Donor 5633

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 12/21/18

Donor Reported Ancestry: West Indies

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Carrier: Alpha Thalassemia (See DNA results below)	Carrier testing recommended for those using this donor Reduced risk to be a carrier for sickle cell anemia and beta thalassemia.
Cystic Fibrosis (CF) carrier screening	Negative by gene sequencing in the CFTR gene	1/1000
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/1061
Expanded Genetic Disease Testing Panel attached- 289 diseases by gene sequencing	Carrier: Congenital Disorder of Glycosylation: Type 1A (PMM2) Carrier: Stargardt Disease (ABCA4) Carrier: Alpha Thalassemia- trait carrier (-a/-a) in the HBA1 and HBA2 genes Negative for other genes sequenced	Carrier testing recommended for those using this donor

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

sema4
a Mount Sinai venture

Patient	Sample	Referring Doctor
Patient Name: Donor 5633 **Date of Birth:** [redacted] **Reference #:** P0715517 **Indication:** Carrier Testing **Test Type:** Alpha Thalassemia Carrier Screen	**Specimen Type:** Blood **Lab #:** [redacted] **Date Collected:** 12/7/2018 **Date Received:** 12/8/2018 **Final Report:** 12/20/2018	[redacted] **Fairfax Cryobank, Inc.** [redacted] [redacted] [redacted] Fax: [redacted]

RESULTS

POSITIVE for alpha-thalassemia
HBA1 copy number: 2
HBA2 copy number: 0
Two copies of the alpha 3.7 deletion detected
HBA1 and *HBA2* sequence analysis: No pathogenic or likely pathogenic variants identified
Alpha-thalassemia trait carrier (-a/-a)

Genes analyzed: *HBA1* (NM_000558.4) and *HBA2* (NM_000517.4)
Inheritance: Autosomal Recessive

Recommendations
Testing of the patient's partner and genetic counseling are recommended.

Individuals of Asian, African, Hispanic and Mediterranean ancestry should also be screened for hemoglobinopathies by CBC and hemoglobin electrophoresis.

Interpretation
This patient carries a homozygous alpha 3.7 deletion, resulting in the loss of two copies of the alpha-globin gene and is therefore a carrier of the alpha-thalassemia trait (-a/-a). No pathogenic or likely pathogenic variants were identified by sequence analysis.

Typically, individuals have four functional alpha-globin genes: 2 copies of *HBA1* and 2 copies of *HBA2*, whose expression is regulated by a cis-acting regulatory element HS-40. Alpha-thalassemia carriers have three (silent carrier) or two (carrier of the alpha-thalassemia trait) functional alpha-globin genes with or without a mild phenotype.

What is alpha-thalassemia?
Alpha-thalassemia is an autosomal recessive condition that affects the red blood cells. It can affect people of any ethnicity, but is more common in people who can trace their ancestry to Southeast Asia, India, equatorial Africa, the Mediterranean, or the Arabian Peninsula. There are two major forms of alpha-thalassemia:

- Hemoglobin Bart syndrome is caused by a loss of all 4 alpha-globin genes (--/--). It is very severe, and fetuses are either stillborn or die shortly after birth.
- Alpha-thalassemia (also called HbH disease) is caused by a loss of 3 alpha-globin genes (-a/--). This disease results in anemia, an enlarged spleen, and mild jaundice. Most individuals are mildly disabled by this

condition. Some people with more severe disease require frequent blood transfusions. The type of disease as well as the severity of symptoms can be predicted based on the genetic variants detected. Carriers may have mild anemia.

Test Methods and Comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Multiplex Ligation-Dependent Probe Amplification (MLPA) (Analytical Detection Rate >99%)
MLPA® probe sets and reagents from MRC-Holland were used for copy number analysis of specific targets versus known control samples. False positive or negative results may occur due to rare sequence variants in target regions detected by MLPA probes. Analytical sensitivity and specificity of the MLPA method are both 99%.

The copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation are assessed. This test is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Carriers of alpha-thalassemia with three or more *HBA* copies on one chromosome, and one or no copies on the other chromosome, may not be detected. With the exception of triplications, other benign alpha-globin gene polymorphisms will not be reported. Analyses of *HBA1* and *HBA2* are performed in association with long-range PCR of the coding regions followed by short-read sequencing.

Long-Range PCR (Analytical Detection Rate >99%)
A long-range PCR was performed to generate a locus-specific amplicon for *HBA1* and *HBA2*. The PCR product was then prepared for short-read NGS sequencing as described below and sequenced. Sequenced reads were mapped back to the original genomic loci and converted to VCF files as described below.

Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)
Samples sequenced on the Illumina HiSeq 2500 platform in the Rapid Run mode or the Illumina NovaSeq platform in the Xp workflow, using 100 bp paired-end reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house.

The coding exons and splice junctions of the tested genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing.

This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions may not be detected, including, but not limited to, UTRs, promoters, and deep intronic areas. This technology may not detect all small insertion/deletions and is not diagnostic for repeat expansions and structural genomic variation. In addition, a mutation(s) in a gene not included on the panel could be present in this patient.

Variant interpretation and classification was performed based on the American College of Medical Genetics Standards and Guidelines for the Interpretation of Sequence Variants (Richards et al, 2015). All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis will not be reported.

Sanger Sequencing (Confirmation method) (Accuracy >99%)
Sanger sequencing, as indicated, was performed using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage (<20 reads) or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

Please note these tests were developed and their performance characteristics were determined by Mount Sinai Genomics, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected

status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

SELECTED REFERENCES

Carrier Screening
Grody W et al. ACMG position statement on prenatal/preconception expanded carrier screening. *Genet Med.* 2013 15:482-3.
Variant Classification:
Richards S et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 May;17(5):405-24

Additional disease-specific references available upon request.

This case has been reviewed and electronically signed by Rebekah Zimmerman, Ph.D., FACMG, Laboratory Director

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D.

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank - [redacted]
[redacted]
Physician: [redacted]
Report Generated: 2018-02-19

Donor 5633

DOB: [redacted]
Gender: Male
Ethnicity: Latin American
Procedure ID: 112233
Kit Barcode: [redacted]
Specimen: Blood, #114417
Specimen Collection: 2018-02-02
Specimen Received: 2018-02-03
Specimen Analyzed: 2018-02-19

TEST INFORMATION

Test: CarrierMap^{SEQ} (Genotyping & Sequencing)
Panel: CarrierMap Expanded v3 - Sequencing
Diseases Tested: 289
Genes Tested: 278
Genes Sequenced: 273

Partner Not Tested

SUMMARY OF RESULTS: MUTATION(S) IDENTIFIED

Disease	Donor 5633	Partner Not Tested
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2) ○ High Impact	Carrier (1 abnormal copy) Mutation: c.470T>C (p.F157S) Method: Genotyping & Sequencing **Reproductive Risk & Next Steps**: Reproductive risk detected. Consider partner testing.	
Stargardt Disease (ABCA4) ○ High Impact	Carrier (1 abnormal copy) Mutation: c.2863G>T Method: Sequencing **Reproductive Risk & Next Steps**: Reproductive risk detected. Consider partner testing.	

No other pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.

For additional disease information, please visit recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

Assay performed by
Reprogenetics
CLIA ID: 31D1054821
3 Regent Street, Livingston, NJ 07039
Lab Technician: Bo Chu

Recombine CLIA # 31D2100763
Reviewed by Pere Colls, PhD, HCLD, Lab Director

ADDITIONAL RESULTS: NO INCREASED REPRODUCTIVE RISK

The following results are not associated with an increased reproductive risk.

Disease (Gene)	**Donor 5633**	**Partner Not Tested**
Spinal Muscular Atrophy: SMN1 Linked (SMN1)*	SMN1 Copy Number: 2 or more copies Method: Genotyping & dPCR	

*SMA Risk Information for Individuals with No Family History of SMA

	Detection Rate	Pre-Test Carrier Risk	Post-Test Carrier Risk (2 SMN1 copies)	Post-Test Carrier Risk (3 SMN1 copies)
European	95%	1/35	1/632	1/3,500
Ashkenazi Jewish	90%	1/41	1/350	1/4,000
Asian	93%	1/53	1/628	1/5,000
African American	71%	1/66	1/121	1/3,000
Hispanic	91%	1/117	1/1,061	1/11,000

For other unspecified ethnicities, post-test carrier risk is assumed to be <1%. For individuals with multiple ethnicities, it is recommended to use the most conservative risk estimate.

Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2)

Congenital Disorder of Glycosylation (CDG) is a clinically heterogeneous disorder which affects multiple parts of the body. CDG Type 1A is caused by mutations in the PMM2 gene, which is normally responsible for synthesizing and attaching sugars to proteins. The most severe cases of this disease are characterized by hydrops fetalis, which leads to death in the womb or soon after birth. Most affected individuals, however, develop signs and symptoms of the condition during infancy, including: weak muscle tone (hypotonia), developmental delay, and a failure to gain weight and grow at the expected rate (failure to thrive). Individuals who survive infancy exhibit intellectual disability, stroke-like episodes, and failure to walk independently. Most affected females do not go through puberty. Affected males experience normal puberty but often have small testes.

○ High Impact

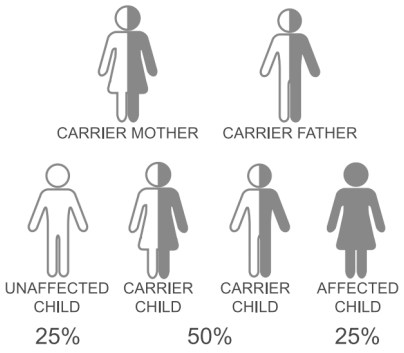
These diseases have a significant impact on life expectancy and quality of life.

Clinical Information

- ✓ Physical Impairment
- ✓ Cognitive Impairment
- ✓ Shortened Lifespan

Effective Treatment

Inheritance:
Autosomal Recessive



Prognosis

Prognosis varies greatly. Approximately 20% of individuals with the severe form die in the first year of life. Individuals with a milder presentation may have very few symptoms. However, the most common presentation is a multisystem disorder with intellectual disability.

Treatment

Treatment includes feeding support in infancy with gastrostomy tubes and specialized food; occupational therapy, physical therapy, and speech therapy for developmental delay; hydration and physical therapy for stroke-like episodes; and surgery for scoliosis.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Danish	90.00%	1/71	1/710
Dutch	39.29%	1/68	1/112
European	55.33%	1/71	1/159

For other unspecified ethnicities, post-test carrier risk is assumed to be <1%. For individuals with multiple ethnicities, it is recommended to use the most conservative risk estimate.

To learn more, visit recombine.com/diseases/congenital-disorder-of-glycosylation-type-1a-pmm2-related

Stargardt Disease (ABCA4)

Stargardt disease is a genetic eye disorder that causes progressive vision loss or macular degeneration. The disorder is caused by mutations in the ABCA4 gene, which normally provides instructions for making proteins that are found in light-sensing (photoreceptor) cells in the retina. Stargardt disease is one of the most frequent causes of macular degeneration in childhood; its onset occurs between 6 and 12 years of age. Visual acuity is severely reduced but peripheral visual fields remain normal throughout life. By 20 years of age, symptoms include wavy vision, blind spots, blurriness, impaired color vision, and difficulty adapting to dim lighting.

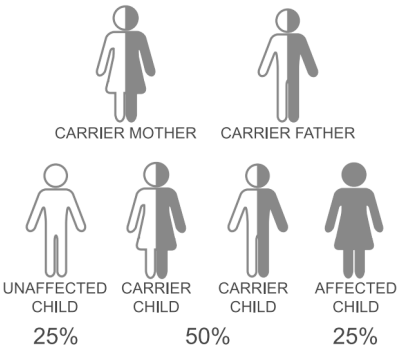
O High Impact

These diseases have a significant impact on life expectancy and quality of life.

Clinical Information

- ✓ Physical Impairment
- Cognitive Impairment
- Shortened Lifespan
- Effective Treatment

Inheritance: Autosomal Recessive



To learn more, visit recombine.com/diseases/stargardt-disease

Prognosis

The long term prognosis for patients with Stargardt disease is widely variable. General health is not affected and some affected individuals are still able to drive.

Treatment

No specific treatments are available for Stargardt disease. Many affected individuals use magnifiers for visual assistance and wear sunglasses to slow the development of the condition.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
General	17.51%	1/51	1/62

For other unspecified ethnicities, post-test carrier risk is assumed to be <1%. For individuals with multiple ethnicities, it is recommended to use the most conservative risk estimate.

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in the genes tested. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Sequencing: Sequencing is performed using a custom next-generation sequencing (NGS) platform. Only the described exons for each gene listed are sequenced. Variants outside of these regions may not be identified. Some splicing mutations may not be identified. Triplet repeat expansions, intronic mutations, and large insertions and deletions may not be detected. All identified variants are curated, and determination of the likelihood of their pathogenicity is made based on examining allele frequency, segregation studies, predicted effect, functional studies, case/control studies, and other analyses. All variants identified via sequencing that are reported to cause disease in the primary scientific literature will be reported. Variants considered to be benign and variants of unknown significance (VUS) are NOT reported. In the sequencing process, interval drop-out may occur, leading to intervals of insufficient coverage. Intervals of insufficient coverage will be reported if they occur.

Spinal Muscular Atrophy: Carrier status for SMA is assessed via copy number analysis by dPCR and via genotyping. Some individuals with a normal number of SMN1 copies (2 copies) may carry both copies of the gene on the same allele/chromosome; this analysis is not able to detect these individuals. Thus, a normal SMN1 result significantly reduces but does not eliminate the risk of being a carrier. Additionally, SMA may be caused by non-deletion mutations in the SMN1 gene; CarrierMap tests for some, but not all, of these mutations. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors. The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

This test was developed and its performance determined by Recombine, Inc., and it has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

Diseases & Mutations Assayed

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CYP11B1): Mutations (1): ♂ Genotyping | c.1343G>A (p.R448H) Sequencing | NM_000497:1-9

17-Alpha-Hydroxylase Deficiency (CYP17A1): Mutations (20): ♂ Genotyping | c.157_159delTTC (p.53delF), c.316T>C (p.S106P), c.715C>T (p.R239X), c.1024C>A (p.P342T), c.286C>T (p.R96W), c.1040G>A (p.R347H), c.1073G>A (p.R358Q), c.51G>A (p.W17X), c.340T>G (p.F114V), c.347A>T (p.D116V), c.1039C>T (p.R347C), c.1084C>T (p.R362C), c.1216T>C (p.W406R), c.985T>G (p.Y329D), c.601T>A (p.Y201N), c.81C>A (p.Y27X), c.287G>A (p.R96Q), c.1226C>G (p.P409R), c.1250T>G (p.F417C), c.278T>G (p.F93C) Sequencing | NM_000102:1-8

17-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD17B3): Mutations (8): ♂ Genotyping | c.695C>T (p.S232L), c.703A>G (p.M235V), c.239G>A (p.R80Q), c.608C>T (p.A203V), c.238C>T (p.R80W), c.166G>A (p.A56T), c.389A>G (p.N130S), c.803G>A (p.C268Y) Sequencing | NM_000197:1-11

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

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

3-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD3B2): Mutations (6): ♂ Genotyping | c.512G>A (p.W171X), c.742_747delGTCCGAinsAACTA (p.V248NfsR249X), c.745C>T (p.R249X), c.29C>A (p.A10E), c.424G>A (p.E142K), c.664C>A (p.P222T) Sequencing | NM_000198:2-4

3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related (MCCC1): Mutations (2): ♂ Genotyping | c.1155A>C (p.R385S), c.1310T>C (p.L437P) Sequencing | NM_020166:1-19

