147P; c.T404C:p.F135S; c.395-8A>G; c.T214C:p.S72P; c.200-201del:p.67-67del; c.T192A:p.C64X; c.G179A:p.G60D

Ataxia with isolated vitamin E deficiency-277460 - Gene:TTPA - Carrier frequency 1/275 - Variants in NM_000370 (1) c.744delA

Ataxia-telangiectasia-208900 - Gene:ATM - Carrier frequency 1/159 - Variants in NM_000051 (1) c.C103T:p.R35X

Autoimmune polyendocrinopathy syndrome, type I-240300 - Gene:AIRE - Carrier frequency 1/355 - Variants in NM_000383 (23) c.T230C:p.F77S; c.T232A:p.W78R; c.G238T:p.V80L; c.T239G:p.V80G; c.A247G:p.K83E; c.A254G:p.Y85C; c.A269G:p.Y90C; c.T278G:p.L93R; c.G342T:p.K114N; c.C371T:p.P124L; c.C415T:p.R139X; c.G463A:p.G155S; c.G682T:p.G228W; c.C755T:p.P252L; c.C769T:p.R257X; c.789delC:p.G263fs; c.G932A:p.C311Y; c.965-977delGCCTGTCCCCTCC; c.967-979del:p.323-327del; c.C977A:p.P326Q; c.1097-1098insC:p.L366fs; c.G1105A:p.G369R; c.1163_1164insA:p.M388fs

Autosomal Recessive Polycystic Kidney Disease (ARPKD)-263200 - Gene:PKHD1 - Carrier frequency 1/101 - Variants in NM_138694 (7) c.T10658C:p.I3553T; c.T10412G:p.V3471G; c.9689delA; c.C9053T:p.S3018F; c.5895insA; c.C1486T:p.R496X; c.C107T:p.T36M

Bardet-Biedl syndrome 1-209900 - Gene:BBS1 - Carrier frequency 1/195 - Variants in NM_024649 (3) c.851delA; c.T1169G:p.M390R; c.G1645T:p.E549X

Bardet-Biedl syndrome 10-209900 - Gene:BBS10 - Carrier frequency 1/195 - Variants in NM_024685 (10) c.A1736G:p.K579R; c.T1245C:p.H415H; c.G1202A:p.G401E; c.T931G:p.S311A; c.A385T:p.I129F; c.C273G:p.C91W; c.271-273insA:p.C91fsX95; c.G101C:p.R34P; c.T32G:p.V11G

Bardet-Biedl syndrome 2-209900 - Gene:BBS2 - Carrier frequency 1/195 - Variants in NM_031885 (7) c.940delA; c.C823T:p.R275X; c.A311C:p.D104A; c.T224G:p.V75G; c.C175T:p.Q59X; c.C72G:p.Y24X

Bare Lymphocyte Syndrome, type II-209920 - Gene:CIITA - Carrier frequency 1/613 - Variants in NM_000246 (7) c.1007-94A>G; c.1007-54C>T; c.G1141T:p.E381X; c.G1230A:p.P410P; c.G1684A:p.A562T; c.C1957A:p.P653T; c.C2356A:p.Q786K

Bartter syndrome, type 4a-602522 - Gene:BSND - Carrier frequency 1/645 - Variants in NM_057176 (8) c.A1T:p.M1L; c.G3A:p.M1I; c.G10A:p.E4K; c.C22T:p.R8W; c.G23A:p.R8Q; c.G28A:p.G10S; c.T35C:p.I12T; c.G139A:p.G47R

Bloom Syndrome-210900 - Gene:BLM - Carrier frequency 1/708 - Variants in NM_000057 (4) c.2406+206->T; c.2487-2488insA:p.L829fs; c.2506-2507del:p.836-836del; c.C2528T:p.T843I

Canavan disease-271900 - Gene:ASPA - Carrier frequency 1/708 - Variants in NM_000049 (7) c.433-2A>G; c.T454C:p.C152R; c.C654A:p.C218X; c.A692G:p.Y231C; c.C693A:p.Y231X; c.A854C:p.E285A; c.C914A:p.A305E

Cartilage-hair hypoplasia-250250 - Gene:RMRP - Carrier frequency 1/225 - Variants in NR_003051 (5) c.G262T; g.35657923-35657925TCT>TGC; c.A70G; n.70dupG; n.71A>G

Cerebrotendinous xanthomatosis-213700 - Gene:CYP27A1 - Carrier frequency 1/113 - Variants in NM_000784 (15) c.C379T:p.R127W; c.G434A:p.G145E; c.G583T:p.E195X; c.G646C:p.A216P; c.C819delT; c.844+1G>A; c.C1016T:p.T339M; c.C1183A:p.R395S; c.C1183T:p.R395C; c.G1214A:p.R405Q; c.1263+1G>A; c.C1420T:p.R474W; c.G1421A:p.R474Q; c.C1435G:p.R479G; c.C1435T:p.R479C

Ceroid lipofuscinosis, neuronal, 5-256731 - Gene:CLN5 - Carrier frequency 1/501 - Variants in NM_006493 (6) c.G225A:p.W75X; c.T772G:p.Y258D; c.G835A:p.D279N; c.918delA:p.T306fs; c.955-970del:p.319-324del; c.C1026A:p.Y342X

Ceroid lipofuscinosis, neuronal, 8-600143 - Gene:CLN8 - Carrier frequency 1/501 - Variants in NM_018941 (23) c.C46A:p.L16M; c.62delG:p.W21fs; c.C70G:p.R24G; c.G84A:p.M28I; c.G88C:p.A30P; c.88delG:p.A30fs; c.180-182del:p.60-61del; c.G209A:p.R70H; c.A227G:p.Q76R; c.T320G:p.I107S; c.C415T:p.H139Y; c.C464T:p.A155V; c.A473G:p.Y158C; c.C507T:p.S169S; c.C509T:p.T170M; c.560-561del:p.187-187del; c.A581G:p.Q194R; c.C610T:p.R204C; c.G611T:p.R204L; c.634-636del:p.212-212del; c.G661A:p.G221S; c.C766G:p.Q256E; c.G789C:p.W263C

Ceroid lipofuscinosis, neuronal, type 1-256730 - Gene:PPT1 - Carrier frequency 1/251 - Variants in NM_000310 (36) c.465-466insA:p.Q156fs; c.G440T:p.G147V; c.T430C:p.Y144H; c.727-2A>T; c.T374G:p.V125G; c.T365C:p.F122S; c.T356C:p.L119P; c.T347A:p.L116Q; c.335delA:p.Y112fs; c.628-1G>T; c.536+2T>C; c.536+1G>A; c.C220G:p.Q74E; c.C181T:p.R61X; c.C147A:p.C49X; c.G146A:p.C49Y; c.C142T:p.R48X; c.C413T:p.S138L; c.398delT:p.M133fs; c.A364T:p.R122W; c.363-3T>G; c.A223C:p.T75P; c.175delG:p.E59fs; c.169-170insA:p.M57fs; c.A163T:p.K55X; c.135-136insTGT:p.C46delinsCC; c.G134A:p.C45Y; c.G125A:p.G42E; c.125-2A>G; c.125-15T>G; c.124+1G>A; c.T117A:p.H39Q; c.G114T:p.W38C; c.114delG:p.W38fs; c.T29A:p.L10X; c.G3A:p.M1I

Ceroid lipofuscinosis, neuronal, type 2-204500 - Gene:TPP1 - Carrier frequency 1/159 - Variants in NM_000391 (10) c.T860A:p.I287N; c.A857G:p.N286S; c.G851T:p.G284V; c.G829A:p.V277M; c.C622T:p.R208X; c.G617A:p.R206H; c.C616T:p.R206C; c.C605T:p.P202L; c.523-1G>A; c.523-1G>C

Cholestasis, progressive-601847 - Gene:ABCB11 - Carrier frequency 1/159 - Variants in NM_003742 (5) c.3767-3768insC:p.T1256fs; c.C3169T:p.R1057X; c.C1723T:p.R575X; c.G1295C:p.R432T; c.A890G:p.E297G

Congenital disorder of glycosylation, type Ib-602579 - Gene:MPI - Carrier frequency 1/1582 - Variants in NM_002435 (1) c.G884A:p.R295H