3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related (MCCC2): Mutations (8): ♂ Genotyping | c.295G>C (p.E99Q), c.499T>C (p.C167R), c.464G>A (p.R155Q), c.569A>G (p.H190R), c.803G>C (p.R268T), c.838G>T (p.D280Y), c.929C>G (p.P310R), c.1309A>G (p.I437V) Sequencing | NM_022132:1-17

3-Methylglutaconic Aciduria: Type 3 (OPA3): Mutations (3): ♂ Genotyping | c.415C>T (p.Q139X), c.320_337delAGCAGCGCCACAAGGAGG (p.Q108_E113del), c.143-1G>C Sequencing | NM_025136:1-2

3-Phosphoglycerate Dehydrogenase Deficiency (PHGDH): Mutations (7): ♂ Genotyping | c.1468G>A (p.V490M), c.403C>T (p.R135W), c.712delG (p.G238fsX), c.1273G>A (p.V425M), c.1117G>A (p.A373T), c.781G>A (p.V261M), c.1129G>A (p.G377S) Sequencing | NM_006623:1-12

5-Alpha Reductase Deficiency (SRD5A2): Mutations (10): ♂ Genotyping | c.736C>T (p.R246W), c.164T>A (p.L55Q), c.344G>A (p.G115D), c.547G>A (p.G183S), c.679C>T (p.R227X), c.682G>A (p.A228T), c.586G>A (p.G196S), c.692A>G (p.H231R), c.635C>G (p.P212R), c.591G>T (p.E197D) Sequencing | NM_000348:1-5

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS): Mutations (6): ♂ Genotyping | c.46C>T (p.R16C), c.74G>A (p.R25Q), c.155A>G (p.N52S), c.259C>T (p.P87S), c.286G>A (p.D96N), c.347A>G (p.D116G) Sequencing | NM_000317:1-6

ARSACS (SACS): Mutations (6): ♂ Genotyping | c.12973C>T (p.R4325X), c.7504C>T (p.R2502X), c.9742T>C (p.W3248R), c.8844delT (p.I2949fs), c.5836T>C (p.W1946R), c.3161T>C (p.F1054S) Sequencing | NM_014363:2-10

Abetalipoproteinemia (MTTP): Mutations (2): ♂ Genotyping | c.2593G>T (p.G865X), c.2211delT Sequencing | NM_000253:2-19

Acrodermatitis Enteropathica (SLC39A4): Mutations (7): ♂ Genotyping | c.1223-1227delCCGGG, c.968-971delAGTC, c.318C>A (p.N106K), c.599C>T (p.P200L), c.1120G>A (p.G374R), c.909G>C (p.Q303H), c.989G>A (p.G330D) Sequencing | NM_130849:1-12

Acute Infantile Liver Failure: TRMU Related (TRMU): Mutations (5): ♂ Genotyping | c.229T>C (p.Y77H), c.815G>A (p.G272D), c.2T>A (p.M1K), c.835G>A (p.V279M), c.1102-3C>G Sequencing | NM_018006:1-11

Acyl-CoA Oxidase I Deficiency (ACOX1): Mutations (5): ♂ Genotyping | c.372delCATGCCCGCCTGGAACTT, c.832A>G (p.M278V), c.926A>G (p.Q309R), c.442C>T (p.R148X), c.532G>T (p.G178C) Sequencing | NM_004035:1-14

Adenosine Deaminase Deficiency (ADA): Mutations (22): ♂ Genotyping | c.986C>T (p.A329V), c.872C>T (p.S291L), c.646G>A (p.G216R), c.632G>A (p.R211H), c.631C>T (p.R211C), c.596A>C (p.Q199P), c.536C>A (p.A179D), c.529G>A (p.V177M), c.467G>A (p.R156H), c.466C>T (p.R156C), c.454C>A (p.L152M), c.445C>T (p.R149W), c.419G>A (p.G140E), c.385G>A (p.V129M), c.320T>C (p.L107P), c.302G>A (p.R101Q), c.302G>T (p.R101L), c.301C>T (p.R101W), c.248C>A (p.A83D), c.220G>T (p.G74C), c.58G>A (p.G20R), c.43C>G (p.H15D) Sequencing | NM_000022:1-12

Alkaptonuria (HGD): Mutations (14): ♂ Genotyping | c.1111_1112insC, c.16-1G>A (IVS1-1G>A), c.174delA, c.342+1G>A (IVS5+1G>A), c.1102A>G (p.M368V), c.140C>T (p.S47L), c.688C>T (p.P230S), c.481G>A (p.G161R), c.808G>A (p.G270R), c.899T>G (p.V300G), c.990G>T (p.R330S), c.457_458insG, c.360T>G (p.C120W), c.1112A>G (p.H371R) Sequencing | NM_000187:1-14

Alpha Thalassemia (HBA1, HBA2): Mutations (9): ♂ Genotyping | SEA deletion, c.207C>A (p.N69K), c.223G>C (p.D75H), c.2T>C, c.207C>G (p.N69K), c.340_351delCTCCCCGCCGAG (p.L114_ E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G

Alpha-1-Antitrypsin Deficiency (SERPINA1): Mutations (4): ♂ Genotyping | c.226_228delTTC (p.76delF), c.1131A>T (p.L377F), c.187C>T (p.R63C), c.1096G>A (p.E366K) Sequencing | NM_001127701:1-7

Alpha-Mannosidosis (MAN2B1): Mutations (3): ♂ Genotyping | c.2426T>C (p.L809P), c.2248C>T (p.R750W), c.1830+1G>C (p.V549_E610del) Sequencing | NM_000528:1-24

Alport Syndrome: COL4A3 Related (COL4A3): Mutations (3): ♂ Genotyping | c.4420_4423delCTTTT, c.4441C>T (p.R1481X), c.4571C>G (p.S1524X) Sequencing | NM_000091:2-52

Alport Syndrome: COL4A4 Related (COL4A4): Mutations (4): ♂ Genotyping | c.3713C>G (p.S1238X), c.4129C>T (p.R1377X), c.4923C>A (p.C1641X), c.3601G>A (p.G1201S) Sequencing | NM_000092:2-48

Amegakaryocytic Thrombocytopenia (MPL): Mutations (23): ♂ Genotyping | c.79+2T>A (IVS1+2T>A), c.127C>T (p.R43X), c.305G>C (p.R102P), c.823C>A (p.P275T), c.304C>T (p.R102C), c.376delT (F126Lfs), c.268C>T (p.R90X), c.235_236delCT (p.L79fs), c.367C>T (p.R123X), c.460T>C (p.W154R), c.1305G>C (p.W435C), c.770G>T (p.R257L), c.407C>T (p.P136L), c.407C>A (p.P136H), c.1781T>G (p.L594W), c.311T>C (p.F104S), c.556C>T (p.Q186X), c.1473G>A (p.W491X), c.1499delT (p.L500fs), c.769C>T (p.R257C), c.1904C>T (p.P635L), c.213-1G>A (IVS2-1G>A), c.1566-1G>T (IVS10-1G>T) Sequencing | NM_005373:1-12

Andermann Syndrome (SLC12A6): Mutations (5): ♂ Genotyping | c.2436delG (p.T813fsX813), c.901delA, c.2023C>T (p.R675X), c.3031C>T (p.R1011X), c.619C>T (p.R207C) Sequencing | NM_133647:1-25

Antley-Bixler Syndrome (POR): Mutations (4): ♂ Genotyping | c.859G>C (p.A287P), c.1615G>A (p.G539R), c.1475T>A (p.V492E), c.1370G>A (p.R457H) Sequencing | NM_000941:2-16

Argininemia (ARG1): Mutations (13): ♂ Genotyping | c.365G>A (p.W122X), c.871C>T (p.R291X), c.869C>G (p.T290S), c.703G>C (p.G235R), c.32T>C (p.I11T), c.413G>T (p.G138V), c.57+1G>A, c.61C>T (p.R21X), c.263_266delAGAA (p.K88fs), c.77delA (p.E26fs), c.844delC (p.L282fs), c.466-2A>G, c.703G>A (p.G235R) Sequencing | NM_000045:1-8

Argininosuccinate Lyase Deficiency (ASL): Mutations (7): ♂ Genotyping | c.446+1G>A (IVS5+1G>A), c.857A>G (p.Q286R), c.1135C>T (p.R379C), c.1153C>T (p.R385C), c.283C>T (p.R95C), c.532G>A (p.V178M), c.1060C>T (p.Q354X) Sequencing | NM_000048:2-17

Aromatase Deficiency (CYP19A1): Mutations (10): ♂ Genotyping | c.1222delC (p.K409fs), c.296+1G>A (IVS3+1G>A), c.468delC, c.629-3C>A (IVS4-3C>A), c.743+2T>C (IVS6+2T>C), c.1123C>T (p.R375C), c.1303C>T (p.R435C), c.1094G>A (p.R365Q), c.1310G>A (p.C437Y), c.628G>A (p.E210K) Sequencing | NM_000103:2-10

Arthrogryposis, Mental Retardation, & Seizures (SLC35A3): Mutations (2): ♂ Genotyping | c.1012A>G (p.S338G), c.514C>T (p.Q172X) Sequencing | NM_001271685:1-8

Asparagine Synthetase Deficiency (ASNS): Mutations (1): ♂ Genotyping | c.1084T>G (p.F362V) Sequencing | NM_001673:3-13

Aspartylglycosaminuria (AGA): Mutations (7): ♂ Genotyping | c.200_201delAG, c.488G>C (p.C163S), c.214T>C (p.S72P), c.916T>C (p.C306R), c.904G>A (p.G302R), c.302C>T (p.A101V), c.179G>A (p.G60D) Sequencing | NM_000027:1-9

Ataxia with Vitamin E Deficiency (TTPA): Mutations (14): ♂ Genotyping | c.744delA, c.575G>A (p.R192H), c.400C>T (p.R134X), c.303T>G (p.H101Q), c.358G>A (p.A120T), c.513_514insTT (p.T172fs), c.219_220insAT, c.175C>T (p.R59W), c.421G>A (p.E141K), c.661C>T (p.R221W), c.486delT (p.W163Gfs), c.736G>C (p.G246R), c.205-1G>C, c.306A>G (p.G102G) Sequencing | NM_000370:2-5

Ataxia-Telangiectasia (ATM): Mutations (20): ♂ Genotyping | c.103C>T (p.R35X), c.1564_1565delGA (p.E522fs), c.3245delATCinsTGAT (p.H1082fs), c.3576G>A (p.K1192K), c.3894insT, c.5712_5713insA (p.S1905fs), c.5762+1126A>G, c.5908C>T (p.Q1970X), c.5932G>T (p.E1978X), c.7268A>G (p.E2423G), c.7271T>G (p.V2424G), c.7327C>T (p.R2443X), c.7517_7520delGAGA (p.R2506fs), c.7630-2A>C, c.7638_7646delTAGAATTTC (p.R2547_S2549delRIS), c.7876G>C (p.A2626P), c.7967T>C (p.L2656P), c.8030A>G (p.Y2677C), c.8480T>G (p.F2827C), c.7449G>A (p.W2483X) Sequencing | NM_000051:2-63

Autosomal Recessive Polycystic Kidney Disease (PKHD1): Mutations (40): ♂ Genotyping | c.5895insA (p.L1966fsX1969), c.9689delA (p.D3230fs), c.107C>T (p.T36M), c.1486C>T (p.R496X), c.10412T>G (p.V3471G), c.10658T>C (p.I3553T), c.10174C>T (p.Q3392X), c.9530T>C (p.I3177T), c.9053C>T (p.S3018F), c.8870T>C (p.I2957T), c.8011C>T (p.R2671X), c.6992T>A (p.I2331K), c.5221G>A (p.V1741M), c.4991C>T (p.S1664F), c.3761_3762delCCinsG (p.A1254fs), c.2414C>T (p.P805L), c.664A>G (p.I222V), c.10036T>C (p.C3346R), c.383delC, c.4220T>G (p.L1407R), c.11612G>A (p.W3871X), c.5984A>G (p.E1995G), c.10637delT (p.V3546fs), c.3747T>G (p.C1249W), c.5750A>G (p.Q1917R), c.10865G>A (p.C3622Y), c.50C>T (p.A17V), c.8063G>T (p.C2688F), c.10402A>G (p.I3468V), c.1529delG (p.G510fs), c.657C>T (p.G219G), c.5513A>G (p.Y1838C), c.10856delA (p.K3619fs), c.5381-9T>G (IVS33-9T>G), c.3229-2A>C (IVS28-2A>C), c.10505A>T (p.E3502V), c.2269A>C (p.I757L), c.4165C>A (p.P1389T), c.10364delC (p.S3455fs), c.7350+653A>G (IVS46+653A>G) Sequencing | NM_138694:2-67

Bardet-Biedl Syndrome: BBS1 Related (BBS1): Mutations (3): ♂ Genotyping | c.851delA, c.1645G>T (p.E549X), c.1169T>G (p.M390R) Sequencing | NM_024649:1-17

Bardet-Biedl Syndrome: BBS10 Related (BBS10): Mutations (3): ♂ Genotyping |

c.271_273ins1bp (p.C91fsX95), c.101G>C (p.R34P), c.931T>G (p.S311A) Sequencing | NM_024685:1-2

Bardet-Biedl Syndrome: BBS11 Related (TRIM32): Mutations (1): ♂ Genotyping | c.388C>T (p.P130S) Sequencing | NM_001099679:2

Bardet-Biedl Syndrome: BBS12 Related (BBS12): Mutations (5): ♂ Genotyping | c.335_337delTAG, c.865G>C (p.A289P), c.1063C>T (p.R355X), c.1114_1115delTT (p.F372X), c.1483_1484delGA (p.E495fsX498) Sequencing | NM_152618:1-2

Bardet-Biedl Syndrome: BBS2 Related (BBS2): Mutations (8): ♂ Genotyping | c.940delA, c.72C>G (p.Y24X), c.224T>G (p.V75G), c.311A>C (p.D104A), c.1895G>C (p.R632P), c.823C>T (p.R275X), c.814C>T (p.R272X), c.1206_1207insA (p.R403fs) Sequencing | NM_031885:1-17

Bare Lymphocyte Syndrome: Type II (CIITA): Mutations (3): ♂ Genotyping | c.1141G>T (p.E381X), c.3317+1G>A (IVS18+1G>A), c.2888+1G>A (IVS13+1G>A) Sequencing | NM_000246:1-19

Bartter Syndrome: Type 4A (BSND): Mutations (6): ♂ Genotyping | c.1A>T, c.22C>T (p.R8W), c.139G>A (p.G47R), c.23G>T (p.R8L), c.28G>A (p.G10S), c.3G>A (p.M1I) Sequencing | NM_057176:1-4

Beta Thalassemia (HBB): Mutations (81): ♂ Genotyping | c.124_127delTTCT (p.F42Lfs), c.17_18delCT, c.20delA (p.E7Gfs), c.217insA (p.S73Kfs), c.223+702_444+342del620insAAGTAGA, c.230delC, c.25_26delAA, c.315+1G>A, c.315+2T>C, c.316-197C>T, c.316-146T>G, c.315+745C>G, c.316-1G>A, c.316-1G>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, c.4delG (p.V2Cfs), c.51delC (p.K18Rfs), c.93-21G>A, c.92+1G>A, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>T, c.-50A>C, c.-78a>g, c.-79A>G, c.-81A>G, c.52A>T (p.K18X), c.-137c>g, c.-138c>t, c.-151C>T, c.118C>T (p.Q40X), c.169G>C (p.G57R), c.295G>A (p.V99M), c.415G>C (p.A139P), c.47G>A (p.W16X), c.48G>A (p.W16X), c.-80t>a, c.2T>C, c.75T>A (p.G25G), c.444+111A>G, c.-29G>A, c.68_74delAAGTTGG, c.92G>C (p.R31T), c.92+1G>T, c.93-15T>G, c.93-1G>C, c.112delT, c.113G>A (p.W38X), c.114G>A (p.W38X), c.126delC, c.444+113A>G, c.250delG, c.225delC, c.383_385delAGG (p.Q128_A129delQAinsP), c.321_322insG (p.N109fs), c.316-1G>T, c.316-2A>C, c.287_288insA (p.L97fs), c.271G>T (p.E91X), c.203_204delTG (p.V68Afs), c.154delC (p.P52fs), c.135delC (p.F46fs), c.92+2T>A, c.92+2T>C, c.90C>T (p.G30G), c.84_85insC (p.L29fs), c.59A>G (p.N20S), c.46delT (p.W16Gfs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.2T>G (p.M1R), c.1A>G (p.M1V), c.-137c>t, c.-136C>G, c.-142C>T, c.-140c>t Sequencing | NM_000518:1-3