Congenital disorder of glycosylation, type Ic-603147 - Gene:ALG6 - Carrier frequency 1/1119 - Variants in NM_013339 (2) c.C998T:p.A333V; c.T1432C:p.S478P

Congenital disorder of glycosylation, type Ia-212065 - Gene:PMM2 - Carrier frequency 1/355 - Variants in NM_000303 (6) c.G349C:p.G117R; c.C357A:p.F119L; c.G385A:p.V129M; c.T395C:p.I132T; c.G415A:p.E139K; c.G422A:p.R141H

Corneal endothelial dystrophy and sensorineural deafness (CDPD)-217400 - Gene:SLC4A11 - Carrier frequency 1/501 - Variants in NM_001174089 (12) c.G2606A:p.R869H; c.C2605T:p.R869C; c.A2566G:p.M856V; c.T2528C:p.L843P; c.G2264A:p.R755Q; c.C2261T:p.T754M; c.2233-2240insTATGACAC:p.Thr747ThrfsX6; c.G1463A:p.R488K; c.G1391A:p.G464D; c.1378delTACGinsA:p.Tyr460-Ala461; c.T637C:p.S213P; c.473delGCTTCGCC:p.Arg158ProfsX4

Crigler-Najjar syndrome, type I-218800 - Gene:UGT1A1 - Carrier frequency 1/501 - Variants in NM_000463 (13) c.-53-40insTA; c.T44G:p.L15R; c.G211A:p.G71R; c.T347A:p.I116K; c.508-513delTTC; c.T524A:p.L175Q; c.C686A:p.P229Q; c.C840A:p.C280X; c.C1021T:p.R341X; c.A1070G:p.Q357R; c.C1124T:p.S375F; c.A1198C:p.N400H; c.A1198G:p.N400D

Cystinosis, nephropathic-219800 - Gene:CTNS - Carrier frequency 1/225 - Variants in NM_001031681 (13) c.18-21delGACT; c.198-218del:p.66-73del; c.198delATTACTATCCTTGAGCTCCCC; c.G283T:p.G95X; c.G329T:p.G110V; c.G413A:p.W138X; c.G414A:p.W138X; c.C416T:p.S139F; c.T473C:p.L158P; c.G506A:p.G169D; c.559-561del:p.187-187del; c.G589A:p.G197R; c.G613A:p.D205N

Diabetes Mellitus, permanent neonatal-606176 - Gene:KCNJ11 - Carrier frequency 1/317 - Variants in NM_000525 (8) c.G602T:p.R201L; c.C601T:p.R201C; c.A499C:p.I167L; c.G497T:p.C166F; c.T179A:p.F60Y; c.T176G:p.V59G; c.G175A:p.V59M; c.G158A:p.G53D

Diastrophic dysplasia-222600 - Gene:SLC26A2 - Carrier frequency 1/140 - Variants in NM_000112 (36) c.C47G:p.S16X; c.G55T:p.G19X; c.C81G:p.I27M; c.254delC:p.A85fs; c.G331T:p.D111Y; c.C398T:p.A133V; c.C403A:p.Q135K; c.G496A:p.G166R; c.C532T:p.R178X; c.699+2T>C; c.700-1G>C; c.705-711del:p.235-237del; c.G764A:p.G255E; c.C835T:p.R279W; c.904-905del:p.302-302del; c.1012-1014del:p.338-338del; c.1018-1020delGTT; c.C1157G:p.A386G; c.C1215G:p.A405A; c.1242-1245del:p.414-415del; c.A1273G:p.N425D; c.A1361C:p.Q454P; c.1391delT:p.L464fs; c.G1451A:p.G484D; c.C1535A:p.T512K; c.1650delG:p.K550fs; c.1723delA:p.K575fs; c.T1798C:p.Y600H; c.T1957A:p.C653S; c.1972delT:p.F658fs; c.1983delA:p.T661fs; c.A1996G:p.T666A; c.G2033T:p.G678V; c.2120-2121del:p.707-707del; c.C2144T:p.A715V

Dihydropyrimidine dehydrogenase deficiency-274270 - Gene:DPYD - Carrier frequency 1/51 - Variants in NM_000110 (6) c.G2657A:p.R886H; c.1905+1G>A; c.1897delC; c.295delTCAT Dubin-Johnson syndrome-237500 - Gene:ABCC2 - Carrier frequency 1/501 - Variants in NM_000392 (6) c.C2302G:p.R768G; c.C2302T:p.R768W; c.2439+2T>C; c.G3449A:p.R1150H; c.A3517T:p.I1173F; c.A4145G:p.Q1382R

Dysautonomia, familial-223900 - Gene:IKBKAP - Carrier frequency 1/708 - Variants in NM_003640 (2) c.2204+6T>C; c.G2087C:p.R696P

Ehlers-Danlos syndrome, type VIIC-225410 - Gene:ADAMTS2 - Carrier frequency 1/1937 - Variants in NM_014244 (1) c.C673T:p.Q225X

Ellis-van Creveld Syndrome-225500 - Gene:EVC2 - Carrier frequency 1/188 - Variants in NM_001166136 (1) c.C3025T:p.Q1009X

Emphysema due to Alpha1 Anti-Trypsin deficiency-613490 - Gene:SERPINA1 - Carrier frequency 1/20 - Variants in NM_000295 (27) c.A1200C:p.E400D; c.C1178T:p.P393L; c.G1159A:p.E387K; c.T1145G:p.M382R; c.A1131T:p.L377F; c.G1093A:p.D365N; c.G1078A:p.A360T; c.A839T:p.D280V; c.C739T:p.R247C; c.A721T:p.K241X; c.G682A:p.E228K; c.A649T:p.K217X; c.C552G:p.Y184X; c.552delC:p.Y184X; c.G415A:p.G139S; c.G374A:p.R125H; c.T347A:p.I116N; c.C326T:p.T109M; c.G272A:p.G91E; c.C230T:p.S77F; c.T194C:p.L65P; c.A77C:p.D26A

Enhanced S-cone syndrome (Retinitis pigmentosa 37,)-268100 - Gene:NR2E3 - Carrier frequency 1/1582 - Variants in NM_014249 (1) c.G932A:p.R311Q

Epidermolysis bullosa dystrophica, AR-226600 - Gene:COL7A1 - Carrier frequency 1/708 - Variants in NM_000094 (10) c.T8393A:p.M2798K; c.C7411T:p.R2471X; c.7345-1G>A; c.5821-1G>A; c.G4039C:p.G1347R; c.2470insG; c.C933A:p.Y311X; c.C933T:p.Y311Y; c.427-2A>G; c.A425G:p.K142R

Epidermolysis bullosa, junctional, Herlitz type-226700 - Gene:LAMB3 - Carrier frequency 1/613 - Variants in NM_001017402 (7) c.3024delT; c.C1903T:p.R635X; c.G1830A:p.W610X; c.C727T:p.Q243X; c.C496T:p.Q166X; c.C430T:p.R144X; c.C124T:p.R42X

Ethylmalonic encephalopathy-602473 - Gene:ETHE1 - Carrier frequency 1/969 - Variants in NM_014297 (4) c.505+1G>T; c.C487T:p.R163W; c.221-222insA; c.G3T:p.M1I

Factor V Deficiency-227400 - Gene:F5 - Carrier frequency 1/501 - Variants in NM_000130 (2) c.C3481T:p.R1161X; c.C2401T:p.Q801X

Thrombophilia due to ACP resistance-188055 - Gene:F5 - Carrier frequency 1/36 - Variants in NM_000130 (7) c.A3899G:p.H1300R; c.3187-3188insA:p.R1063fs; c.G2127A:p.M709I; c.G1001C:p.R334T; c.A1000G:p.R334G; c.G439T:p.E147X

Factor XI deficiency, autosomal recessive-612416 - Gene:F11 - Carrier frequency 1/501 - Variants in NM_000128 (21) c.G403T:p.E135X; c.G419A:p.C140Y; c.C438A:p.C146X; c.A452G:p.Y151C; c.A809T:p.K270I; c.C827G:p.S276C; c.G830A:p.G277D; c.T901C:p.F301L; c.G943A:p.E315K; c.T959C:p.L320P; c.C965T:p.T322I; c.C976T:p.R326C; c.C992T:p.T331I; c.G1021A:p.E341K; c.T1079C:p.L360P; c.T1378G:p.F460V; c.T1442G:p.I481S; c.G1684A:p.G562S; c.G1693A:p.E565K; c.1714-1716+11delAAGgtaacagagtg; c.1716+1G>A