Beta-Hexosaminidase Pseudodeficiency (HEXA): Mutations (2): ♂ Genotyping | c.739C>T (p.R247W), c.745C>T (p.R249W) Sequencing | NM_000520:1-14

Beta-Ketothiolase Deficiency (ACAT1): Mutations (19): ♂ Genotyping | c.1006-1G>C, c.1006-2A>C, c.1083insA, c.826+1G>T, c.278A>G (p.N93S), c.433C>G (p.Q145E), c.814C>T (p.Q272X), c.1136G>T (p.G379V), c.1138G>A (p.A380T), c.547G>A (p.G183R), c.997G>C (p.A333P), c.2T>A (p.M1K), c.935T>C (p.I312T), c.99T>A (p.Y33X), c.149delC (p.T50Nfs), c.253_255delGAA (p.85delE), c.455G>C (p.G152A), c.380C>T (p.A127V), c.371A>G (p.K124R) Sequencing | NM_000019:1-12

Biotinidase Deficiency (BTD): Mutations (21): ♂ Genotyping | c.98_104delGCGGCTGinsTCC (p.C33FfsX68), c.1368A>C (p.Q456H), c.755A>G (p.D252G), c.1612C>T (p.R538C), c.235C>T (p.R79C), c.100G>A (p.G34S), c.1330G>C (p.D444H), c.511G>A (p.A171T), c.1207T>G (p.F403V), c.470G>A (p.R157H), c.1595C>T (p.T532M), c.1489C>T (p.P497S), c.341G>T (p.G114V), c.1052delC (p.T351fs), c.393delC (p.F131Lfs), c.1049delC (p.A350fs), c.1239delC (p.Y414Ifs), c.1240_1251delTATCTCCACGTC (p.Y414_V417del), c.278A>G (p.Y93C), c.595G>A (p.V199M), c.933delT (p.S311Rfs) Sequencing | NM_000060:1-4

Bloom Syndrome (BLM): Mutations (25): ♂ Genotyping | c.2207_2212delATCTGAinsTAGATTC (p.Y736Lfs), c.2407insT, c.557_559delCAA (p.S186X), c.1284G>A (p.W428X), c.1701G>A (p.W567X), c.1933C>T (p.Q645X), c.2528C>T (p.T843I), c.2695C>T (p.R899X), c.3107G>T (p.C1036F), c.2923delC (p.Q975K), c.3558+1G>T, c.3875-2A>G, c.2074+2T>A, c.2343_2344dupGA (p.781EfsX), c.318_319insT (p.L107fs), c.380delC (p.127Tfs), c.3564delC (p.1188Dfs), c.4008delG (p.1336Rfs), c.947C>G (p.S316X), c.2193+1_2193+9del9, c.1642C>T (p.Q548X), c.3143delA (p.1048NfsX), c.356_357delTA (p.C120Hfs), c.4076+1delG, c.3281C>A (p.S1094X) Sequencing | NM_000057:2-22

Canavan Disease (ASPA): Mutations (8): ♂ Genotyping | c.433-2A>G, c.854A>C (p.E285A), c.693C>A (p.Y231X), c.914C>A (p.A305E), c.71A>G (p.E24G), c.654C>A (p.C218X), c.2T>C (p.M1T), c.79G>A (p.G27R) Sequencing | NM_000049:1-6

Carnitine Palmitoyltransferase IA Deficiency (CPT1A): Mutations (10): ♂ Genotyping | c.1079A>G (p.E360G), c.1361A>G (p.D454G), c.1241C>T (p.A414V), c.1436C>T (p.P479L), c.2126G>A (p.G709E), c.2129G>A (p.G710E), c.1493A>G (p.Y498C), c.1339C>T (p.R447X), c.2156G>A (p.G719D), c.96T>G (p.Y32X) Sequencing | NM_001876:2-19

Carnitine Palmitoyltransferase II Deficiency (CPT2): Mutations (19): ♂ Genotyping | c.109_110insGC, c.1238_1239delAG, c.1737delC, c.1923_1935delGAAGGCCTTAGAA, c.534_558delGAACCCTGCAAAAAGTGACACTATCinsT, c.1649A>G (p.Q550R), c.1883A>C (p.Y628S), c.359A>G (p.Y120C), c.983A>G (p.D328G), c.149C>A (p.P50H), c.1810C>T (p.P604S), c.1891C>T (p.R631C), c.338C>T (p.S113L), c.370C>T (p.R124X), c.680C>T (p.P227L), c.1646G>A (p.G549D), c.452G>A (p.R151Q), c.520G>A (p.E174K), c.1148T>A (p.F383Y) Sequencing | NM_000098:1-5

Carnitine-Acylcarnitine Translocase Deficiency (SLC25A20): Mutations (7): ♂ Genotyping | c.199-10T>G (IVS2-10T>G), c.897_898insC (p.N300fs), c.496C>T (p.R166X), c.84delT (p.H29Tfs), c.713A>G (p.Q238R), c.576G>A (p.W192X), c.106-2A>T Sequencing | NM_000387:1-9

Carpenter Syndrome (RAB23): Mutations (2): ♂ Genotyping | c.434T>A (p.L145X), c.408_409insT (p.136fsX) Sequencing | NM_016277:2-7

Cartilage-Hair Hypoplasia (RMRP): Mutations (2): ♂ Genotyping | n.71A>G, c.263G>T Sequencing | NR_003051:1

Cerebrotendinous Xanthomatosis (CYP27A1): Mutations (14): ♂ Genotyping | c.1263+1G>A, c.844+1G>A, c.1016C>T (p.T339M), c.1183C>T (p.R395C), c.1420C>T (p.R474W), c.1435C>T (p.R479C), c.379C>T (p.R127W), c.819delT (p.D273fs), c.1214G>A (p.R405Q), c.1421G>A (p.R474Q), c.434G>A (p.G145E), c.583G>T (p.E195X), c.646G>C (p.A216P), c.1183C>A (p.R395S) Sequencing | NM_000784:1-9

Chediak-Higashi Syndrome (LYST): Mutations (4): ♂ Genotyping | c.3085C>T (p.Q1029X), c.9590delA (p.Y3197fs), c.1902_1903insA (p.A635Sfs), c.118_119insG (p.A40fs) Sequencing | NM_000081:3-53

Cholesteryl Ester Storage Disease (LIPA): Mutations (4): ♂ Genotyping | c.1024G>A (p.G342R), c.894G>A (p.Q298X), c.883C>T (p.H295Y), c.652C>T (p.R218X) Sequencing | NM_001127605:2-10

Choreoacanthocytosis (VPS13A): Mutations (1): ♂ Genotyping | c.6058delC (p.P2020fs) Sequencing | NM_033305:1-72

Chronic Granulomatous Disease: CYBA Related (CYBA): Mutations (12): ♂ Genotyping | c.354C>A (p.S118R), c.467C>A (p.P156Q), c.281A>G (p.H94R), c.7C>T (p.Q3X), c.70G>A (p.G24R), c.244delC (p.P82fs), c.171_172insG (p.K58fs), c.373G>A (p.A125T), c.174delG (p.K58fs), c.385_388delGAGC (p.E129SfsX61), c.369+1G>A (IVS5+1G>A), c.71G>A (p.G24E) Sequencing | NM_000101:1-5

Citrin Deficiency (SLC25A13): Mutations (8): ♂ Genotyping | c.1180G>A (p.G394S), c.674C>A (p.S225X), c.1766G>A (p.R589Q), c.851_854delGTAT (p.R284fs), c.1802_1803insA (p.Y601fs), c.1180+1G>A, c.1663_1664insGAGATTACAGGTGGCTGCCCGGG (p.A555fs), c.1314+1G>A Sequencing | NM_001160210:1-18

Citrullinemia: Type I (ASS1): Mutations (11): ♂ Genotyping | c.1194-1G>C, c.970+5G>A, c.928A>C (p.K310Q), c.835C>T (p.R279X), c.1085G>T (p.G362V), c.470G>A (p.R157H), c.539G>A (p.S180N), c.970G>A (p.G324S), c.535T>C (p.W179R), c.1168G>A (p.G390R), c.421-2A>G (IVS6-2A>G) Sequencing | NM_000050:3-16

Classical Galactosemia (GALT): Mutations (18): ♂ Genotyping | c.253-2A>G, c.563A>G (p.Q188R), c.626A>G (p.Y209C), c.404C>T (p.S135L), c.413C>T (p.T138M), c.505C>A (p.Q169K), c.997C>G (p.R333G), c.607G>A (p.E203K), c.855G>T (p.K285N), c.1138T>C (p.X380R), c.221T>C (p.L74P), c.425T>A (p.M142K), c.512T>C (p.F171S), c.584T>C (p.L195P), c.134_138delCAGCT, c.-1039_753del3162, c.820+51_*789del2294ins12, c.404C>G (p.S135W) Sequencing | NM_000155:1-11

Cockayne Syndrome: Type A (ERCC8): Mutations (3): ♂ Genotyping | c.966C>A (p.Y322X), c.37G>T (p.E13X), c.479C>T (p.A160V) Sequencing | NM_000082:1-12

Cockayne Syndrome: Type B (ERCC6): Mutations (7): ♂ Genotyping | c.1550G>A (p.W517X), c.2203C>T (p.R735X), c.1518delG (p.K506Nfs), c.1357C>T (p.R453X), c.972_973insA (p.E325Rfs), c.1974_1975insTGTC (p.T659fs), c.1034_1035insT (p.K345fs) Sequencing | NM_000124:2-21

Cohen Syndrome (VPS13B): Mutations (9): ♂ Genotyping | c.6578T>G (p.L2193R), c.7051C>T (p.R2351X), c.4471G>T (p.E1491X), c.2911C>T (p.R971X), c.7934G>A (p.G2645D), c.10888C>T (p.Q3630X), c.8459T>C (p.I2820T), c.9259_9260insT (p.L3087fs), c.3348_3349delCT (p.C1117fx) Sequencing | NM_017890:2-51,53-62

Combined Pituitary Hormone Deficiency: PROP1 Related (PROP1): Mutations (11): ♂ Genotyping | c.218G>A (p.R73H), c.150delA (p.G50fsX), c.358C>T (p.R120C), c.112_124delTCGAGTGCTCCAC (p.S38fsX), c.2T>C, c.157delA (p.R53fsX), c.212G>A (p.R71H), c.217C>T (p.R73C), c.582G>A (p.W194X), c.109+1G>T, c.301delAG (p.S101fsX) Sequencing | NM_006261:1-3

Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2): Mutations (5): ♂ Genotyping | c.357C>A (p.F119L), c.422G>A (p.R141H), c.338C>T (p.P113L), c.691G>A (p.V231M), c.470T>C (p.F157S) Sequencing | NM_000303:1-8

Congenital Disorder of Glycosylation: Type 1B: MPI Related (MPI): Mutations (1): ♂ Genotyping | c.884G>A (p.R295H) Sequencing | NM_002435:1-8

Congenital Disorder of Glycosylation: Type 1C: ALG6 Related (ALG6): Mutations (4): ♂ Genotyping | c.257+5G>A, c.895_897delATA, c.998C>T (p.A333V), c.1432T>C (p.S478P) Sequencing | NM_013339:2-15

Congenital Ichthyosis: ABCA12 Related (ABCA12): Mutations (8): ♂ Genotyping | c.4139A>G (p.N1380S), c.4951G>A (p.G1651S), c.4142G>A (p.G1381E), c.4541G>A (p.R1514H), c.4615G>A (p.E1539K), c.7323delC (p.V2442Sfs), c.6610C>T (p.R2204X), c.3535G>A (p.G1179R) Sequencing | NM_173076:1-53

Congenital Insensitivity to Pain with Anhidrosis (NTRK1): Mutations (12): ♂ Genotyping | c.1729G>C (p.G577R), c.2339G>C (p.R780P), c.25C>T (p.Q9X), c.1076A>G (p.Y359C), c.1759A>G (p.M587V), c.207_208delTG (p.E70Afs), c.1550G>A (p.G517E), c.717+4A>T, c.429-1G>C, c.1660delC (p.R554fs), c.2046+3A>C, c.2084C>T (p.P695L) Sequencing | NM_002529:2-17

Congenital Lipoid Adrenal Hyperplasia (STAR): Mutations (12): ♂ Genotyping | c.178+1_178+2insT (IVS2+3insT), c.201_202delCT, c.466-11T>A (IVS4-11T>A), c.64+1G>T (IVS1+1G>T), c.562C>T (p.R188C), c.772C>T (p.Q258X), c.545G>A (p.R182H), c.545G>T

(p.R182L), c.559G>A (p.V187M), c.650G>C (p.R217T), c.749G>A (p.W250X), c.64+1G>A Sequencing | NM_000349:1-7

Congenital Myasthenic Syndrome: CHRNE Related (CHRNE): Mutations (12): ♂ Genotyping | c.1327delG (p.E443fs), c.865C>T (p.L289F), c.911delT (p.L304fs), c.344+1G>A, c.850A>C (p.T284P), c.422C>T (p.P141L), c.250C>G (p.R84G), c.500G>T (p.R167L), c.991C>T (p.R331W), c.37G>A (p.G13R), c.613_619delTGGGCCA (p.W205fs), c.1353_1354insG (p.N452Efs) Sequencing | NM_000080:1-12

Congenital Myasthenic Syndrome: DOK7 Related (DOK7): Mutations (6): ♂ Genotyping | c.601C>T (p.R201X), c.539G>C (p.G180A), c.548_551delTCCT (p.F183fs), c.1263_1264insC (p.S422fs), c.101-1G>T, c.331+1G>T Sequencing | NM_173660:3-7

Congenital Myasthenic Syndrome: RAPSN Related (RAPSN): Mutations (11): ♂ Genotyping | c.264C>A (p.N88K), c.41T>C (p.L14P), c.807C>A (p.Y269X), c.548_549insGTTCT (p.L183fs), c.46_47insC (p.L16fs), c.133G>A (p.V45M), c.848T>C (p.L283P), c.484G>A (p.E162K), c.490C>T (p.R164C), c.-210A>G, c.193-15C>A (IVS1-15C>A) Sequencing | NM_005055:1-8

Congenital Neutropenia: Recessive (HAX1): Mutations (6): ♂ Genotyping | c.121_125insG, c.130_131insA, c.431insG, c.91delG, c.256C>T (p.R86X), c.568C>T (p.Q190X) Sequencing | NM_006118:1-7

Corneal Dystrophy and Perceptive Deafness (SLC4A11): Mutations (8): ♂ Genotyping | c.1459_1462delTACGinsA (p.487_488delYAinsT), c.2313_2314insTATGACAC, c.554_561delGCTTCGCC (p.R185fs), c.2566A>G (p.M856V), c.1463G>A (p.R488K), c.2528T>C (p.L843P), c.637T>C (p.S213P), c.2321+1G>A Sequencing | NM_001174090:1-20

Corticosterone Methyloxidase Deficiency (CYP11B2): Mutations (3): ♂ Genotyping | c.1492A>G (p.T498A), c.541C>T (p.R181W), c.1382T>C (p.L461P) Sequencing | NM_000498:1-9

Crigler-Najjar Syndrome (UGT1A1): Mutations (11): ♂ Genotyping | c.508_513delTTC (p.170delF), c.1070A>G (p.Q357R), c.1021C>T (p.R341X), c.1124C>T (p.S375F), c.840C>A (p.C280X), c.991C>T (p.Q331X), c.923G>A (p.G308E), c.1198A>G (p.N400D), c.992A>G (p.Q331R), c.44T>G (p.L15R), c.524T>A (p.L175Q) Sequencing | NM_000463:1-5

Cystic Fibrosis (CFTR): Mutations (149): ♂ Genotyping | c.1029delC, c.1153_1154insAT, c.1477delCA, c.1519_1521delATC (p.507delI), c.1521_1523delCTT (p.508delF), c.1545_1546delTA (p.Y515Xfs), c.1585-1G>A, c.164+12T>C, c.1680-886A>G, c.1680-1G>A, c.1766+1G>A, c.1766+1G>T, c.1766+5G>T, c.1818del84, c.1911delG, c.1923delCTCAAAACTinsA, c.1973delGAAATTCAATCCTinsAGAAA, c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2051_2052delAAinsG (p.K684SfsX38), c.2174insA, c.261delTT, c.2657+5G>A, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2988+1G>A, c.3039delC, c.3140-26A>G, c.325delTATinsG, c.3527delC, c.3535delACCA, c.3691delT, c.3717+12191C>T, c.3744delA, c.3773_3774insT (p.L1258fs), c.442delA, c.489+1G>T, c.531delT, c.579+1G>T, c.579+5G>A (IVS4+5G>A), c.803delA (p.N268fs), c.805_806delAT (p.I269fs), c.933_935delCTT (p.311delF), c.946delT, c.1645A>C (p.S549R), c.2128A>T (p.K710X), c.1000C>T (p.R334W), c.1013C>T (p.T338I), c.1364C>A (p.A455E), c.1477C>T (p.Q493X), c.1572C>A (p.C524X), c.1654C>T (p.Q552X), c.1657C>T (p.R553X), c.1721C>A (p.P574H), c.2125C>T (p.R709X), c.223C>T (p.R75X), c.2668C>T (p.Q890X), c.3196C>T (p.R1066C), c.3276C>G (p.Y1092X), c.3472C>T (p.R1158X), c.3484C>T (p.R1162X), c.349C>T (p.R117C), c.3587C>G (p.S1196X), c.3712C>T (p.Q1238X), c.3764C>A (p.S1255X), c.3909C>G (p.N1303K), c.1040G>A (p.R347H), c.1040G>C (p.R347P), c.1438G>T (p.G480C), c.1558G>T (p.V520F), c.1624G>T (p.G542X), c.1646G>A (p.S549N), c.1646G>T (p.S549I), c.1652G>A (p.G551D), c.1675G>A (p.A559T), c.1679G>C (p.R560T), c.178G>T (p.E60X), c.254G>A (p.G85E), c.271G>A (p.G91R), c.274G>T (p.E92X), c.3209G>A (p.R1070Q), c.3266G>A (p.W1089X), c.3454G>C (p.D1152H), c.350G>A (p.R117H), c.3611G>A (p.W1204X), c.3752G>A (p.S1251N), c.3846G>A (p.W1282X), c.3848G>T (p.R1283M), c.532G>A (p.G178R), c.988G>T (p.G330X), c.1090T>C (p.S364P), c.3302T>A (p.M1101K), c.617T>G (p.L206W), c.14C>T (p.P5L), c.19G>T (p.E7X), c.171G>A (p.W57X), c.313delA (p.I105fs), c.328G>C (p.D110H), c.580-1G>T, c.1055G>A (p.R352Q), c.1075C>A (p.Q359K), c.1079C>A (p.T360K), c.1647T>G (p.S549R), c.1976delA (p.N659fs), c.2290C>T (p.R764X), c.2737_2738insG (p.Y913X), c.3067_3072delATAGTG (p.I1023_V1024delT), c.3536_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.54-5940_273+10250del21080bp (p.S18fs), c.4364C>G (p.S1455X), c.4003C>T (p.L1335F), c.2538G>A (p.W846X), c.200C>T (p.P67L), c.4426C>T (p.Q1476X), c.1116+1G>A, c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.263T>G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.658C>T (p.Q220X), c.868C>T (p.Q290X), c.1526delG (p.G509fs), c.2908+1085_3367+260del7201, c.11C>A (p.S4X), c.3878_3881delTATT (p.V1293fs), c.3700A>G (p.I1234V), c.416A>T (p.H139L), c.366T>A (p.Y122X), c.3767_3768insC (p.A1256fs), c.613C>T (p.P205S), c.293A>G (p.Q98R), c.3731G>A (p.G1244E), c.535C>A (p.Q179K), c.3368-2A>G, c.455T>G (p.M152R), c.1610_1611delAC (p.D537fs), c.3254A>G (p.H1085R), c.496A>G (p.K166E), c.1408_1417delGTGATTATGG (p.V470fs), c.1585-8G>A, c.2909G>A (p.G970D), c.653T>A (p.L218X), c.1175T>G (p.V392G), c.3139_3139+1delGG, c.3717+4A>G (IVS22+4A>G) Sequencing | NM_000492:1-27

Cystinosis (CTNS): Mutations (14): ♂ Genotyping | c.18_21delGACT, c.198_218delTATTACTATCCTTGAGCTCCC , c.283G>T (p.G95X), c.414G>A (p.W138X), c.506G>A (p.G169D), c.613G>A (p.D205N), c.473T>C (p.L158P), c.329G>T (p.G110V), c.416C>T (p.S139F), c.589G>A (p.G197R), c.969C>G (p.N323K), c.1015G>A (p.G339R), c.-39155_848del57119, c.199_219delATTACTATCCTTGAGCTCCCC (p.I67_P73del) Sequencing | NM_001031681:1,3-13

Cystinuria: Non-Type I (SLC7A9): Mutations (15): ♂ Genotyping | c.508G>A (p.V170M), c.313G>A (p.G105R), c.583G>A (p.G195R), c.775G>A (p.G259R), c.997C>T (p.R333W), c.131T>C (p.I44T), c.782C>T (p.P261L), c.695A>G (p.Y232C), c.544G>A (p.A182T), c.368C>T (p.T123M), c.614_615insA (p.K205fs), c.604+2T>C, c.605-3C>A (IVS5-3C>A), c.1445C>T (p.P482L), c.368_369delCG (p.T123fs) Sequencing | NM_001243036:2-13

Cystinuria: Type I (SLC3A1): Mutations (10): ♂ Genotyping | c.1400T>C (p.M467T), c.2033T>C (p.L678P), c.542G>A (p.R181Q), c.1955C>G (p.T652R), c.1843C>A (p.P615T), c.1085G>A (p.R362H), c.1597T>A (p.Y533N), c.647C>T (p.T216M), c.808C>T (p.R270X), c.452A>G (p.Y151C) Sequencing | NM_000341:1-10

D-Bifunctional Protein Deficiency (HSD17B4): Mutations (6): ♂ Genotyping | c.46G>A (p.G16S), c.63G>T (p.L21F), c.422_423delAG, c.652G>T (p.V218L), c.1369A>T (p.N457Y), c.1369A>G (p.N457D) Sequencing | NM_000414:1-24

Diabetes: Recessive Permanent Neonatal (ABCC8): Mutations (2): ♂ Genotyping | c.215A>G (p.N72S), c.1144G>A (p.E382K) Sequencing | NM_000352:1-39

Du Pan Syndrome (GDF5): Mutations (4): ♂ Genotyping | c.1309delTTG, c.1306C>A (p.P436T), c.1133G>A (p.R378Q), c.1322T>C (p.L441P) Sequencing | NM_000557:1-2

Dyskeratosis Congenita: RTEL1 Related (RTEL1): Mutations (5): ♂ Genotyping | c.2869C>T (p.R981W), c.2920C>T (p.R974X), c.1548G>T (p.M516I), c.2216G>T (p.G763V), c.3791G>A (p.R1264H) Sequencing | NM_001283009:2-35

Dystrophic Epidermolysis Bullosa: Recessive (COL7A1): Mutations (11): ♂ Genotyping | c.2470_2471insG, c.5820G>A (p.P1940P), c.933C>A (p.Y311X), c.4039G>C (p.G1347R), c.8393T>A (p.M2798K), c.425A>G (p.K142R), C.8441-14_8435delGCTCTTGGCTCCAGGACCCCT, c.4783-1G>A, c.7344G>A (p.V2448X), c.4991G>C (p.G1664A), c.497_498insA (p.V168GfsX179) Sequencing | NM_000094:1-118

Ehlers-Danlos Syndrome: Type VIIC (ADAMTS2): Mutations (2): ♂ Genotyping | c.673C>T (p.Q225X), c.2384G>A (p.W795X) Sequencing | NM_014244:2-22

Ellis-van Creveld Syndrome: EVC Related (EVC): Mutations (10): ♂ Genotyping | c.919T>C (p.S307P), c.1694delC (p.A565VfsX23), c.734delT (p.L245fs), c.910-911insA (p.R304fs), c.2635C>T (p.Q879X), c.1868T>C (p.L623Q), c.1858_1879delTTGGGCCGACTGGGCGGCCTC (p.L620_L626del), c.1886+5G>T, c.1098+1G>A, c.1018C>T (p.R340X) Sequencing | NM_153717:2-21

Ellis-van Creveld Syndrome: EVC2 Related (EVC2): Mutations (1): ♂ Genotyping | c.3025C>T (p.Q1009X) Sequencing | NM_147127:1-22

Enhanced S-Cone (NR2E3): Mutations (5): ♂ Genotyping | c.932G>A (p.R311Q), c.227G>A (p.R76Q), c.119-2A>C, c.226C>T (p.R76W), c.747+1G>C (IVS5+1G>C) Sequencing | NM_016346:1-8

Ethylmalonic Aciduria (ETHE1): Mutations (4): ♂ Genotyping | c.505+1G>T, c.487C>T (p.R163W), c.3G>T (p.M1I), c.488G>A (p.R163Q) Sequencing | NM_014297:1-7

Familial Chloride Diarrhea (SLC26A3): Mutations (6): ♂ Genotyping | c.344delT (p.I115fs), c.559G>T (p.G187X), c.951delGGT (p.V318del), c.1386G>A (p.W462X), c.371A>T (p.H124L), c.2023_2025dupATC (p.I675L) Sequencing | NM_000111:2-21

Familial Dysautonomia (IKBKAP): Mutations (4): ♂ Genotyping | c.2204+6T>C, c.2741C>T (p.P914L), c.2087G>C (p.R696P), c.2128C>T (p.Q710X) Sequencing | NM_003640:2-37

Familial Hyperinsulinism: Type 1: ABCC8 Related (ABCC8): Mutations (11): ♂ Genotyping | c.3989-9G>A, c.4159_4161delTTC (p.1387delF), c.4258C>T (p.R1420C), c.4477C>T (p.R1493W), c.2147G>T (p.G716V), c.4055G>C (p.R1352P), c.560T>A (p.V187D), c.4516G>A (p.E1506K), c.2506C>T (p.Q836X), c.579+2T>A, c.1333-1013A>G (IVS8-1013A>G) Sequencing | NM_000352:1-39

Familial Hyperinsulinism: Type 2: KCNJ11 Related (KCNJ11): Mutations (6): ♂ Genotyping | c.776A>G (p.H259R), c.36C>A (p.Y12X), C.C761T (p.P254L), c.G-134T, c.844G>A (p.E282K), c.440T>C (p.L147P) Sequencing | NM_000525:1

Familial Mediterranean Fever (MEFV): Mutations (12): ♂ Genotyping | c.2076_2078delAAT (p.692delI), c.2080A>G (p.M694V), c.2084A>G (p.K695R), c.1437C>G (p.F479L), c.800C>T (p.T267I), c.1958G>A (p.R653H), c.2040G>A (p.M680I), c.2040G>C (p.M680I), c.2082G>A (p.M694I), c.2230G>T (p.A744S), c.2282G>A (p.R761H), c.2177T>C (p.V726A) Sequencing | NM_000243:1-10

Fanconi Anemia: Type A (FANCA): Mutations (10): ♂ Genotyping | c.295C>T (p.Q99X), c.1115_1118delTTGG, c.3720_3724delAAACA (p.E1240Dfs), c.513G>A (p.W171X), c.1606delT (p.S536fs), c.3558_3559insG (p.R1187Efs), c.1615delG (p.D539fs), c.890_893delGCTG (p.C297fs), c.2172_2173insG (p.T724fs), c.4275delT (p.R1425fs) Sequencing | NM_000135:1-43

Fanconi Anemia: Type C (FANCC): Mutations (8): ♂ Genotyping | c.456+4A>T, c.67delG, c.37C>T (p.Q13X), c.553C>T (p.R185X), c.1661T>C (p.L554P), c.1642C>T (p.R548X), c.66G>A (p.W22X), c.65G>A (p.W22X) Sequencing | NM_000136:2-15

Fanconi Anemia: Type G (FANCG): Mutations (5): ♂ Genotyping | c.1480+1G>C, c.307+1G>C, c.1794_1803delCTGGATCCGT (p.W599Pfs), c.637_643delTACCGCC (p.Y213K+4X), c.925-2A>G Sequencing | NM_004629:1-14

Fanconi Anemia: Type J (BRIP1): Mutations (1): ♂ Genotyping | c.2392C>T (p.R798X) Sequencing | NM_032043:2-20

Fumarase Deficiency (FH): Mutations (1): ♂ Genotyping | c.1431_1433insAAA Sequencing | NM_000143:1-10

GM1-Gangliosidoses (GLB1): Mutations (17): ♂ Genotyping | c.1480-2A>G,

c.75+2_75+3insT, c.1772A>G (p.Y591C), c.947A>G (p.Y316C), c.1051C>T (p.R351X), c.1369C>T (p.R457X), c.145C>T (p.R49C), c.202C>T (p.R68W), c.245C>T (p.T82M), c.601C>T (p.R201C), c.622C>T (p.R208C), c.1370G>A (p.R457Q), c.176G>A (p.R59H), c.367G>A (p.G123R), c.152T>C (p.I51T), c.1771T>A (p.Y591N), c.1577_1578insG Sequencing | NM_000404:1-16

GRACILE Syndrome (BCS1L): Mutations (12): ♂ Genotyping | c.232A>G (p.S78G), c.103G>C (p.G35R), c.148A>G (p.T50A), c.166C>T (p.R56X), c.133C>T (p.R45C), c.296C>T (p.P99L), c.464G>C (p.R155P), c.547C>T (p.R183C), c.548G>A (p.R183H), c.550C>T (p.R184C), c.830G>A (p.S277N), c.1057G>A (p.V353M) Sequencing | NM_004328:1-9

Galactokinase Deficiency (GALK1): Mutations (7): ♂ Genotyping | c.1144C>T (p.Q382X), c.1045G>A (p.G349S), c.1031C>T (p.T344M), c.238G>T (p.E80X), c.94G>A (p.V32M), c.82C>A (p.P28T), c.593C>T (p.A198V) Sequencing | NM_000154:1-8

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

Gitelman Syndrome (SLC12A3): Mutations (11): ♂ Genotyping | c.1926-1G>T, c.2883+1G>T, c.1046C>T (p.P348L), c.1763C>T (p.A588V), c.622C>T (p.R208W), c.1889G>T (p.G629V), c.1961G>A (p.R654H), c.1868T>C (p.L623P), c.1180+1G>T (IVS9+1G>T), c.1670-191C>T, c.2548+253C>T Sequencing | NM_000339:1-26