Fanconi anemia, complementation group C-227645 - Gene:FANCC - Carrier frequency 1/201 - Variants in NM_000136 (6) c.T1661C:p.L554P; c.C1642T:p.R548X; c.C553T:p.R185X; c.456+4A>T; c.66delG; c.C37T:p.Q13X

Fibular hypoplasia and complex brachydactyly / Du pan syndrome-228900 - Gene:GDF5 - Carrier frequency 1/1119 - Variants in NM_000557 (12) c.G1471A:p.E491K; c.T1461G:p.Y487X; c.G1424A:p.S475N; c.T1322C:p.L441P; c.T1315A:p.S439T; c.G1313T:p.R438L; c.1309delTTG; c.C1306A:p.P436T; c.G1199A:p.C400Y; c.G1133A:p.R378Q; c.T1118G:p.L373R; c.C901T:p.R301X

Fructose intolerance-229600 - Gene:ALDOB - Carrier frequency 1/72 - Variants in NM_000035 (8) c.T1027C:p.Y343H; c.C1005G:p.N335K; c.T612G:p.Y204X; c.547-552del:p.183-184del; c.C524A:p.A175D; c.G448C:p.A150P; c.T442C:p.W148R; c.357_360del:p.119_120del

Fumarase deficiency-606812 - Gene:FH - Carrier frequency 1/613 - Variants in NM_000143 (2) c.1433-1434insAAA:p.N478delinsKN; c.C1421T:p.T474I

GM1-gangliosidosis, types I, II-230500 - Gene:GLB1 - Carrier frequency 1/195 - Variants in NM_000404 (24) c.A1379G:p.Y460C; c.T1378C:p.Y460H; c.T1771A:p.Y591N; c.G1134T:p.W378C; c.A1105G:p.T369A; c.1480-2A>G; c.G1052A:p.R351H; c.C1051T:p.R351C; c.G977A:p.R326Q; c.C976T:p.R326X; c.G920A:p.G307E; c.C658T:p.R220X; c.A554G:p.Y185C; c.G425T:p.W142L; c.C622T:p.R208C; c.C601T:p.R201C; c.G367A:p.G123R; c.T247C:p.Y83H; c.C245T:p.T82M; c.C202T:p.R68W; c.G176A:p.R59H; c.T152C:p.I51T; c.C145T:p.R49C; c.75+2_75+3insT

GRACILE Syndrome-603358 - Gene:BCS1L - Carrier frequency 1/501 - Variants in NM_001257344 (7) c.G103C:p.G35R; c.C133T:p.R45C; c.A148G:p.T50A; c.C166T:p.R56X; c.A232G:p.S78G; c.C296T:p.P99L; c.320+1G>T

Gaucher disease, types I and II-230800,230900 - Gene:GBA - Carrier frequency 1/159 - Variants in NM_000157 (22) c.G1343A:p.R448H; c.G1288A:p.G430S; c.T1251C:p.S417S; c.G1244A:p.R415H; c.C1243T:p.R415C; c.G1222C:p.A408P; c.T1187C:p.L396P; c.G1183A:p.D395N; c.C1100G:p.P367R; c.A1082T:p.D361V; c.G1081C:p.D361H; c.C1058T:p.P353L; c.1002-1056del:p.334-352del; c.G1048T:p.V350F; c.G1036T:p.V346L; c.G985A:p.G329S; c.C967G:p.L323V; c.A965G:p.N322S; c.G93C:p.K31N; c.115+1G>A; c.93-94insG:p.Q32fs; c.84_85insG:p.L29fs

Gitelman syndrome-263800 - Gene:SLC12A3 - Carrier frequency 1/101 - Variants in NM_000339 (13) c.C625T:p.R209W; c.A697G:p.M233V; c.C1046T:p.P349L; c.G1189A:p.V397M; c.T1261C:p.C421R; c.G1710A:p.A570A; c.C1763T:p.A588V; c.T1868C:p.L623P; c.G1889T:p.G630V; c.1923-1G>T; c.C1956A:p.P652P; c.G1964A:p.R655H; c.2744+1G>T

Glycogen storage disease II / Pompe disease-232300 - Gene:GAA - Carrier frequency 1/159 - Variants in NM_000152 (5) c.T-45G; c.G271A:p.D91N; c.G1927A:p.G643R; c.C1935A:p.D645E; c.C2560T:p.R854X

Glycogen storage disease IIIa-232400 - Gene:AGL - Carrier frequency 1/159 - Variants in NM_000028 (4) c.C135T:p.F45F; c.C1171T:p.R391X; c.4403delT:p.L1468fs; c.4455delT:p.S1486PfsX18

Glycogen storage disease IV-232500 - Gene:GBE1 - Carrier frequency 1/501 - Variants in NM_000158 (3) c.A986C:p.Y329S; c.C784T:p.R262C

Glycogen storage disease Ia-232200 - Gene:G6PC - Carrier frequency 1/159 - Variants in NM_000151 (19) c.79delC; c.A113T:p.D38V; c.T229C:p.W77R; c.C247T:p.R83C; c.G248A:p.R83H; c.G328A:p.E110K; c.G370A:p.A124T; c.376-377insTA:p.V126fs; c.T497G:p.V166G; c.G474A:p.W158X; c.G562C:p.G188R; c.G648T:p.L216L; c.C724T:p.Q242X; c.G750A:p.W250X; c.G809T:p.G270V; c.C883T:p.R295C; c.979-981del:p.327-327del; c.T1022A:p.I341N; c.C1039T:p.Q347X

Glycogen storage disease Ib-232220 - Gene:SLC37A4 - Carrier frequency 1/159 - Variants in NM_001164280 (15) c.G1126A:p.G376S; c.C1118A:p.A373D; c.G1099A:p.A367T; c.G1063T:p.E355X; c.1042-1043del:p.348-348del; c.G1016A:p.G339D; c.G1015T:p.G339C; c.T352C:p.W118R; c.G287A:p.W96X; c.G263A:p.G88D; c.T254C:p.L85P; c.G202A:p.G68R; c.A163C:p.S55R; c.C162A:p.S54R; c.G149A:p.G50E

Glycogen storage disease VII-232800 - Gene:PFKM - Carrier frequency 1/708 - Variants in NM_000289 (4) c.G116A:p.R39Q; c.G116T:p.R39L; c.C283T:p.R95X; c.593+1G>A

Hemochromatosis, type 3-604250 - Gene:TFR2 - Carrier frequency 1/867 - Variants in NM_003227 (13) c.A1556C:p.Q519P; c.G817A:p.A273T; c.721-723del:p.241-241del; c.C673T:p.R225X; c.C436T:p.Q146X; c.C237G:p.Y79X; c.C201G:p.I67M; c.T2A:p.M1K; c.C224T:p.A75V; c.C97A:p.H33N; c.88-89insC:p.R30fs; c.84insC; c.G64A:p.V22I

Hemochromatosis: Type 2A: HFE2 Related-602390 - Gene:HFE2 - Carrier frequency 1/1582 - Variants in NM_213652 (5) c.T326A:p.I109N; c.T164C:p.I55T; c.G281T:p.G94V; c.C285A:p.C95X; c.C298T:p.R100X

Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related-226700 - Gene:LAMC2 - Carrier frequency 1/607 - Variants in NM_005562 (1) c.C283T:p.R95X

Hermansky-Pudlak syndrome 3-614072 - Gene:HPS3 - Carrier frequency 1/434 - Variants in NM_032383 (1) c.1691+2T>G

Holocarboxylase synthetase deficiency-253270 - Gene:HLCS - Carrier frequency 1/225 - Variants in NM_000411 (6) c.1795+5G>A; c.G1741A:p.G581S; c.G1711A:p.D571N; c.780delG; c.T710C:p.L237P; c.T647G:p.L216R

Hyperinsulinemic hypoglycemia, familial, type 1-256450 - Gene:ABCC8 - Carrier frequency 1/113 - Variants in NM_000352 (23) c.G4516A:p.E1506K; c.C4477T:p.R1493W; c.G4451A:p.G1484E; c.4412-14C>T; c.G4307A:p.R1436Q; c.C4306T:p.R1436X; c.A4270G:p.I1424V; c.C4258T:p.R1420C; c.G4198A:p.G1400R; c.4159-4161delTTC:p.1387-1387del; c.4156-4158del:p.1386-1386del; c.G4136T:p.R1379L; c.C4135T:p.R1379C; c.G4055C:p.R1352P; c.3988+138G>A; c.G2147T:p.G716V; c.2117-12C>A; c.G1144A:p.E382K; c.T560A:p.V187D; c.413-5G>A; c.T257G:p.V86G; c.A215G:p.N72S

Hyperinsulinemic hypoglycemia, familial, type 2-601820 - Gene:KCNJ11 - Carrier frequency 1/501 - Variants in NM_000525 (25) c.G1001A:p.G334D; c.T997A:p.F333I; c.A989G:p.Y330C; c.G964A:p.E322K; c.G902T:p.R301L; c.A886G:p.I296V; c.G844A:p.E282K; c.A776G:p.H259R; c.C761T:p.P254L; c.T755C:p.V252A; c.A544G:p.I182V; c.G510C:p.K170N; c.A509G:p.K170R; c.T440C:p.L147P; c.G157C:p.G53R; c.A155G:p.Q52R; c.G149C:p.R50P; c.T124C:p.C42R; c.T103G:p.F35V; c.A67G:p.K23E; c.A54G:p.A18A; c.C36A:p.Y12X; c.T5C:p.L2P; c.G-134T

Hyperoxaluria III-613616 - Gene:HOGA1 - Carrier frequency 1/708 - Variants in NM_138413 (3) c.G371T:p.G124V; c.449-451del:p.150-151del; c.944_946delAGG

Hyperoxaluria, primary, type I-259900 - Gene:AGXT - Carrier frequency 1/501 - Variants in NM_000030 (61) c.1-3AAT; c.T2C:p.M1T; c.G3T:p.M1I; c.26delC:p.T9fs; c.26-27del:p.9-9del; c.C27A:p.T9T; c.C32A:p.P11H; c.33insC; c.A65G:p.N22S; c.T74G:p.L25R; c.T77C:p.L26P; c.82delC:p.P28fs; c.C106T:p.R36C; c.G107A:p.R36H; c.115-116insCA:p.A39fs; c.G121A:p.G41R; c.121delG:p.G41fs; c.G122A:p.G41E; c.G125A:p.G42E; c.C130T:p.Q44X; c.G139A:p.G47R;c.165+16A>G;
c.165+18>CGGGGGGCCTGGGTCTCACCCATGTTCCCACCCACAGATCGTGGACGAGGGAAGGGGGTCACTGCCTCCTCACT;
c.165+40A>C; c.165+44T>A; c.165+83C>T; c.165+84C>T; c.165+86C>T; c.424-2A>G; c.445delG:p.V149fs; c.447-454del:p.149-152del; c.T449C:p.L150P; c.T454A:p.F152I; c.T457G:p.L153V; c.459delA:p.L153fs; c.G466A:p.G156R; c.C473A:p.S158X; c.G481A:p.G161S; c.G489A:p.L163L; c.T497C:p.L166P; c.G508A:p.G170R; c.G518A:p.C173Y; c.518-520GGA; c.C519A:p.C173X; c.524+91C>T; c.C697T:p.R233C; c.G698A:p.R233H; c.G705A:p.T235T; c.724-725insT:p.L242fs; c.G727C:p.D243H; c.T731C:p.I244T; c.G737A:p.W246X; c.G738A:p.W246X; c.G742T:p.A248S; c.743delC:p.A248fs; c.G753A:p.W251X; c.T757C:p.C253R; c.776+1G>A

Hyperoxaluria, primary, type II-260000 - Gene:GRHPR - Carrier frequency 1/708 - Variants in NM_012203 (11) c.84-2A>G; c.101delG:p.W34fs; c.G102A:p.W34X; c.103delG; c.T203C:p.L68P; c.288-11C>T; c.C295T:p.R99X; c.G337A:p.E113K; c.374delG:p.R125fs; c.403-404del:p.135-135del; c.404+1_404+4del

Hypophosphatasia, infantile-241500 - Gene:ALPL - Carrier frequency 1/159 - Variants in NM_001177520 (15) c.G254T:p.G85V; c.G295A:p.A99T; c.G304A:p.A102T; c.G340A:p.E114K; c.A389C:p.Q130P; c.A650C:p.D217A; c.G661A:p.E221K; c.T748C:p.F250L; c.G770A:p.G257D; c.A902T:p.D301V; c.G928A:p.G310R; c.A1019G:p.N340S; c.T1075C:p.Y359H; c.G1135A:p.G379R; c.1328delT:p.L443fs

Inclusion Body Myopathy, type 2-600737 - Gene:GNE - Carrier frequency 1/388 - Variants in NM_001190384 (3) c.T1805C:p.M602T; c.G1756T:p.V586M; c.G1663A:p.V555M

Joubert syndrome 2-608091 - Gene:TMEM216 - Carrier frequency 1/159 - Variants in NM_001173990 (5) c.G218A:p.R73H; c.G218T:p.R73L; c.G230C:p.G77A; c.C253A:p.R85R; c.T341G:p.L114R

Krabbe disease-245200 - Gene:GALC - Carrier frequency 1/159 - Variants in NM_000153 (3) c.G1084T:p.E362X; c.G788A:p.G263D; c.C550T:p.R184C

Leber congenital amaurosis 5-604537 - Gene:LCA5 - Carrier frequency 1/1174 - Variants in NM_001122769 (1) c.C835T:p.Q279X

Leigh syndrome, French-Canadian type-220111 - Gene:LRPPRC - Carrier frequency 1/708 - Variants in NM_133259 (1) c.C1061T:p.A354V

Limb-Girdle Muscular Dystrophy: Type 2I; Muscular dystrophy-dystroglycanopathy -607155 - Gene:FKRP - Carrier frequency 1/708 - Variants in NM_001039885 (13) c.C160G:p.R54G; c.G235A:p.V79M; c.C400T:p.R134W; c.T899C:p.V300A; c.T919A:p.Y307N; c.A926G:p.Y309C; c.C946A:p.P316T; c.C1154A:p.S385X; c.G1213T:p.V405L; c.C1343T:p.P448L; c.C1364A:p.A455D; c.A1387G:p.N463D; c.T1486A:p.X496R

Lipoid adrenal hyperplasia-201710 - Gene:STAR - Carrier frequency 1/159 - Variants in NM_000349 (13) c.C772T:p.Q258X; c.G749A:p.W250X; c.G650C:p.R217T; c.C577T:p.R193X; c.C562T:p.R188C; c.G559A:p.V187M; c.G545T:p.R182L; c.G545A:p.R182H; c.466-11T>A; c.201-202delCT; c.178+3T>T; c.135delT:p.P45fs; c.64+1G>T

Lipoprotein lipase deficiency-238600 - Gene:LPL - Carrier frequency 1/501 - Variants in NM_000237 (17) c.A548G:p.D183G; c.C596G:p.S199C; c.G607A:p.A203T; c.C621G:p.D207E; c.G644A:p.G215E; c.T662C:p.I221T; c.G665A:p.G222E; c.C693G:p.D231E; c.C701T:p.P234L; c.T755C:p.I252T; c.C798G:p.C266W; c.C808T:p.R270C; c.G809A:p.R270H; c.T811A:p.S271T; c.G829A:p.D277N; c.C987A:p.Y329X

McArdle disease / Glycogen Storage Disease: Type V-232600 - Gene:PYGM - Carrier frequency 1/225 - Variants in NM_001164716 (15) c.2128-2130delTTC; c.T2128C:p.W710R; c.G1792A:p.G598R; c.C1732G:p.Q578E; c.G1699A:p.E567K; c.G1563A:p.K521K; c.C1462T:p.R488X; c.T1458G:p.Y486X; c.A1364C:p.K455T; c.A1627T:p.K543X; c.G1357T:p.E453X; c.G349A:p.G117S; c.C255A:p.Y85X; c.C148T:p.R50X; c.A1G:p.M1V