Globoid Cell Leukodystrophy (GALC): Mutations (10): ♂ Genotyping | c.1153G>T (p.E385X), c.857G>A (p.G286D), c.2002A>C (p.T668P), c.1700A>C (p.Y567S), c.1586C>T (p.T529M), c.1472delA (p.K491fs), c.913A>G (p.I305V), c.683_694delATCTCTGGGAGTinsCTC (p.N228_S232del5insTP), c.246A>G (p.I82M), c.1161+6555_*9573del31670bp Sequencing | NM_000153:2-17

Glutaric Acidemia: Type I (GCDH): Mutations (8): ♂ Genotyping | c.1204C>T (p.R402W), c.1262C>T (p.A421V), c.743C>T (p.P248L), c.1093G>A (p.E365K), c.877G>A (p.A293T), c.1083-2A>C (IVS10-2A>C), c.680G>C (p.R227P), c.1198G>A (p.V400M) Sequencing | NM_000159:2-12

Glutaric Acidemia: Type IIA (ETFA): Mutations (5): ♂ Genotyping | c.797C>T (p.T266M), c.470T>G (p.V157G), c.346G>A (p.G116R), c.809_811delTAG (p.V270_A271delinsA), c.963+1delG Sequencing | NM_000126:1-12

Glutaric Acidemia: Type IIB (ETFB): Mutations (2): ♂ Genotyping | c.764G>A (p.R255Q), c.655G>A (p.D219N) Sequencing | NM_001014763:1-5, NM_001985:1

Glutaric Acidemia: Type IIC (ETFDH): Mutations (8): ♂ Genotyping | c.1448C>T (p.P483L), c.2T>C (p.M1T), c.250G>A (p.A84T), c.524G>T (p.R175L), c.380T>A (p.L127H), c.524G>A (p.R175H), c.1130T>C (p.L377P), c.36delA (p.A12fs) Sequencing | NM_004453:1-13

Glycine Encephalopathy: AMT Related (AMT): Mutations (6): ♂ Genotyping | c.959G>A (p.R320H), c.878-1G>A, c.826G>C (p.D276H), c.574C>T (p.Q192X), c.139G>A (p.G47R), c.125A>G (p.H42R) Sequencing | NM_000481:1-9

Glycine Encephalopathy: GLDC Related (GLDC): Mutations (5): ♂ Genotyping | c.2284G>A (p.G762R), c.2266_2268delTTC (p.756delF), c.1691G>T (p.S564I), c.1545G>C (p.R515S), c.2T>C (p.M1T) Sequencing | NM_000170:1-25

Glycogen Storage Disease: Type IA (G6PC): Mutations (13): ♂ Genotyping | c.376_377insTA, c.79delC, c.979_981delTTC (p.327delF), c.1039C>T (p.Q347X), c.247C>T (p.R83C), c.724C>T (p.Q242X), c.248G>A (p.R83H), c.562G>C (p.G188R), c.648G>T, c.809G>T (p.G270V), c.113A>T (p.D38V), c.975delG (p.L326fs), c.724delC Sequencing | NM_000151:1-5

Glycogen Storage Disease: Type IB (SLC37A4): Mutations (5): ♂ Genotyping | c.1042_1043delCT, c.796G>T (p.G266C), c.1016G>A (p.G339D), c.1099G>A (p.A367T), c.352T>C (p.W118R) Sequencing | NM_001164277:3-11

Glycogen Storage Disease: Type II (GAA): Mutations (13): ♂ Genotyping | c.1935C>A (p.D645E), c.2560C>T (p.R854X), c.-32-13T>G (IVS1-13T>G), c.525delT (p.E176Rfs), c.710C>T (p.A237V), c.896T>G (p.L299R), c.953T>C (p.M318T), c.1561G>A (p.E521K), c.1585_1586delTCinsGT (p.S529V), c.1634C>T (p.P545L), c.1927G>A (p.G643R), c.2173C>T (p.R725W), c.2707_2709delK (p.903delK) Sequencing | NM_001079804:2-20

Glycogen Storage Disease: Type III (AGL): Mutations (15): ♂ Genotyping | c.17_18delAG, c.4455delT (p.S1486fs), c.1222C>T (p.R408X), c.16C>T (p.Q6X), c.1384delG (p.V462X), c.2039G>A (p.W680X), c.2590C>T (p.R864X), c.2681+1G>A, c.3439A>G (p.R1147G), c.3682C>T (p.R1228X), c.3965delT (p.V1322AfsX27), c.3980G>A (p.W1327X), c.4260-12A>G (IVS32-12A>G), c.4342G>C (p.G1448R), c.2681+1G>T Sequencing | NM_000642:2-34

Glycogen Storage Disease: Type IV (GBE1): Mutations (3): ♂ Genotyping | c.986A>C (p.Y329S), c.691+2T>C (IVS5+2T>C), c.986A>G (p.Y329C) Sequencing | NM_000158:1-16

Glycogen Storage Disease: Type V (PYGM): Mutations (10): ♂ Genotyping | c.2128_2130delTTC (p.710delF), c.1627A>T (p.K543X), c.1628A>C (p.K543T), c.148C>T (p.R50X), c.255C>A (p.Y85X), c.613G>A (p.G205S), c.2392T>C (p.W798R), c.1827G>A (p.K609K), c.632delG (p.S211fs), c.808C>T (p.R270X) Sequencing | NM_005609:1-20

Glycogen Storage Disease: Type VII (PFKM): Mutations (4): ♂ Genotyping | c.450+1G>A, c.329G>T (p.R110L), c.283C>T (p.R95X), c.2214delC (p.P739Qfs) Sequencing | NM_001166686:2-25

Guanidinoacetate Methyltransferase Deficiency (GAMT): Mutations (4): ♂ Genotyping | c.506G>A (p.C169Y), c.327G>A, c.309_310insCCGGGACTGGGCC (p.L99_A103fs), c.148A>C (p.M50L) Sequencing | NM_000156:1-6

HMG-CoA Lyase Deficiency (HMGCL): Mutations (7): ♂ Genotyping | c.914_915delTT, c.122G>A (p.R41Q), c.208G>C (p.V70L), c.835G>A (p.E279K), c.561+1G>A, c.109G>T (p.E37X), c.561+1G>T Sequencing | NM_000191:1-9

Hemochromatosis: Type 2A: HFE2 Related (HFE2): Mutations (1): ♂ Genotyping | c.959G>T (p.G320V) Sequencing | NM_213653:2-4

Hemochromatosis: Type 3: TFR2 Related (TFR2): Mutations (4): ♂ Genotyping | c.2069A>C (p.Q690P), c.750C>G (p.Y250X), c.515T>A (p.M172K), c.88_89insC (p.E60X) Sequencing | NM_003227:1-18

Hemoglobinopathy: Hb C (HBB): Mutations (1): ♂ Genotyping | c.19G>A (p.E7K) Sequencing | NM_000518:1-3

Hemoglobinopathy: Hb D (HBB): Mutations (1): ♂ Genotyping | c.364G>C (p.E122Q) Sequencing | NM_000518:1-3

Hemoglobinopathy: Hb E (HBB): Mutations (1): ♂ Genotyping | c.79G>A (p.E27K) Sequencing | NM_000518:1-3

Hemoglobinopathy: Hb O (HBB): Mutations (1): ♂ Genotyping | c.364G>A (p.E122K) Sequencing | NM_000518:1-3

Hereditary Fructose Intolerance (ALDOB): Mutations (10): ♂ Genotyping | c.357_360delAAAC, c.1005C>G (p.N335K), c.524C>A (p.A175D), c.448G>C (p.A150P), c.612T>G (p.Y204X), c.865_867delCTT (p.289delL), c.720C>A (p.C240X), c.442T>C (p.W148R), c.178C>T (p.R60X), c.10C>T (p.R4X) Sequencing | NM_000035:2-9

Hereditary Spastic Paraplegia: TECPR2 Related (TECPR2): Mutations (1): ♂ Genotyping | c.3416delT (p.L1139fs) Sequencing | NM_014844:2-20

Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3): Mutations (1): ♂ Genotyping | c.1981C>T (p.R661X) Sequencing | NM_000227:1-38

Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3): Mutations (6): ♂ Genotyping | c.3024delT, c.124C>T (p.R42X), c.1903C>T (p.R635X), c.430C>T (p.R144X), c.727C>T (p.Q243X), c.3247C>T (p.Q1083X) Sequencing | NM_000228:2-23

Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2): Mutations (1): ♂ Genotyping | c.283C>T (p.R95X) Sequencing | NM_005562:1-23

Hermansky-Pudlak Syndrome: Type 1 (HPS1): Mutations (1): ♂ Genotyping | c.1470_1486dup16 (p.H497Qfs) Sequencing | NM_000195:3-20

Hermansky-Pudlak Syndrome: Type 3 (HPS3): Mutations (4): ♂ Genotyping | c.1189C>T (p.R397W), c.1691+2T>G, c.2589+1G>C, c.1163+1G>A Sequencing | NM_032383:1-17

Hermansky-Pudlak Syndrome: Type 4 (HPS4): Mutations (7): ♂ Genotyping | c.1876C>T (p.Q626X), c.526C>T (p.Q176X), c.957_958insGCTTGTCCAGATGGCAGGAAGGAG (p.E319_N320ins8), c.634C>T (p.R212X), c.397G>T (p.E133X), c.649G>T (p.E217X), c.2039delC (p.P680fs) Sequencing | NM_152841:1-12

Holocarboxylase Synthetase Deficiency (HLCS): Mutations (7): ♂ Genotyping | c.1795+5G>A (IVS10+5G>A), c.780delG, c.710T>C (p.L237P), c.1522C>T (p.R508W), c.1648G>A (p.V550M), c.1513G>C (p.G505R), c.772_781delACAAGCAAGG (p.T258fs) Sequencing | NM_001242785:4-12

Homocystinuria Caused by CBS Deficiency (CBS): Mutations (8): ♂ Genotyping | c.919G>A (p.G307S), c.833T>C (p.I278T), c.1006C>T (p.R336C), c.959T>C (p.V320A), c.797G>A (p.R266K), c.572C>T (p.T191M), c.341C>T (p.A114V), c.969G>A (p.W324X) Sequencing | NM_001178008:3-17

Hurler Syndrome (IDUA): Mutations (8): ♂ Genotyping | c.1598C>G (p.P533R), c.208C>T (p.Q70X), c.1205G>A (p.W402X), c.979G>C (p.A327P), c.266G>A (p.R89Q), c.1960T>G (p.X654G), c.152G>A (p.G51D), c.1037T>G (p.L346R) Sequencing | NM_000203:2-8, 11-14

Hypophosphatasia (ALPL): Mutations (5): ♂ Genotyping | c.1559delT, c.1133A>T (p.D378V), c.1001G>A (p.G334D), c.571G>A (p.E191K), c.979T>C (p.F327L) Sequencing | NM_000478:2-12

Inclusion Body Myopathy: Type 2 (GNE): Mutations (3): ♂ Genotyping | c.2228T>C (p.M743T), c.1807G>C (p.V603L), c.131G>C (p.C44S) Sequencing | NM_001128227:1-12

Infantile Cerebral and Cerebellar Atrophy (MED17): Mutations (1): ♂ Genotyping | c.1112T>C (p.L371P) Sequencing | NM_004268:1-12

Isolated Microphthalmia: VSX2 Related (VSX2): Mutations (4): ♂ Genotyping | c.599G>A (p.R200Q), c.599G>C (p.R200P), c.679C>T (p.R227W), c.371-1G>A Sequencing | NM_182894:1-5

Isovaleric Acidemia (IVD): Mutations (1): ♂ Genotyping | c.941C>T (p.A314V) Sequencing | NM_002225:1-12

Joubert Syndrome (TMEM216): Mutations (2): ♂ Genotyping | c.218G>T (p.R73L), c.218G>A (p.R73H) Sequencing | NM_001173991:1-5

Lamellar Ichthyosis: Type 1 (TGM1): Mutations (1): ♂ Genotyping | c.877-2A>G (IVS5-2A>G) Sequencing | NM_000359:2-15

Laryngoonychocutaneous Syndrome (LAMA3): Mutations (1): ♂ Genotyping | c.151_152insG (p.V51GfsX3) Sequencing | NM_000227:1-38

Leber Congenital Amaurosis: CEP290 Related (CEP290): Mutations (1): ♂ Genotyping | c.2991+1655A>G (p.C998X) Sequencing | NM_025114:2-54

Leber Congenital Amaurosis: GUCY2D Related (GUCY2D): Mutations (3): ♂

Genotyping | c.1694T>C (p.F565S), c.2943delG (p.G982V), c.387delC (p.P130Lfx) Sequencing | NM_000180:2-19

Leber Congenital Amaurosis: LCA5 Related (LCA5): Mutations (3): ♂ Genotyping | c.835C>T (p.Q279X), c.1476_1477insA (p.P493TfsX1), c.1151delC Sequencing | NM_001122769:2-8

Leber Congenital Amaurosis: RDH12 Related (RDH12): Mutations (6): ♂ Genotyping | c.565C>T (p.Q189X), c.184C>T (p.R62X), c.464C>T (p.T155I), c.677A>G (p.Y226C), c.146C>T (p.T49M), c.295C>A (p.L99I) Sequencing | NM_152443:3-9

Leigh Syndrome: French-Canadian (LRPPRC): Mutations (1): ♂ Genotyping | c.1061C>T (p.A354V) Sequencing | NM_133259:1-38

Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related (EIF2B5): Mutations (9): ♂ Genotyping | c.338G>A (p.R113H), c.271A>G (p.T91A), c.1882T>C (p.W628R), c.1157G>T (p.G386V), c.584G>A (p.R195H), c.925G>C (p.V309L), c.944G>A (p.R315H), c.166T>G (p.F56V), c.167T>G (p.F56C) Sequencing | NM_003907:1-16

Leydig Cell Hypoplasia (Luteinizing Hormone Resistance) (LHCGR): Mutations (13): ♂ Genotyping | c.1822_1827delCTGGTT (p.608_609delLV), c.1777G>C (p.A593P), c.1660C>T (p.R554X), c.1060G>A (p.E354K), c.1635C>A (p.C545X), c.391T>C (p.C131R), c.1027T>A (p.C343S), c.1627T>C (p.C543R), c.1505T>C (p.L502P), c.430G>T (p.V144F), c.1847C>A (p.S616Y), c.455T>C (p.I152T), c.537-3C>A Sequencing | NM_000233:1-11

Limb-Girdle Muscular Dystrophy: Type 2A (CAPN3): Mutations (6): ♂ Genotyping | c.1715G>A (p.R572Q), c.1469G>A (p.R490Q), c.550delA (p.T184fs), c.2306G>A (p.R769Q), c.2362_2363delAGinsTCATCT (p.R788Sfs), c.1525G>T (p.V509F) Sequencing | NM_000070:1-24

Limb-Girdle Muscular Dystrophy: Type 2B (DYSF): Mutations (5): ♂ Genotyping | c.4989_4993delGCCCGinsCCCC (p.E1663fs), c.2833delG (p.A945fs), c.5830C>T (p.R1944X), c.2271C>A (p.Y758X), c.5174+5G>A Sequencing | NM_001130987:1-56

Limb-Girdle Muscular Dystrophy: Type 2C (SGCG): Mutations (4): ♂ Genotyping | c.848G>A (p.C283Y), c.787G>A (p.E263K), c.525delT (p.F175fsX), c.87_88insT (p.G30fs) Sequencing | NM_000231:2-8

Limb-Girdle Muscular Dystrophy: Type 2D (SGCA): Mutations (1): ♂ Genotyping | c.229C>T (p.R77C) Sequencing | NM_000023:1-9

Limb-Girdle Muscular Dystrophy: Type 2E (SGCB): Mutations (6): ♂ Genotyping | c.341C>T (p.S114F), c.452C>G (p.T151R), c.272G>C (p.R91P), c.272G>T (p.R91L), c.299T>A (p.M100K), c.323T>G (p.L108R) Sequencing | NM_000232:2-6

Limb-Girdle Muscular Dystrophy: Type 2F (SGCD): Mutations (5): ♂ Genotyping | c.493C>T (p.R165X), c.89G>A (p.W30X), c.784G>A (p.E262K), c.391G>C (p.A131P), c.653delC (p.A218fs) Sequencing | NM_001128209:2-8

Limb-Girdle Muscular Dystrophy: Type 2I (FKRP): Mutations (1): ♂ Genotyping | c.826C>A (p.L276I) Sequencing | NM_001039885:1-4

Lipoprotein Lipase Deficiency (LPL): Mutations (1): ♂ Genotyping | c.644G>A (p.G215E) Sequencing | NM_000237:1-10

Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA): Mutations (2): ♂ Genotyping | c.1132C>T (p.Q378X), c.1528G>C (p.E510Q) Sequencing | NM_000182:1-20

Lysinuric Protein Intolerance (SLC7A7): Mutations (4): ♂ Genotyping | c.1228C>T (p.R410X), c.726G>A (p.W242X), c.1384_1385insATCA (p.R462fs), c.895-2A>T Sequencing | NM_001126105:3-11

MTHFR Deficiency: Severe (MTHFR): Mutations (6): ♂ Genotyping | c.1721T>G (p.V574G), c.1408G>T (p.E470X), c.1166G>A (p.W389X), c.652G> T (p.V218L), c.523G>A (p.A175T), c.474A>T (p.G158G) Sequencing | NM_005957:2-12

Malonyl-CoA Decarboxylase Deficiency (MLYCD): Mutations (5): ♂ Genotyping | c.560C>G (p.S187X), c.8G>A (p.G3D), c.1064_1065delTT (p.F355fs), c.949-14A>G, c.638_641delGTGA (p.S213fs) Sequencing | NM_012213:1-5

Maple Syrup Urine Disease: Type 1A (BCKDHA): Mutations (4): ♂ Genotyping | c.860_867delGAGGCCCC, c.868G>A (p.G290R), c.1312T>A (p.Y438N), c.288+1G>A Sequencing | NM_000709:1-9

Maple Syrup Urine Disease: Type 1B (BCKDHB): Mutations (6): ♂ Genotyping | c.1114G>T (p.E372X), c.548G>C (p.R183P), c.832G>A (p.G278S), c.970C>T (p.R324X), c.487G>T (p.E163X), c.853C>T (p.R285X) Sequencing | NM_183050:1-10

Maple Syrup Urine Disease: Type 2 (DBT): Mutations (15): ♂ Genotyping | c.670G>T (p.E224X), c.581C>G (p.S194X), c.1355A>G (p.H452R), c.294C>G (p.I98M), c.1448G>T (p.X483L), c.75_76delAT (p.C26Wfs), c.901C>T (p.R301C), c.363_364delCT (p.Y122Lfs), c.1193T>C (p.L398P), c.1169A>G (p.D390G), c.1209+5G>C (IVS9+5G>C), c.1232C>A (p.P411Q), c.939G>C (p.K313N), c.788T>G (p.M263R), c.1202T>C (p.I401T) Sequencing | NM_001918:1-11

Maple Syrup Urine Disease: Type 3 (DLD): Mutations (8): ♂ Genotyping | c.104_105insA, c.685G>T (p.G229C), c.214A>G (p.K72E), c.1081A>G (p.M361V), c.1123G>A (p.E375K), c.1178T>C (p.I393T), c.1463C>T (p.P488L), c.1483A>G (p.R495G) Sequencing | NM_000108:1-14

Maroteaux-Lamy Syndrome (ARSB): Mutations (6): ♂ Genotyping | c.629A>G (p.Y210C), c.1178A>C (p.H393P), c.284G>A (p.R95Q), c.944G>A (p.R315Q), c.1143-8T>G, c.1143-1G>C Sequencing | NM_000046:1-8

Meckel Syndrome: Type 1 (MKS1): Mutations (5): ♂ Genotyping | c.1408-35_1408-7del29 (p.G470fs), c.80+2T>C (IVS1+2T>C), c.1024+1G>A (IVS11+1G>A), c.417G>A (p.E139X), c.50insCCGGG (p.D19AfsX) Sequencing | NM_017777:1-18

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (ACADM): Mutations (8): ♂ Genotyping | c.985A>G (p.K329E), c.362C>T (p.T121I), c.583G>A (p.G195R), c.799G>A (p.G267R), c.199T>C (p.Y67H), c.262C>T (p.L88F), c.616C>T (p.R206C), c.617G>A (p.C206H) Sequencing | NM_001127328:1-12

Megalencephalic Leukoencephalopathy (MLC1): Mutations (6): ♂ Genotyping | c.176G>A (p.G59E), c.278C>T (p.S93L), c.135_136insC (p.C46fsX), c.908_918delTGCTGCTGCTGinsGCA (p.V303GfsX96), c.880C>T (p.P294S), c.178-10T>A Sequencing | NM_139202:2-12

Metachromatic Leukodystrophy (ARSA): Mutations (18): ♂ Genotyping | c.1210+1G>A, c.465+1G>A (IVS2+1G>A), c.862A>C (p.T288P), c.1136C>T (p.P379L), c.1283C>T (p.P428L), c.827C>T (p.T276M), c.542T>G (p.I181S), c.1232C>T (p.T411I), c.769G>C (p.D257H), c.739G>A (p.G247R), c.641C>T (p.A214V), c.302G>A (p.G101D), c.293C>T (p.S98F), c.257G>A (p.R86Q), c.263G>A (p.G88D), c.1114C>T (p.R372W), c.292_293delTCinsCT (p.S98L), c.302G>T (p.G101V) Sequencing | NM_001085425:2-9

Methylmalonic Acidemia: MMAA Related (MMAA): Mutations (14): ♂ Genotyping | c.64C>T (p.R22X), c.161G>A (p.W54X), c.266T>C (p.L89P), c.283C>T (p.Q95X), c.358C>T (p.Q120X), c.397C>T (p.Q133X), c.433C>T (p.R145X), c.503delC (p.T168MfsX9), c.562G>C (p.G188R), c.650T>A (p.L217X), c.653G>A (p.G218E), c.733+1G>A, c.988C>T (p.R330X), c.1076G>A (p.R359Q) Sequencing | NM_172250:2-7

Methylmalonic Acidemia: MMAB Related (MMAB): Mutations (11): ♂ Genotyping | c.700C>T (p.Q234X), c.656A>G (p.Y219C), c.572G>A (p.R191Q), c.571C>T (p.R191W), c.569G>A (p.R190H), c.568C>T (p.R190C), c.556C>T (p.R186W), c.403G>A (p.A135T), c.291-1G>A, c.287T>C (p.I96T), c.197-1G>T Sequencing | NM_052845:1-9

Methylmalonic Acidemia: MUT Related (MUT): Mutations (23): ♂ Genotyping | c.2150G>T (p.G717V), c.2099T>A (p.M700K), c.2080C>T (p.R694W), c.2054T>G (p.L685R), c.1867G>A (p.G623R), c.1280G>A (p.G427D), c.1106G>A (p.R369H), c.1105C>T (p.R369C), c.1097A>G (p.N366S), c.935G>T (p.G312V), c.691T>A (p.Y231N), c.655A>T (p.N219Y), c.643G>A (p.G215S), c.607G>A (p.G203R), c.572C>A (p.A191E), c.521T>C (p.F174S), c.322C>T (p.R108C), c.313T>C (p.W105R), c.299A>G (p.Y100C), c.284C>G (p.P95R), c.281G>T (p.G94V), c.278G>A (p.R93H), c.643G>T (p.G215C) Sequencing | NM_000255:2-13

Methylmalonic Aciduria and Homocystinuria: Type cblC (MMACHC): Mutations (5): ♂ Genotyping | c.271_272insA (p.R91KfsX14), c.331C>T (p.R111X), c.394C>T (p.R132X), c.482G>A (p.R161Q), c.609G>A (p.W203X) Sequencing | NM_015506:1-4

Mitochondrial Complex I Deficiency: NDUFS6 Related (NDUFS6): Mutations (1): ♂ Genotyping | c.344G>A (p.C115Y) Sequencing | NM_004553:1-4

Mitochondrial DNA Depletion Syndrome: MNGIE Type (TYMP): Mutations (6): ♂ Genotyping | c.866A>C (p.E289A), c.433G>A (p.G145R), c.665A>G (p.K222R), c.457G>A (p.G153S), c.516+2T>C (IVS4+2T>C), c.1425_1426insC (p.S476Lfs) Sequencing | NM_001257989:2-8,10

Mitochondrial Myopathy and Sideroblastic Anemia (PUS1): Mutations (2): ♂ Genotyping | c.430C>T (p.R144W), c.658G>T (p.E220X) Sequencing | NM_025215:1-6

Mitochondrial Trifunctional Protein Deficiency: HADHB Related (HADHB): Mutations (7): ♂ Genotyping | c.182G>A (p.R61H), c.788A>G (p.D263G), c.740G>A (p.R247H), c.1331G>A (p.R444K), c.1364T>G (p.V455G), c.776_777insT (p.G259fs), c.1175C>T (p.A392V) Sequencing | NM_000183:2-16

Morquio Syndrome: Type A (GALNS): Mutations (6): ♂ Genotyping | c.205T>G (p.F69V), c.485C>T (p.S162F), c.1156C>T (p.R386C), c.901G>T (p.G301C), c.337A>T (p.I113F), c.178G>A (p.D60N) Sequencing | NM_000512:2-14

Morquio Syndrome: Type B (GLB1): Mutations (8): ♂ Genotyping | c.1527G>T (p.W509C), c.1313G>A (p.G438E), c.1445G>A (p.R482H), c.247T>C (p.Y83H), c.1444C>T (p.R482C), c.1498A>G (p.T500A), c.1223A>C (p.Q408P), c.817_818delTGinsCT (p.W273L) Sequencing | NM_000404:1-16

Mucolipidosis: Type II/III (GNPTAB): Mutations (3): ♂ Genotyping | c.3503_3504delTC (p.L1168QfsX5), c.3565C>T (p.R1189X), c.1120T>C (p.F374L) Sequencing | NM_024312:1-21

Mucolipidosis: Type IV (MCOLN1): Mutations (5): ♂ Genotyping | c.-1015_788del6433, c.406-2A>G, c.1084G>T (p.D362Y), c.304C>T (p.R102X), c.244delC (p.L82fsX) Sequencing | NM_020533:1-14

Multiple Pterygium Syndrome (CHRNG): Mutations (6): ♂ Genotyping | c.715C>T (p.R239C), c.13C>T (p.Q5X), c.320T>G (p.V107G), c.401_402delCT (p.P134fs), c.1408C>T (p.R470X), c.136C>T (p.R46X) Sequencing | NM_005199:1-12

Multiple Sulfatase Deficiency (SUMF1): Mutations (1): ♂ Genotyping | c.463T>C (p.S155P) Sequencing | NM_182760:1-9

Muscle-Eye-Brain Disease (POMGNT1): Mutations (3): ♂ Genotyping | c.1539+1G>A , c.1324C>T (p.R442C), c.1478C>G (p.P493R) Sequencing | NM_001243766:2-23

Navajo Neurohepatopathy (MPV17): Mutations (1): ♂ Genotyping | c.149G>A (p.R50Q) Sequencing | NM_002437:2-8

Nemaline Myopathy: NEB Related (NEB): Mutations (2): ♂ Genotyping | c.7434_7536del2502bp, c.8890-2A>G (IVS63-2A>G) Sequencing | NM_001164508:63-66,86,95-96,103,105,143,168-172, NM_004543:3-149

Nephrotic Syndrome: Type 1 (NPHS1): Mutations (5): ♂ Genotyping | c.121_122delCT (p.L41Dfs), c.1481delC, c.3325C>T (p.R1109X), c.3478C>T (p.R1160X), c.2335-1G>A Sequencing | NM_004646:1-29

Nephrotic Syndrome: Type 2 (NPHS2): Mutations (27): ♂ Genotyping | c.976_977insA (p.T326fsX345), c.964C>T (p.R322X), c.948delT (p.A317L), c.871C>T (p.R291W), c.868G>A (p.V290M), c.862G>A (p.A288T), c.855_856delAA (p.Q285fsX302), c.851C>T (p.A284V), c.779T>A (p.V260E), c.714G>T (p.R238S), c.706_714del CTAGAGAGG (p.L236_R238del), c.622G>A (p.A208T), c.555delT (p.F185fsX186), c.538G>A (p.V180M), c.503G>A (p.R168H), c.502C>A (p.R168S), c.502C>T (p.R168C), c.479A>G (p.D160G), c.467delT (p.L156fsX180), c.467_468insT (p.L156fsX166), c.419delG (p.G140fsX180), c.413G>A (p.R138Q), c.412C>T (p.R138X), c.353C>T (p.P118L), c.274G>T (p.G92C), c.104_105insG (p.G35fsX69), c.85G>A (p.A29T) Sequencing | NM_014625:1-8

Neuronal Ceroid-Lipofuscinosis: CLN5 Related (CLN5): Mutations (7): ♂ Genotyping | c.1175_1176delAT (p.Y392X), c.225G>A (p.W75X), c.835G>A (p.D279N), c.335G>A (p.R112H), c.377G>A (p.C126Y), c.1054G>T (p.E352X), c.1121A>G (p.Y374C) Sequencing | NM_006493:1-4

Neuronal Ceroid-Lipofuscinosis: CLN6 Related (CLN6): Mutations (8): ♂ Genotyping | c.663C>G (p.Y221X), c.460_462delATC (p.I154del), c.368G>A (p.G123D), c.308G>A (p.R103Q), c.214G>T (p.E72X), c.200T>C (p.L67P), c.139C>T (p.L47F), c.17G>C (p.R6T) Sequencing | NM_017882:2-7

Neuronal Ceroid-Lipofuscinosis: CLN8 Related (CLN8): Mutations (4): ♂ Genotyping | c.70C>G (p.R24G), c.789G>C (p.W263C), c.88G>C (p.A30P), c.610C>T (p.R204C) Sequencing | NM_018941:2-3

Neuronal Ceroid-Lipofuscinosis: MFSD8 Related (MFSD8): Mutations (2): ♂ Genotyping | c.881C>A (p.T294K), c.754+2T>A Sequencing | NM_152778:2-13

Neuronal Ceroid-Lipofuscinosis: PPT1 Related (PPT1): Mutations (8): ♂ Genotyping | c.223A>C (p.T75P), c.364A>T (p.R122W), c.451C>T (p.R151X), c.29T>A (p.L10X), c.656T>A (p.L219Q), c.322G>C (p.G108R), c.236A>G (p.D79G), c.134G>A (p.C45Y) Sequencing | NM_000310:1-9

Neuronal Ceroid-Lipofuscinosis: TPP1 Related (TPP1): Mutations (9): ♂ Genotyping | c.523-1G>A, c.509-1G>C, c.622C>T (p.R208X), c.851G>T (p.G284V), c.1340G>A (p.R477H), c.1094G>A (p.C365Y), c.1093T>C (p.C365R), c.857A>G (p.N286S), c.616C>T (p.R206C) Sequencing | NM_000391:1-13

Niemann-Pick Disease: Type A (SMPD1): Mutations (6): ♂ Genotyping | c.996delC, c.1493G>T (p.R498L), c.911T>C (p.L304P), c.1267C>T (p.H423Y), c.1734G>C (p.K578N), c.1493G>A (p.R498H) Sequencing | NM_000543:1-6

Niemann-Pick Disease: Type B (SMPD1): Mutations (3): ♂ Genotyping | c.1828_1830delCGC (p.610delR), c.880C>A (p.Q294K), c.1280A>G (p.H427R) Sequencing | NM_000543:1-6