Metachromatic leukodystrophy-250100 - Gene:ARSA - Carrier frequency 1/113 - Variants in NM_000487 (95) c.T1471G:p.C491G; c.C1462T:p.Q488X; c.1407-1417del:p.469-473del; c.C1412G:p.A471G; c.G1306A:p.D436N; c.G1294T:p.D432Y; c.A1292C:p.Y431S; c.T1289C:p.L430P; c.C1283T:p.P428L; c.C1279A:p.P427T; c.C1232T:p.T411I; c.C1229T:p.T410I; c.A1222G:p.S408G; c.1210+1G>A; c.C1195T:p.H399Y; c.G1175A:p.R392Q; c.C1174T:p.R392W; c.C1156T:p.R386C; c.G1150A:p.E384K; c.C1149T:p.D383D; c.C1136T:p.P379L; c.T1132A:p.Y378N; c.G1115A:p.R372Q; c.C1114T:p.R372W; c.G1107C:p.K369N; c.A1010T:p.D337V; c.G946A:p.A316T; c.G942T:p.E314D; c.G938A:p.R313Q; c.G931A:p.G311S; c.G929T:p.G310V; c.G925A:p.E309K; c.T922C:p.Y308H; c.C917T:p.T306M; c.G912C:p.K304N; c.G905T:p.C302F; c.T899C:p.L300S; c.C890A:p.S297Y; c.G887A:p.C296Y; c.G884A:p.G295D; c.G883A:p.G295S; c.G869A:p.R290H; c.C868T:p.R290C; c.A862C:p.T288P; c.A851G:p.N284S; c.G847T:p.D283Y; c.C827T:p.T276M; c.G769C:p.D257H; c.G763A:p.E255K; c.758-759insT:p.L253fs; c.C755A:p.S252Y; c.T746C:p.F249S; c.G739A:p.G247R; c.G737A:p.R246H; c.C736T:p.R246C; c.C697A:p.P233T; c.C685T:p.H229Y; c.C677T:p.A226V; c.T661G:p.F221V; c.C641T:p.A214V; c.G640C:p.A214P; c.A608G:p.Y203C; c.C577A:p.P193T; c.G576C:p.Q192H; c.T548A:p.L183Q; c.T542G:p.I181S; c.G521A:p.C174Y; c.G514A:p.G172S; c.G511A:p.D171N; c.C506G:p.P169R; c.T472C:p.C158R; c.C470T:p.P157L; c.G467A:p.G156D; c.459+1G>A; c.G465C:p.Q155H; c.G460T:p.D154Y; c.C449T:p.P150L; c.C433G:p.R145G; c.C418G:p.H140D; c.C413T:p.P138L; c.C412T:p.P138S; c.T410C:p.L137P; c.G370A:p.G124S; c.G361A:p.G121R; c.G302T:p.G101V; c.292-294CTC; c.C293T:p.S98F; c.G290A:p.S97N; c.C286G:p.P96A; c.G263A:p.G88D; c.G257A:p.R86Q; c.C256T:p.R86W; c.T233C:p.L78P

Mucolipidosis III alpha/beta, and type II-252500,252600 - Gene:GNPTAB - Carrier frequency 1/251 - Variants in NM_024312 (17) c.G3598A:p.E1200K; c.3569-3570insA:p.N1190fs; c.C3565T:p.R1189X; c.3523-3529del:p.1175-1177del; c.3503-3504del:p.1168-1168del; c.3487-3490del:p.1163-1164del; c.3474-3475del:p.1158-1159del; c.A3458G:p.N1153S; c.3443-3446del:p.1148-1149del; c.3435-1G>A; c.A1220G:p.D407G; c.T1208C:p.I403T; c.1206-1207insT:p.I403fs; c.C1196T:p.S399F; c.1194-1195insGCTG:p.S399fs; c.C1123T:p.R375X; c.T1120C:p.F374L

Mucolipidosis IV-252650 - Gene:MCOLN1 - Carrier frequency 1/101 - Variants in NM_020533 (5) g.511-6943del; c.C304T:p.R102X; c.405+153A>G; c.C964T:p.R322X; c.G1084T:p.D362Y

Mucopolysaccharidosis Ih / Hurler Syndrome-607014 - Gene:IDUA - Carrier frequency 1/210 - Variants in NM_000203 (7) c.C192A:p.Y64X; c.C208T:p.Q70X; c.G266A:p.R89Q; c.G979C:p.A327P; c.T1037G:p.L346R; c.C1091T:p.T364M; c.A1096C:p.T366P

Muscular dystrophy, limb-girdle, type 2D-608099 - Gene:SGCA - Carrier frequency 1/293 - Variants in NM_000023 (2) c.C229T:p.R77C; c.G293A:p.R98H

Muscular dystrophy, limb-girdle, type 2E-604286 - Gene:SGCB - Carrier frequency 1/525 - Variants in NM_000232 (7) c.T552G:p.Y184X; c.C452G:p.T151R; c.C341T:p.S114F; c.T323G:p.L108R; c.T299A:p.M100K; c.G272T:p.R91L; c.G272C:p.R91P

Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)-253280 - Gene:POMGNT1 - Carrier frequency 1/708 - Variants in NM_001243766 (11) c.1413+211G>T; c.1539+1G>A; c.C1478G:p.P493R; c.G1469A:p.C490Y; c.G1425A:p.W475X; c.1350-1354del:p.450-452del; c.G1342C:p.G448R; c.C1324T:p.R442C; c.T1319G:p.L440R; c.1284+80A>G; c.G1274C:p.W425S

Myopathy due to myoadenylate deaminase deficiency-612874 - Gene:AMPD1 - Carrier frequency 1/501 - Variants in NM_000036 (3) c.G1361A:p.R454H; c.C1249T:p.R417W

Nephrotic syndrome, type 1 (Finnish Type)-256300 - Gene:NPHS1 - Carrier frequency 1/501 - Variants in NM_004646 (17) c.C3422T:p.S1141F; c.C3418T:p.R1140C; c.3387+148A>G; c.3357-3358insGG:p.T1120fs; c.C3325T:p.R1109X; c.G1583T:p.C528F; c.C1555T:p.P519S; c.1481delC:p.S494fs; c.C320T:p.A107V; c.G319A:p.A107T; c.G313A:p.D105N; c.C286G:p.L96V; c.248-249insA:p.Y83-R84delinsX; c.G191C:p.W64S; c.139delG:p.A47fs; c.121-122del:p.41-41del; c.58+1G>T

Neutropenia, severe congenital 3, autosomal recessive-610738 - Gene:HAX1 - Carrier frequency 1/126 - Variants in NM_001018837 (8) c.91delG; c.121-125insG; c.130insA; c.C112T:p.R38X; c.T245G:p.L82R; c.424insG; c.T277C:p.F93L; c.C424T:p.Q142X

Niemann-Pick Disease, Type C2-607625 - Gene:NPC2 - Carrier frequency 1/867 - Variants in NM_006432 (2) c.G58T:p.E20X; c.27delG:p.L9fs

Niemann-Pick Disease: Type C1-257220 - Gene:NPC1 - Carrier frequency 1/181 - Variants in NM_000271 (9) c.A3467G:p.N1156S; c.A3263G:p.Y1088C; c.T3182C:p.I1061T; c.G3160A:p.A1054T; c.C3107T:p.T1036M; c.C3104T:p.A1035V; c.C2793T:p.N931N; c.A2783C:p.Q928P; c.G2665A:p.V889M

Niemann-Pick disease, type A-257200 - Gene:SMPD1 - Carrier frequency 1/501 - Variants in NM_000543 (5) c.T788A:p.L263X; c.T911C:p.L304P; c.991delC:p.P331fs; c.G1493T:p.R498L; c.G1735A:p.G579S

Niemann-Pick disease, type B-607616 - Gene:SMPD1 - Carrier frequency 1/225 - Variants in NM_000543 (11) c.G730A:p.G244R; c.G742A:p.E248K; c.841-842insTCCCCGCA:p.I281fs; c.C880A:p.Q294K; c.994delC; c.G1006A:p.G336S; c.G1026T:p.W342C; c.1496-1498TAC; c.C1598T:p.P533L; c.1828_1830del:p.610_610del

OTC deficiency / Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome-238970 - Gene:SLC25A15 - Carrier frequency 1/142 - Variants in NM_014252 (6) c.562-564delTTC; c.C535T:p.R179X; c.G538A:p.E180K; c.553-555del:p.185-185del; c.C564G:p.F188L; c.G569A:p.G190D

Peroxisomal acyl-CoA oxidase deficiency-264470 - Gene:ACOX1 - Carrier frequency 1/969 - Variants in NM_001185039 (5) c.A812G:p.Q271R; c.A718G:p.M240V; c.G418T:p.G140C; c.C328T:p.R110X; c.258_275del:p.86_92del

Prothrombin thrombophilia / Factor II deficiency -613679 - Gene:F2 - Carrier frequency 1/708 - Variants in NM_000506 (13) c.G598A:p.E200K; c.734delT:p.L245fs; c.C940T:p.R314C; c.G1027A:p.E343K; c.G1054A:p.E352K; c.T1139C:p.M380T; c.C1273T:p.R425C; c.G1274A:p.R425H; c.G1292A:p.R431H; c.C1381T:p.R461W; c.C1785G:p.D595E; c.G1787T:p.R596L; c.G1802T:p.G601V

Pycnodysostosis-265800 - Gene:CTSK - Carrier frequency 1/225 - Variants in NM_000396 (2) c.A990G:p.X330W; c.T926C:p.L309P

Pyruvate dehydrogenase E1-beta deficiency-614111 - Gene:PDHB - Carrier frequency 1/251 - Variants in NM_000925 (2) c.C1030T:p.P344S; c.A395G:p.Y132C

Retinitis pigmentosa 59-613861 - Gene:DHDDS - Carrier frequency 1/501 - Variants in NM_001243564 (1) c.A124G:p.K42E

Rhizomelic chondrodysplasia punctata, type 1-215100 - Gene:PEX7 - Carrier frequency 1/159 - Variants in NM_000288 (7) c.G649A:p.G217R; c.C653T:p.A218V; c.C694T:p.R232X; c.A854G:p.H285R; c.870-871insCAA:p.C290delinsCQ; c.T875A:p.L292X; c.903+1G>C

Salla disease-604369 - Gene:SLC17A5 - Carrier frequency 1/578 - Variants in NM_012434 (8) c.C1090T:p.R364C; c.1007-1008del:p.336-336del; c.C1001G:p.P334R; c.G983A:p.G328E; c.507delA:p.A169fs; c.A406G:p.K136E; c.A406T:p.K136X; c.G309A:p.W103X

Segawa syndrome, recessive (tyrosine hydroxylase deficiency)-605407 - Gene:TH - Carrier frequency 1/708 - Variants in NM_000360 (2) c.T614C:p.L205P; c.G605A:p.R202H

Sjogren-Larsson syndrome-270200 - Gene:ALDH3A2 - Carrier frequency 1/251 - Variants in NM_000382 (1) c.C943T:p.P315S

Smith-Lemli-Opitz syndrome-270400 - Gene:DHCR7 - Carrier frequency 1/88 - Variants in NM_001163817 (29) c.G1342A:p.E448K; c.G1337A:p.R446Q; c.T470C:p.L157P; c.G1228A:p.G410S; c.C1210T:p.R404C; c.G1139A:p.C380Y; c.G1055A:p.R352Q; c.C1054T:p.R352W; c.G976T:p.V326L; c.964-1G>C; c.G744T:p.W248C; c.G730A:p.G244R; c.G725A:p.R242H; c.C724T:p.R242C; c.C506T:p.S169L; c.G453A:p.W151X; c.G452C:p.W151S; c.A356T:p.H119L; c.T326C:p.L109P; c.G321C:p.Q107H; c.T296C:p.L99P; c.C292T:p.Q98X; c.C278T:p.T93M; c.T203C:p.L68P; c.A185T:p.D62V; c.T176G:p.M59R; c.A172G:p.I58V; c.C151T:p.P51S; c.C149A:p.A50D

Spastic ataxia, Charlevoix-Saguenay type (ARSACS)-270550 - Gene:SACS - Carrier frequency 1/501 - Variants in NM_014363 (13) c.C12973T:p.R4325X; c.G12220C:p.A4074P; c.C12160T:p.Q4054X; c.G10907A:p.R3636Q; c.T9742C:p.W3248R; c.8844delT:p.P2948fs; c.C7504T:p.R2502X; c.6273delT; c.T5836C:p.W1946R; c.5263-5264del:p.1755-1755del; c.C4933T:p.R1645X; c.T3161C:p.F1054S; c.C3107T:p.P1036L

Tay-Sachs disease-272800 - Gene:HEXA - Carrier frequency 1/284 - Variants in NM_000520 (21) c.1330+293G>C; c.G1360A:p.G454S; c.C1351G:p.L451V; c.1278-1279insTATC; c.G1260C:p.W420C; c.C1177T:p.R393X; c.G1176A:p.W392X; c.G987A:p.W329X; c.805+1G>A; c.G805A:p.G269S; c.G772C:p.D258H; c.G749A:p.G250D; c.C745T:p.R249W; c.C739T:p.R247W; c.C540G:p.Y180X; c.T538C:p.Y180H; c.G533T:p.R178L; c.G533A:p.R178H; c.C532T:p.R178C; c.G509A:p.R170Q; c.C508T:p.R170W

Thrombocytopenia, congenital amegakaryocytic-604498 - Gene:MPL - Carrier frequency 1/914 - Variants in NM_005373 (2) c.79+2T>A; c.C127T:p.R43X

Usher syndrome, type 1F-602083 - Gene:PCDH15 - Carrier frequency 1/80 - Variants in NM_001142767 (2) c.G674A:p.G225D; c.C622T:p.R208X

Usher syndrome, type 3A-276902 - Gene:CLRN1 - Carrier frequency 1/537 - Variants in NM_001195794 (10) c.T300G:p.Y100X; c.T221C:p.L74P; c.T359A:p.M120K; c.C189A:p.Y63X; c.T144G:p.N48K; c.G127A:p.G43R; c.T118G:p.C40G; c.C92T:p.P31L; c.A57T:p.A19A; c.A6C:p.P2P

Walker-Warburg syndrome-253800 - Gene:FKTN - Carrier frequency 1/355 - Variants in NM_001079802 (1) c.1167insA

Wilson disease-277900 - Gene:ATP7B - Carrier frequency 1/80 - Variants in NM_000053 (29) c.C2586A:p.H862Q; c.A2480G:p.H827R; c.3060+16G>T; c.G2433A:p.A811A; c.G2424A:p.L808L; c.C2394T:p.N798N; c.G2388A:p.A796A;

c.G2352A:p.T784T; c.C2351T:p.T784M; c.T2332C:p.C778R; c.C2309T:p.T770M; c.G2285A:p.R762Q; c.2866-13G>C; c.C2135T:p.A712V; c.G2337A:p.W779X; c.G2336A:p.W779X; c.G2333T:p.R778L; c.C2332G:p.R778G; c.C2332T:p.R778W; c.2304-2305insC:p.M769fs; c.A2305G:p.M769V; c.2304delC; c.C2297G:p.T766R; c.G2293A:p.D765N; c.G2128A:p.G710S; c.T2123C:p.L708P; c.2122-8T>G; c.G1366C:p.V456L; c.1340delAAAC:p.Q447LfsX50

Zellweger syndrome-214100 - Gene:PEX10 - Carrier frequency 1/138 - Variants in NM_002617 (10) c.G881A:p.W294X; c.874-875delCT; c.C870G:p.H290Q; c.C868G:p.H290D; c.G843A:p.R281R; c.G835T:p.E279X; c.A820G:p.T274A; c.814-815del:p.272-272del; c.C730T:p.R244X; c.704_705insA:p.Q235fs

Zellweger syndrome-1-214100 - Gene:PEX1 - Carrier frequency 1/138 - Variants in NM_000466 (2) c.2537-2546TCATGGTG; c.G2528A:p.G843D

von Willebrand disease types 2A, 2B, 2M and 2N-613554 - Gene:VWF - Carrier frequency 1/19 - Variants in NM_000552 (22) c.2540-2541insA:p.N847fs; c.2516delG:p.G839fs; c.T2451A:p.H817Q; c.G2447A:p.R816Q; c.C2446T:p.R816W; c.2443-1G>C; c.C2435T:p.P812L; c.2435delC:p.P812fs; c.G2411T:p.C804F; c.A2398G:p.M800V; c.A2384G:p.Y795C; c.C2372T:p.T791M; c.A2365G:p.T789A; c.G2363A:p.C788Y; c.T2362C:p.C788R; c.G2359A:p.E787K; c.G2354A:p.G785E; c.G2345C:p.R782P; c.C2344T:p.R782W; c.G2313T:p.M771I; c.A2287G:p.R763G; c.A2284G:p.K762E