Niemann-Pick Disease: Type C1 (NPC1): Mutations (14): ♂ Genotyping | c.2783A>C (p.Q928P), c.3263A>G (p.Y1088C), c.3467A>G (p.N1156S), c.3107C>T (p.T1036M), c.3182T>C (p.I1061T), c.2974G>C (p.G992R), c.2932C>T (p.R978C), c.2848G>A (p.V950M), c.2665G>A (p.V889M), c.2324A>C (p.Q775P), c.1133T>C (p.V378A), c.530G>A (p.C177Y), c.337T>C (p.C113R), c.2974G>T (p.G992W) Sequencing | NM_000271:1-25

Niemann-Pick Disease: Type C2 (NPC2): Mutations (11): ♂ Genotyping | c.58G>T (p.E20X), c.436C>T (p.Q146X), c.358C>T (p.P120S), c.352G>T (p.E118X), c.332delA (p.N111Ifs), c.295T>C (p.C99R), c.199T>C (p.S67P), c.190+5G>A, c.141C>A (p.C47X), c.133C>T (p.Q45X), c.115G>A (p.V39M) Sequencing | NM_006432:1-5

Nijmegen Breakage Syndrome (NBN): Mutations (1): ♂ Genotyping | c.657_661delACAAA (p.K219fs) Sequencing | NM_002485:1-16

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutations (29): ♂ Genotyping | c.167delT, c.235delC, c.313_326delAAGTTCATCAAGGG, c.358delGAG (p.120delE), c.35delG, c.370C>T (p.Q124X), c.427C>T (p.R143W), c.109G>A (p.V37I), c.231G>A (p.W77X), c.551G>C (p.R184P), c.71G>A (p.W24X), c.229T>C (p.W77R), c.269T>C (p.L90P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T, c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147K), c.-23+1G>A, c.550C>T (p.R184W), c.-259C>T Sequencing | NM_004004:1-2

Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related (LOXHD1): Mutations (2): ♂ Genotyping | c.2008C>T (p.R670X), c.4714C>T (p.R1572X) Sequencing | NM_144612:1-40

Nonsyndromic Hearing Loss and Deafness: MYO15A Related (MYO15A): Mutations (10): ♂ Genotyping | c.453_455delCGAinsTGGACGCCTGGTCGGGCAGTGG (p.E152GfsX81), c.7801A>T (p.K2601X), c.6337A>T (p.I2113F), c.3866+1G>T, c.3313G>T (p.E1105X), c.3334delG (p.G1112fs), c.8148G>T (p.Q2716H), c.6331A>T (p.N2111Y), c.3685C>T (p.Q1229X), c.3866+1G>A Sequencing | NM_016239:2-65

Oculocutaneous Albinism: Type 1 (TYR): Mutations (27): ♂ Genotyping | c.272G>A (p.C91Y), c.242C>T (p.P81L), c.265T>C (p.C89R), c.1A>G (p.M1V), c.140G>A (p.G47D), c.325G>A (p.G109R), c.568delG (p.G191Dfs), c.707G>A (p.W236X), c.832C>T (p.R278X), c.1118C>A (p.T373K), c.229C>T (p.R77W), c.823G>T (p.V275F), c.32G>A (p.W11X), c.149C>T (p.S50L), c.1467_1468insT (p.A490Cfs), c.820-2A>G, c.892C>T (p.R298W), c.1064C>T (p.A355V), c.1090A>C (p.N364H), c.1150C>G (p.P384A), c.1184+1G>A, c.1309G>A (p.D437N), c.1469C>A (p.A490D), c.133_134insC (p.P45fs), c.710delA (p.D237fs), c.978delA (p.Q326fs), c.1138_1158delTCTGCCAACGATCCTATCTTC (p.S380_F386del) Sequencing | NM_000372:1-5

Oculocutaneous Albinism: Type 3 (TYRP1): Mutations (6): ♂ Genotyping | c.1067G>A (p.R356Q), c.497C>G (p.S166X), c.107delT, c.1057_1060delAACA (p.N353fs), c.1103delA (p.K368fs), c.1120C>T (p.R374X) Sequencing | NM_000550:2-8

Oculocutaneous Albinism: Type 4 (SLC45A2): Mutations (2): ♂ Genotyping | c.469G>A (p.D157N), c.563G>T (p.G188V) Sequencing | NM_016180:1-7

Omenn Syndrome: DCLRE1C Related (DCLRE1C): Mutations (1): ♂ Genotyping | c.597C>A (p.Y199X) Sequencing | NM_001033855:1-14

Omenn Syndrome: RAG2 Related (RAG2): Mutations (1): ♂ Genotyping | c.685C>T (p.R229W) Sequencing | NM_000536:1-2

Ornithine Translocase Deficiency (SLC25A15): Mutations (3): ♂ Genotyping | c.562_564delTTC (p.188delF), c.95C>G (p.T32R), c.535C>T (p.R179X) Sequencing | NM_014252:2-7

Osteopetrosis: TCIRG1 Related (TCIRG1): Mutations (6): ♂ Genotyping | c.1674-1G>A, c.1392C>A (p.C464X), c.117+4A>T, c.1213G>A (p.G405R), c.1331G>T (p.R444L), c.922delC (p.Q308fs) Sequencing | NM_006019:1-20

POLG Related Disorders: Autosomal Recessive (POLG): Mutations (16): ♂ Genotyping | c.695G>A (p.R232H), c.752C>T (p.T251I), c.1399G>A (p.A467T), c.1760C>T (p.P587L), c.2243G>C (p.W748S), c.2542G>A (p.G848S), c.3488T>G (p.M1163R), c.911T>G (p.L304R), c.8G>C (p.R3P), c.2617G>T (p.E873X), c.2794C>T (p.H932Y), c.3151G>C (p.G1051R), c.2591A>G (p.N864S), c.1491G>C (p.Q497H), c.679C>T (p.R227W), c.3218C>T (p.P1073L) Sequencing | NM_001126131:2-23

Papillon-Lefevre Syndrome (CTSC): Mutations (11): ♂ Genotyping | c.815G>A (p.R272H), c.96T>G (p.Y32X), c.380A>C (p.H127P), c.1287G>C (p.W429C), c.856C>T (p.Q286X), c.755A>T (p.Q252L), c.628C>T (p.R210X), c.857A>G (p.Q286R), c.890-1G>A, c.1047delA (p.G350Vfs), c.1056delT (p.Y352fs) Sequencing | NM_001814:1-7

Pendred Syndrome (SLC26A4): Mutations (7): ♂ Genotyping | c.1001+1G>A, c.1151A>G (p.E384G), c.1246A>C (p.T416P), c.2168A>G (p.H723R), c.707T>C (p.L236P), c.716T>A (p.V239D), c.919-2A>G Sequencing | NM_000441:1-21

Persistent Mullerian Duct Syndrome: Type I (AMH): Mutations (6): ♂ Genotyping | c.1144G>T (p.E382X), c.571C>T (p.R191X), c.1518C>G (p.H506Q), c.1574G>A (p.C525Y), c.17_18delTC, c.283C>T (p.R95X) Sequencing | NM_000479:1-4

Persistent Mullerian Duct Syndrome: Type II (AMHR2): Mutations (14): ♂ Genotyping | c.232+1G>A, c.1330_1356delCTGGGCAATACCCCTACCTCTGATGAG, c.596delA, c.1217G>A (p.R406Q), c.742G>A (p.E248K), c.1277A>G (p.D426G), c.846T>G (p.H282Q), c.1373T>C (p.V458A), c.1471G>C (p.D491H), c.1510C>T (p.R504C), c.118G>T (p.G40X), c.289C>T (p.R97X), c.160C>T (p.R54C), c.425G>T (p.G142V) Sequencing | NM_020547:1-11

Phenylalanine Hydroxylase Deficiency (PAH): Mutations (61): ♂ Genotyping | c.1066-11G>A (IVS10-11G>A), c.1315+1G>A (IVS12+1G>A), c.1241A>G (p.Y414C), c.1222C>T (p.R408W), c.754C>T (p.R252W), c.1223G>A (p.R408Q), c.473G>A (p.R158Q), c.782G>A (p.R261Q), c.814G>T (p.G272X), c.143T>C (p.L48S), c.194T>C (p.I65T), c.896T>G (p.F299C), c.842C>T (p.P281L), c.838G>A (p.E280K), c.117C>G (p.F39L), c.3G>A (p.M1I), c.1A>G (p.M1V), c.611A>G (p.Y204C), c.721C>T (p.R241C), c.727C>T (p.R243X), c.1139C>T (p.T380M), c.926C>T (p.A309V), c.898G>T (p.A300S), c.734T>C (p.V245A), c.818C>T (p.S273F), c.997C>T (p.L333F), c.199T>C (p.S67P), c.1042C>G (p.L348V), c.136G>A (p.G46S), c.728G>A (p.R243Q), c.745C>T (p.L249F), c.581T>C (p.L194P), c.722G>T (p.R241L), c.829T>G (p.Y277D), c.899C>T (p.A300V), c.926C>A (p.A309D), c.1045T>C (p.S349P), c.1157A>G (p.Y386C), c.1169A>G (p.E390G), c.331C>T (p.R111X), c.241_256delACCCATTTGGATAAAC (p.T81fs), c.442-1G>A (IVS4-1G>A), c.463_464insTGTGTACC (p.R155fs), c.569T>G (p.V190G), c.682G>T (p.E228X), c.755G>A (p.R252Q), c.770G>T (p.G257V), c.781C>T (p.R261X), c.800A>G (p.Q267R), c.842+5G>A (IVS7+5G>A), c.856G>A (p.E286K), c.904delT (p.F302fs), c.913-7A>G (IVS8-7A>G), c.935G>T (p.G312V), c.1068C>G (p.Y356X), c.1238G>C (p.R413P), c.1301C>A (p.A434D), c.842+2T>A (IVS7+2T>A), c.764T>C (p.L255S), c.722G>A (p.R241H), c.533A>G (p.E178G) Sequencing | NM_000277:1-13

Polyglandular Autoimmune Syndrome: Type I (AIRE): Mutations (5): ♂ Genotyping | c.769C>T (p.R257X), c.254A>G (p.Y85C), c.1163_1164insA (p.M388IfsX36), c.967_979delCTGTCCCCTCCGC (p.L323SfsX51), c.415C>T (p.R139X) Sequencing | NM_000383:1-14

Pontocerebellar Hypoplasia: EXOSC3 Related (EXOSC3): Mutations (4): ♂ Genotyping | c.395A>C (p.D132A), c.294_303delTGTTTACTGG (p.V99Wfs), c.92G>C (p.G31A), c.238G>T (p.V80F) Sequencing | NM_016042:1-4

Pontocerebellar Hypoplasia: RARS2 Related (RARS2): Mutations (3): ♂ Genotyping | c.35A>G (p.Q12R), c.110+5A>G, c.1024A>G (p.M342V) Sequencing | NM_020320:1-20

Pontocerebellar Hypoplasia: SEPSECS Related (SEPSECS): Mutations (1): ♂ Genotyping | c.1001A>G (p.Y334C) Sequencing | NM_016955:1-11

Pontocerebellar Hypoplasia: TSEN54 Related (TSEN54): Mutations (3): ♂ Genotyping | c.919G>T (p.A307S), c.736C>T (p.Q246X), c.1027C>T (p.Q343X) Sequencing | NM_207346:3-11

Pontocerebellar Hypoplasia: VPS53 Related (VPS53): Mutations (2): ♂ Genotyping | c.2084A>G (p.Q695R), c.1556+5G>A Sequencing | NM_001128159:1-22

Pontocerebellar Hypoplasia: VRK1 Related (VRK1): Mutations (2): ♂ Genotyping |

c.1072C>T (p.R358X), c.397C>T (p.R133C) Sequencing | NM_003384:2-13

Primary Carnitine Deficiency (SLC22A5): Mutations (12): ♂ Genotyping | c.506G>A (p.R169Q), c.396G>A (p.W132X), c.1195C>T (p.R399W), c.1433C>T (p.P478L), c.43G>T (p.G15W), c.1324_1325delGCinsAT (p.A442I), c.632A>G (p.Y211C), c.1202_1203insA (p.Y401fsX), c.844C>T (p.R282X), c.505C>T (p.R169W), c.1196G>A (p.R399Q), c.95A>G (p.N32S) Sequencing | NM_003060:1-10

Primary Ciliary Dyskinesia: DNAI1 Related (DNAI1): Mutations (5): ♂ Genotyping | c.282_283insAATA (p.G95Nfs), c.1543G>A (p.G515S), c.48+2_48+3insT, c.1658_1669delCCAAGGTCTTCA (p.Thr553_Phe556del), c.1490G>A (p.G497D) Sequencing | NM_012144:1-20

Primary Ciliary Dyskinesia: DNAI2 Related (DNAI2): Mutations (4): ♂ Genotyping | c.1494+1G>A, c.346-3T>G, c.787C>T (p.R263X), c.1304G>A (p.W435X) Sequencing | NM_023036:2-13

Primary Congenital Glaucoma (CYP1B1): Mutations (9): ♂ Genotyping | c.1405C>T (p.R469W), c.1093G>T (p.G365W), c.155C>T (p.P52L), c.1064_1076delGAGTGCAGGCAGA (p.R355Hfs), c.1410_1422delCATTGGCGAAGAA (p.C470fs), c.862_863insC, c.1199_1200insTCATGCCACC, c.182G>A (p.G61E), c.535delG (p.A179fs) Sequencing | NM_000104:2-3

Primary Hyperoxaluria: Type 1 (AGXT): Mutations (11): ♂ Genotyping | c.508G>A (p.G170R), c.454T>A (p.F152I), c.731T>C (p.I244T), c.121G>A (p.G41R), c.198C>G (p.Y66X), c.245G>A (p.G82E), c.466G>A (p.G156R), c.613T>C (p.S205P), c.697C>T (p.R233C), c.698G>A (p.R233H), c.738G>A (p.W246X) Sequencing | NM_000030:1-11

Primary Hyperoxaluria: Type 2 (GRHPR): Mutations (3): ♂ Genotyping | c.103delG, c.404+3delAAGT, c.295C>T (p.R99X) Sequencing | NM_012203:1-9

Primary Hyperoxaluria: Type 3 (HOGA1): Mutations (2): ♂ Genotyping | c.944_946delAGG (p.315delE), c.860G>T (p.G287V) Sequencing | NM_138413:1-7

Progressive Familial Intrahepatic Cholestasis: Type 2 (ABCB11): Mutations (5): ♂ Genotyping | c.3767_3768insC, c.890A>G (p.E297G), c.1723C>T (p.R575X), c.3169C>T (p.R1057X), c.1295G>C (p.R432T) Sequencing | NM_003742:2-28

Propionic Acidemia: PCCA Related (PCCA): Mutations (13): ♂ Genotyping | c.862A>G (p.R288G), c.937C>T (p.R313X), c.1196G>A (p.R399Q), c.1685C>G (p.S562X), 916_917insT, c.1192T>C (p.C398R), c.229C>T (p.R77W), c.590G>A (p.G197E), c.1643+1G>A (IVS18+1G>A), c.890A>G (p.Q297R), c.1644-6C>G (IVS18-6C>G), c.1746G>A (p.S582S), c.1268C>T (p.P423L) Sequencing | NM_000282:1-24

Propionic Acidemia: PCCB Related (PCCB): Mutations (13): ♂ Genotyping | c.280G>T (p.G94X), c.335G>A (p.G112D), c.457G>C (p.A153P), c.502G>A (p.E168K), c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA (p.G407fs), c.1228C>T (p.R410W), c.1283C>T (p.T428I), c.1304A>G (p.Y435C), c.1495C>T (p.R499X), c.1534C>T (p.R512C), c.1539_1540insCCC (p.R514PfsX38), c.1556T>C (p.L519P), c.1606A>G (p.N536D) Sequencing | NM_000532:1-15