Duchenne muscular dystrophy-310200 - Gene:DMD - Carrier frequency 1/1651 - Coding Region of NM_000109 (+18 variants)

G6PD deficiency / Favism-134700 - Gene:G6PD - Carrier frequency 1/51 - Coding Region of NM_000402 (+56 variants)

Adrenoleukodystrophy-300100 - Gene:ABCD1 - Carrier frequency 1/10001 - Variants in NM_000033 (35) c.67-83del:p.23-28del; c.G395A:p.W132X; c.C420A:p.I140I; c.G421A:p.A141T; c.A443G:p.N148S; c.T520G:p.Y174D; c.G796A:p.G266R; c.C838T:p.R280C; c.G871A:p.E291K; c.871-873del:p.291-291del; c.901-1G>A:IVS1-1G>A; c.C1165G:p.R389G; c.G1202A:p.R401Q; c.C1252T:p.R418W; c.1364-1365insC:p.V455fs; c.C1390T:p.R464X; c.1394-2A>G; c.1415-1416del:p.472-472del; c.G1429T:p.E477X; c.C1451G:p.P484R; c.C1544T:p.S515F; c.1551delC:p.F517fs; c.C1552T:p.R518W; c.T1592C:p.L531P; c.1634+1G>A:IVS6+1G>A; c.1635-2A>G:IVS6-2A>G; c.1780+4G>A; c.C1817T:p.S606L; c.C1849T:p.R617C; c.G1850A:p.R617H; c.1865+1G>A:IVS8+1G>A; c.1866-10G>A:IVS8-10G>A; c.1937delC; c.1937-1938insGG:p.A646fs; c.G1950A:p.A650A

Alport syndrome, X-linked-301050 - Gene:COL4A5 - Carrier frequency 1/25001 - Variants in NM_000495 (24) c.4689-30-4689-9delinsCA; c.T4690C:p.C1564R; c.G4691A:p.C1564Y; c.G4691C:p.C1564S; c.T4699C:p.C1567R; c.G4702A:p.E1568K; c.4732-4738del:p.1578-1580del; c.C4751T:p.P1584L; c.T4756C:p.C1586R; c.G4757T:p.C1586F; c.G4766T:p.G1589V; c.T4768G:p.W1590G; c.4778-4779del:p.1593-1593del; c.G4787A:p.G1596D; c.A4790G:p.Y1597C; c.T4791A:p.Y1597X; c.G4803A:p.M1601I; c.4831delG:p.G1611fs; c.T4894G:p.C1632G; c.G4913A:p.C1638Y; c.4938-4939insT:p.S1646fs; c.T4946G:p.L1649R; c.4968delC:p.D1656fs; c.G4976A:p.S1659N

Androgen insensitivity-300068 - Gene:AR - Carrier frequency 1/20001 - Variants in NM_000044 (37) c.G4A:p.E2K; c.T521G:p.L174X; c.C1645T:p.P549S; c.T1748A:p.F583Y; c.C1937A:p.A646D; c.G1943A:p.S648N; c.T2033C:p.L678P; c.A2069C:p.H690P; c.T2123G:p.L708R; c.C2137T:p.L713F; c.G2157A:p.W719X; c.G2164A:p.A722T; c.G2191A:p.V731M; c.C2222G:p.S741C; c.G2231A:p.G744E; c.A2291G:p.Y764C; c.C2323T:p.R775C; c.G2324A:p.R775H; c.G2343T:p.M781I; c.A2362G:p.M788V; c.G2391A:p.W797X; c.T2423C:p.M808T; c.C2521T:p.R841C; c.G2522A:p.R841H; c.G2567A:p.R856H; c.C2571G:p.F857L; c.T2596C:p.S866P; c.G2599A:p.V867M; c.T2610G:p.I870M; c.C2623T:p.H875Y; c.A2632G:p.T878A; c.C2633G:p.T878S; c.A2650T:p.K884X; c.C2667T:p.S889S; c.A2708G:p.Q903R

Charcot-Marie-Tooth Disease with Deafness: X-Linked: PRPS1 Related (CMTX5)-311070 - Gene:PRPS1 - Carrier frequency 1/100001 - Variants in NM_002764 (8) c.A129C:p.E43D; c.G154C:p.D52H; c.G193A:p.D65N; c.G259A:p.A87T; c.A341G:p.N114S; c.T344C:p.M115T; c.C385A:p.L129I; c.A398C:p.Q133P

Charcot-Marie-Tooth neuropathy, X-linked-302800 - Gene:GJB1 - Carrier frequency 1/16668 - Variants in NM_000166 (25) c.G37T:p.V13L; c.C43T:p.R15W; c.T89A:p.I30N; c.G123C:p.E41D; c.T145C:p.S49P; c.C164T:p.T55I; c.G187A:p.V63I; c.A194G:p.Y65C; c.C223T:p.R75W; c.224delG:p.R75fs; c.C254G:p.S85C; c.G283A:p.V95M; c.304delGAG:p.102delE; c.T397C:p.W133R; c.T407C:p.V136A; c.G415A:p.V139M; c.C424T:p.R142W; c.T467G:p.L156R; c.C514T:p.P172S; c.G536A:p.C179Y; c.G556A:p.E186K; c.561-562insACCGTCTTC:p.K187delinsKTVF; c.A614G:p.N205S; c.C658T:p.R220X

Chronic granulomatous disease, X-linked-306400 - Gene:CYBB - Carrier frequency 1/250001 - Variants in NM_000397 (45) c.A532C:p.T178P; c.G535A:p.G179R; c.C578T:p.S193F; c.G607T:p.E203X; c.T613A:p.F205I; c.C625T:p.H209Y; c.A626G:p.H209R; c.T627A:p.H209Q; c.C654A:p.G218G; c.C664A:p.H222N; c.A665G:p.H222R; c.G667T:p.G223X; c.G668T:p.G223V; c.C671G:p.A224G; c.A674T:p.E225V; c.T730C:p.C244R; c.G731A:p.C244Y; c.C907A:p.H303N; c.C911G:p.P304R; c.A919C:p.T307P; c.G925A:p.E309K; c.G965A:p.G322E; c.A973T:p.I325F; c.T997C:p.S333P;

c.C1012T:p.H338Y; c.C1016A:p.P339H; c.T1025A:p.L342Q; c.C1031T:p.S344F; c.G1067C:p.R356P; c.1138-1139insG:p.W380fs; c.G1166A:p.G389E; c.G1166C:p.G389A; c.T1214G:p.M405R; c.G1222A:p.G408R; c.G1223A:p.G408E; c.C1244A:p.P415H; c.T1259C:p.L420P; c.T1264C:p.S422P; c.1314+19C>G; c.A1499G:p.D500G; c.T1514G:p.L505R; c.T1546C:p.W516R; c.G1548T:p.W516C

Emery-Dreifuss muscular dystrophy 1, X-linked-310300 - Gene:EMD - Carrier frequency 1/150001 - Variants in NM_000117 (4) c.G470A:p.R157Q; c.C547A:p.P183T; c.C548A:p.P183H

Fabry disease-301500 - Gene:GLA - Carrier frequency 1/1501 - Variants in NM_000169 (33) c.G1145A:p.C382Y; c.T1095A:p.Y365X; c.G1081C:p.G361R; c.C1066T:p.R356W; c.G1025A:p.R342Q; c.C1024T:p.R342X; c.G1020A:p.W340X; c.G983C:p.G328A; c.G982A:p.G328R; c.C979A:p.Q327K; c.G902T:p.R301L; c.C890T:p.S297F; c.G888A:p.M296I; c.A886G:p.M296V; c.G861A:p.W287X; c.C835G:p.Q279E; c.A815G:p.N272S; c.T806C:p.V269A; c.A797T:p.D266V; c.A791T:p.D264V; c.G680A:p.R227Q; c.C679T:p.R227X; c.C666A:p.Y222X; c.369+2T>G; c.C334T:p.R112C; c.286-287insA:p.M96fs; c.T166G:p.C56G; c.G131A:p.W44X; c.C118T:p.P40S; c.A101G:p.N34S; c.G58C:p.A20P; c.-10C>T

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

Hemophilia A, factor VIII deficiency, X-linked-306700 - Gene:F8 - Carrier frequency 1/3001 - Variants in NM_000132 (119) c.G572T:p.R191L; c.C571T:p.R191X; c.G563A:p.R188H; c.C562T:p.R188C; c.C551T:p.P184L; c.C550T:p.P184S; c.507-511del:p.169-171del; c.G339T:p.W113C; c.294delG:p.G98fs; c.G278T:p.R93L; c.G6683A:p.R2228Q; c.C277T:p.R93X; c.G226C:p.A76P; c.T149C:p.L50S; c.G140A:p.R47H; c.C139T:p.R47C; c.G128T:p.R43L; c.C127T:p.R43C; c.C113T:p.T38I; c.C110A:p.P37Q; c.G101A:p.R34H; c.C91T:p.R31X; c.59-60del:p.20-20del; c.C6413A:p.S2138Y; c.6412-6413del:p.2138-2138del; c.G6404C:p.R2135P; c.C6403T:p.R2135X; c.A6371G:p.Y2124C; c.T6360G:p.F2120L; c.A6278G:p.D2093G; c.5961delA:p.K1987fs; c.C5953T:p.R1985X; c.C5938T:p.H1980Y; c.G5936T:p.G1979V; c.G5900A:p.G1967D; c.G5882A:p.W1961X; c.G5879T:p.R1960L; c.C5878T:p.R1960X; c.4858delC:p.L1620X; c.4328-4331del:p.1443-1444del; c.4121-4124del:p.1374-1375del; c.2962-2963del:p.988-988del; c.2945-2946insA:p.N982fs; c.C2440T:p.R814X; c.A2383T:p.R795X; c.G2215A:p.E739K; c.T2029C:p.F677L; c.C1988T:p.A663V; c.C1965G:p.Y655X; c.T1958C:p.V653A; c.G1957A:p.V653M; c.A1892G:p.N631S; c.C1834T:p.R612C; c.A1814C:p.Y605S; c.G1812C:p.W604C; c.G1808T:p.S603I; c.C1804T:p.R602X; c.T1786C:p.S596P; c.T1754C:p.I585T; c.C1750A:p.Q584K; c.C1730T:p.S577F; c.T1729C:p.S577P; c.G1726T:p.E576X; c.A1682G:p.D561G; c.A1660G:p.S554G; c.G1649A:p.R550H; c.C1648T:p.R550C; c.C1636T:p.R546W; c.G1630A:p.D544N; c.T1481C:p.I494T; c.A1475G:p.Y492C; c.T1474C:p.Y492H; c.A1226G:p.E409G; c.T1214G:p.I405S; c.1197-1198del:p.399-400del; c.C1175T:p.S392L; c.T1174C:p.S392P; c.G1172A:p.R391H; c.C1171T:p.R391C; c.1078-1079del:p.360-360del; c.1075-1078del:p.359-360del; c.G1064A:p.R355Q; c.C1063T:p.R355X; c.G1043C:p.C348S; c.T1042C:p.C348R; c.G1033C:p.V345L; c.T1026A:p.Y342X; c.G986A:p.C329Y; c.T985C:p.C329R; c.T980C:p.L327P; c.943delG:p.A315fs; c.A940G:p.T314A; c.T935C:p.F312S; c.C923T:p.S308L; c.907delG:p.A303fs; c.G902T:p.R301L; c.A896T:p.N299I; c.C881T:p.T294I; c.A872G:p.E291G; c.T854G:p.V285G; c.849delT:p.P283fs; c.G832A:p.G278R; c.G822T:p.W274C; c.G797A:p.G266E; c.T592G:p.C198G; c.C566T:p.S189L; c.A554C:p.K185T; c.G541A:p.V181M; c.C493T:p.P165S; c.G491T:p.G164V; c.C410T:p.T137I; c.A404G:p.D135G; c.A398G:p.Y133C; c.A396C:p.E132D; c.G121T:p.G41C; c.A104G:p.Y35C; c.A89T:p.E30V; c.T77G:p.L26R; c.C43T:p.R15X

Hemophilia B, factor IX deficiency-306900 - Gene:F9 - Carrier frequency 1/15001 - Variants in NM_000133 (51) c.A278G:p.D93G; c.A287C:p.Q96P; c.C301G:p.P101A; c.G316A:p.G106S; c.G328A:p.D110N; c.A329G:p.D110G; c.C571T:p.R191C; c.G572A:p.R191H; c.A580G:p.T194A; c.C655T:p.Q219X; c.C676T:p.R226W; c.G677A:p.R226Q; c.G679T:p.V227F; c.G682C:p.V228L; c.G697A:p.A233T; c.C709T:p.Q237X; c.A710T:p.Q237L; c.A872T:p.E291V; c.C880T:p.R294X; c.G881A:p.R294Q; c.C892T:p.R298X; c.C907T:p.H303Y; c.A917G:p.N306S; c.C998T:p.P333L; c.G1009C:p.A337P; c.C1025T:p.T342M; c.T1031C:p.I344T; c.T1058C:p.V353A; c.G1064T:p.G355V; c.G1069A:p.G357R; c.G1070A:p.G357E; c.G1088T:p.G363V; c.G1120T:p.V374F; c.C1135T:p.R379X; c.G1136A:p.R379Q; c.T1144A:p.C382S; c.C1150T:p.R384X; c.G1151C:p.R384P; c.T1169C:p.I390T; c.A1180G:p.M394V; c.G1187C:p.C396S; c.G1189C:p.A397P; c.C1217T:p.S406L; c.G1226T:p.G409V; c.G1228C:p.D410H; c.A1231G:p.S411G; c.G1232T:p.S411I; c.C1240A:p.P414T; c.T1256A:p.V419E; c.C1307A:p.A436E; c.T1328C:p.I443T

Hypohidrotic Ectodermal Dysplasia, X-Linked-305100 - Gene:EDA - Carrier frequency 1/50001 - Variants in NM_001005609 (3) c.C1007T:p.T336M; c.G1039A:p.A347T; c.C1066G:p.Q356E

Mucopolysaccharidosis II / Hunter Syndrome: X-linked-309900 - Gene:IDS - Carrier frequency 1/51501 - Variants in NM_001166550 (15) c.G1505C:p.W502S; c.G1466C:p.G489A; c.G1464T:p.M488I; c.G1425A:p.W475X; c.G1403T:p.R468L; c.G1403A:p.R468Q; c.C1402T:p.R468W; c.A1394T:p.Q465L; c.C1327T:p.R443X; c.T1264G:p.C422G; c.A404G:p.K135R; c.G401A:p.G134E; c.C359A:p.P120H; c.G253A:p.A85T; c.15-49A>T
Myotubular myopathy, X-linked-310400 - Gene:MTM1 - Carrier frequency 1/25001 - Variants in NM_000252 (3) c.A566G:p.N189S; c.C670T:p.R224X; c.C721T:p.R241C
Pyruvate dehydrogenase E1-alpha deficiency: X-linked-312170 - Gene:PDHA1 - Carrier frequency 1/375001 - Variants in NM_000284 (6) c.C522G:p.F174L; c.A555C:p.L185F; c.T634A:p.Y212N; c.A680C:p.D227A; c.C694G:p.R232G; c.1145_1146insATCA
Retinoschisis: X-linked-312700 - Gene:RS1 - Carrier frequency 1/6251 - Variants in NM_000330 (5) c.G276C:p.W92C; c.T267A:p.Y89X; c.A266G:p.Y89C; c.C262T:p.Q88X; c.253_255del:p.85_85del
Severe combined immunodeficiency, X-linked-300400 - Gene:IL2RG - Carrier frequency 1/100001 - Variants in NM_000206 (18) c.C923A:p.S308X; c.T878A:p.L293Q; c.C865T:p.R289X; c.G710A:p.W237X; c.C664T:p.R222C; c.T662C:p.F221S; c.T515C:p.L172P; c.T458A:p.I153N; c.T455C:p.V152A; c.454+1G>A; c.T452C:p.L151P; c.A355T:p.K119X; c.T343C:p.C115R; c.G341A:p.G114D; c.A314G:p.Y105C; c.A292T:p.K98X; c.269+208G>T; c.T186A:p.C62X

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