Pseudocholinesterase Deficiency (BCHE): Mutations (1): ♂ Genotyping | c.293A>G (p.D98G) Sequencing | NM_000055:2-4

Pycnodysostosis (CTSK): Mutations (2): ♂ Genotyping | c.990A>G (p.X330W), c.926T>C (p.L309P) Sequencing | NM_000396:2-8

Pyruvate Carboxylase Deficiency (PC): Mutations (15): ♂ Genotyping | c.1892G>A (p.R631Q), c.184C>T (p.R62C), c.2540C>T (p.A847V), c.1351C>T (p.R451C), c.467G>A (p.R156Q), c.1828G>T (p.A610S), c.2229G>T (p.M743I), c.434T>C (p.V145A), c.1748G>T (p.R583L), c.2491_2492delGT (p.V831fs), c.3409_3410delCT (p.L1137fs), c.2493_2494delGT (p.F832Xfs), c.2876_2877insT (p.F959fs), c.2473+2_2473+5delTAGG, c.1828G>A (p.A610T) Sequencing | NM_022172:2-21

Pyruvate Dehydrogenase Deficiency (PDHB): Mutations (2): ♂ Genotyping | c.395A>G (p.Y132C), c.1030C>T (p.P344S) Sequencing | NM_000925:1-10

Renal Tubular Acidosis and Deafness (ATP6V1B1): Mutations (7): ♂ Genotyping | c.242T>C (p.L81P), c.232G>A (p.G78R), c.1248+1G>C, c.585+1G>A, c.497delC (p.T166fs), c.1037C>G (p.P346R), c.1155_1156insC (p.I386fs) Sequencing | NM_001692:1-14

Retinal Dystrophies: RLBP1 Related (RLBP1): Mutations (3): ♂ Genotyping | c.700C>T (p.R234W), c.141G>A (p.K47=), c.141+2T>C Sequencing | NM_000326:3-9

Retinal Dystrophies: RPE65 Related (RPE65): Mutations (12): ♂ Genotyping | c.1292A>G (p.Y431C), c.1102T>C (p.Y368H), c.11+5G>A, c.700C>T (p.R234X), c.1087C>A (p.P363T), c.1022T>C (p.L341S), c.271C>T (p.R91W), c.1355T>G (p.V452G), c.1543C>T (p.R515W), c.907A>T (p.K303X), c.1067delA (p.N356fs), c.95-2A>T (IVS2-2A>T) Sequencing | NM_000329:1-14

Retinitis Pigmentosa: CERKL Related (CERKL): Mutations (5): ♂ Genotyping | c.420delT (p.I141Lfs), c.598A>T (p.K200X), c.780delT (p.P261Lfs), c.769C>T (p.R257X), c.238+1G>A (IVS1+1G>A) Sequencing | NM_201548:1-13

Retinitis Pigmentosa: DHDDS Related (DHDDS): Mutations (1): ♂ Genotyping | c.124A>G (p.K42E) Sequencing | NM_024887:2-9

Retinitis Pigmentosa: FAM161A Related (FAM161A): Mutations (5): ♂ Genotyping | c.685C>T (p.R229X), c.1309A>T, c.1355_1356delCA (p.T452fs), c.1567C>T (p.R523X), c.1786C>T (p.R596X) Sequencing | NM_001201543:1-7

Rhizomelic Chondrodysplasia Punctata: Type I (PEX7): Mutations (8): ♂ Genotyping | c.903+1G>C, c.649G>A (p.G217R), c.875T>A (p.L292X), c.40A>C (p.T14P), c.45_52insGGGACGCC (p.H18RfsX35), c.120C>G (p.Y40X), c.345T>G (p.Y115X), c.653C>T (p.A218V) Sequencing | NM_000288:1-10

Salla Disease (SLC17A5): Mutations (5): ♂ Genotyping | c.802_816delTCATCATTAAGAAAT (p.L336fsX13), c.406A>G (p.K136E), c.115C>T (p.R39C), c.548A>G (p.H183R), c.1001C>G (p.P334R) Sequencing | NM_012434:1-11

Sandhoff Disease (HEXB): Mutations (14): ♂ Genotyping | c.76delA, c.445+1G>A, c.850C>T (p.R284X), c.508C>T (p.R170X), c.796T>G (p.Y266D), c.845G>A (p.G282E), c.800_816delCACCAAATGATGTCCGT (p.T267fs), c.1082+5G>A, c.1250C>T (p.P417L), c.1615C>T (p.R539C), c.1514G>A (p.R505Q), c.1303_1304delAG (p.R435fs), c.1509-26G>A, c.1597C>T (p.R533C) Sequencing | NM_000521:1-14

Sanfilippo Syndrome: Type A (SGSH): Mutations (11): ♂ Genotyping | c.734G>A (p.R245H), c.220C>T (p.R74C), c.197C>G (p.S66W), c.449G>A (p.R150Q), c.1339G>A (p.E447K), c.1105G>A (p.E369K), c.1298G>A (p.R433Q), c.383C>T (p.P128L), c.617G>C (p.R206P), c.892T>C (p.S298P), c.1080delC (p.T360fs) Sequencing | NM_000199:1-8

Sanfilippo Syndrome: Type B (NAGLU): Mutations (10): ♂ Genotyping | c.2021G>A (p.R674H), c.889C>T (p.R297X), c.1928G>A (p.R643H), c.1927C>T (p.R643C), c.1562C>T (p.P521L), c.1444C>T (p.R482W), c.1693C>T (p.R565W), c.1694G>C (p.R565P), c.700C>T (p.R234C), c.1876C>T (p.R626X) Sequencing | NM_000263:2-6

Sanfilippo Syndrome: Type C (HGSNAT): Mutations (13): ♂ Genotyping | c.848C>T (p.P283L,p.P311L), c.962T>G (p.L321X), c.1529T>A (p.M510K), c.1030C>T (p.R344C), c.1553C>T (p.S518F), c.1150C>T (p.R384X), c.493+1G>A (IVS4+1G>A), c.372-2A>G (IVS3-2A>G), c.1622C>T (p.S541L), c.852-1G>A, c.525_526insT (p.A175fsX), c.1345insG (p.D449fsX), c.234+1G>A (IVS2+1G>A) Sequencing | NM_152419:2-18

Sanfilippo Syndrome: Type D (GNS): Mutations (5): ♂ Genotyping | c.1063C>T (p.R355X), c.1168C>T (p.Q390X), c.1226insG (p.R409fsX), c.1138insGTCCT (p.D380fsX), c.1169delA (p.Q390fsX) Sequencing | NM_002076:1-14

Short-Chain Acyl-CoA Dehydrogenase Deficiency (ACADS): Mutations (5): ♂ Genotyping | c.1058C>T (p.S353L), c.1138C>T (p.R380W), c.1147C>T (p.R383C), c.319C>T (p.R107C), c.575C>T (p.A192V) Sequencing | NM_000017:1-10

Sickle-Cell Anemia (HBB): Mutations (1): ♂ Genotyping | c.20A>T (p.E7V) Sequencing | NM_000518:1-3

Sjogren-Larsson Syndrome (ALDH3A2): Mutations (2): ♂ Genotyping | c.943C>T (p.P315S), c.1297_1298delGA (p.E433fs) Sequencing | NM_001031806:1-10

Sly Syndrome (GUSB): Mutations (5): ♂ Genotyping | c.526C>T (p.L176F), c.1244C>T (p.P415L), c.1222C>T (p.P408S), c.1856C>T (p.A629V), c.1429C>T (p.R477W) Sequencing | NM_000181:1-12

Smith-Lemli-Opitz Syndrome (DHCR7): Mutations (50): ♂ Genotyping | c.964-1G>C, c.356A>T (p.H119L), c.1054C>T (p.R352W), c.1210C>T (p.R404C), c.278C>T (p.T93M), c.1055G>A (p.R352Q), c.1139G>A (p.C380Y), c.1337G>A (p.R446Q), c.452G>A (p.W151X), c.453G>A (p.W151X), c.744G>T (p.W248C), c.976G>T (p.V326L), c.326T>C (p.L109P), c.470T>C (p.L157P), c.1342G>A (p.E448K), c.1228G>A (p.G410S), c.906C>G (p.F302L), c.725G>A (p.R242H), c.724C>T (p.R242C), c.506C>T (p.S169L), c.1A>G, c.670G>A (p.E224K), c.818T>G (p.V273G), c.203T>C (p.L68P), c.292C>T (p.Q98X), c.532A>T (p.I178F), c.545G>T (p.W182L), c.682C>T (p.R228W), c.575C>T (p.S192F), c.1295A>G (p.Y432C), c.1039G>A (p.G347S), c.1079T>C (p.L360P), c.1424T>C (p.F475S), c.1190C>T (p.S397L), c.1351T>C (p.C451R), c.853_855delTTC (p.285delF), c.1327C>T (p.R443C), c.151C>T (p.P51S), c.296T>C (p.L99P), c.443T>G (p.L148R), c.502T>A (p.F168I), c.523G>C (p.D175H), c.536C>T (p.P179L), c.728C>G (p.P243R), c.852C>A (p.F284L), c.861C>A (p.N287K), c.970T>C (p.Y324H), c.1384T>C (p.Y462H), c.1406G>C (p.R469P), c.111G>A (p.W37X) Sequencing | NM_001360:3-9

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

Stargardt Disease (ABCA4): Mutations (16): ♂ Genotyping | c.3083C>T (p.A1028V), c.52C>T (p.R18W), c.5338C>G (p.P1780A), c.1018T>G (p.Y340D), c.2461T>A (p.W821R), c.2565G>A (p.W855X), c.3106G>A (p.E1036K), c.3210_3211insGT (p.S1071Vfs), c.634C>T (p.R212C), c.3113C>T (p.A1038V), c.1622T>C (p.L541P), c.3364G>A (p.E1122K), c.6079C>T (p.L2027F), c.2588G>C (p.G863A), c.1938-1G>A, c.571-2A>G Sequencing | NM_000350:1-50

Stuve-Wiedemann Syndrome (LIFR): Mutations (9): ♂ Genotyping | c.2472_2476delTATGT, c.2434C>T (p.R812X), c.2274_2275insT, c.1789C>T (pR597X), c.1601-2A>G, c.1620_1621insA, c.756_757insT (p.K253X), c.653_654insT, c.170delC Sequencing | NM_002310:2-20

Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2): Mutations (7): ♂ Genotyping | c.1018_1020delGTT (p.340delV), c.-26+2T>C, c.532C>T (p.R178X), c.835C>T (p.R279W), c.1957T>A (p.C653S), c.398C>T (p.A133V), c.764G>A (p.G255E) Sequencing | NM_000112:1-3

Tay-Sachs Disease (HEXA): Mutations (78): ♂ Genotyping | c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.532C>T (p.R178C), c.533G>A (p.R178H), c.805G>A (p.G269S), c.1510C>T (p.R504C), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.1003A>T (p.I335F), c.910_912delTTC (p.305delF), c.749G>A (p.G250D), c.632T>C (p.F211S),

c.629C>T (p.S210F), c.613delC, c.611A>G (p.H204R), c.598G>A (p.V200M), c.590A>C (p.K197T), c.571-1G>T, c.540C>G (p.Y180X), c.538T>C (p.Y180H), c.533G>T (p.R178L), c.508C>T (p.R170W), c.409C>T (p.R137X), c.380T>G (p.L127R), c.346+1G>C, c.116T>G (p.L39R), c.78G>A (p.W26X), c.1A>G (p.M1V), c.1495C>T (p.R499C), c.459+5G>A (IVS4+5G>A), c.1422-2A>G, c.535C>T (p.H179Y), c.1141delG (p.V381fs), c.796T>G (p.W266G), c.155C>A (p.S52X), c.426delT (p.F142fs), c.413-2A>G, c.570+3A>G, c.536A>G (p.H179R), c.1146+1G>A, c.736G>A (p.A246T), c.1302C>G (p.F434L), c.778C>T (p.P260S), c.1008G>T (p.Q336H), c.1385A>T (p.E462V), c.964G>A (p.D322N), c.340G>A (p.E114K), c.1432G>A (p.G478R), c.1178G>C (p.R393P), c.805+1G>C, c.1426A>T (p.R476X), c.623A>T (p.D208V), c.1537C>T (p.Q513X), c.1511G>T (p.R504L), c.1307_1308delTA (p.I436fs), c.571-8A>G, c.624_627delTCCT (p.D208fs), c.1211_1212delTG (p.L404fs), c.621T>G (p.D207E), c.1511G>A (p.R504H), c.1177C>T (p.R393X), c.2T>C (p.M1T), c.1292G>A (p.W431X), c.947_948insA (p.Y316fs), c.607T>G (p.W203G), c.1061_1063delTCT (p.F354_Y355delinsX), c.615delG (p.L205fs), c.805+2T>C, c.1123delG (p.E375fs), c.1121A>G (p.Q374R), c.1043_1046delTCAA (p.F348fs), c.1510delC (p.R504fs), c.1451T>C (p.L484P), c.964G>T (p.D322Y), c.1351C>G (p.L451V), c.571-2A>G (IVS5-2A>G) Sequencing | NM_000520:1-14

Trichohepatoenteric Syndrome: Type 1 (TTC37): Mutations (9): ♂ Genotyping | c.3847G>A (p.D1283N), c.751G>A (p.G251R), c.2251C>T (p.Q751X), c.439C>T (p.Q147X), c.2808G>A (p.W936X), c.2515+1G>C, c.4620+1G>C, c.1632+1delG, c.2578-7delTTTTT Sequencing | NM_014639:4-43

Tyrosine Hydroxylase Deficiency (TH): Mutations (1): ♂ Genotyping | c.698G>A (p.R233H) Sequencing | NM_199292:1-14

Tyrosinemia: Type I (FAH): Mutations (10): ♂ Genotyping | c.1062+5G>A, c.554-1G>T, c.607-6T>G, c.707-1G>C, c.782C>T (p.P261L), c.1069G>T (p.E357X), c.786G>A (p.W262X), c.698A>T (p.D233V), c.1009G>A (p.G337S), c.192G>T (p.Q64H) Sequencing | NM_000137:1-14

Tyrosinemia: Type II (TAT): Mutations (5): ♂ Genotyping | c.169C>T (p.R57X), c.668C>G (p.S223X), c.1249C>T (p.R417X), c.1085G>T (p.G362V), c.236-5A>G Sequencing | NM_000353:2-12

Usher Syndrome: Type 1B (MYO7A): Mutations (13): ♂ Genotyping | c.93C>A (p.C31X), c.448C>T (p.R150X), c.634C>T (p.R212C), c.635G>A (p.R212H), c.700C>T (p.Q234X), c.1797G>A (p.M599I), c.1996C>T (p.R666X), c.2476G>A (p.A826T), c.3719G>A (p.R1240Q), c.5581C>T (p.R1861X), c.6025delG (p.A2009fs), c.640G>A (p.G214R), c.1190C>A (p.A397D) Sequencing | NM_000260:2-49

Usher Syndrome: Type 1C (USH1C): Mutations (5): ♂ Genotyping | c.496+1G>A, c.238_239insC, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.36+1G>T Sequencing | NM_153676:1-27

Usher Syndrome: Type 1D (CDH23): Mutations (14): ♂ Genotyping | c.172C>T (p.Q58X), c.3367C>T (p.Q1123X), c.3617C>G (p.P1206R), c.3713_3714delCT (p.S1238fs), c.3880C>T (p.Q1294X), c.4069C>T (p.Q1357X), c.4488G>C (p.Q1496H), c.4504C>T (p.R1502X), c.5237G>A (p.R1746Q), c.5985C>A (p.Y1995X), c.6307G>T (p.E2103X), c.7549A>G (p.S2517G), c.8230G>A (p.G2744S), c.8497C>G (p.R2833G) Sequencing | NM_022124:2-68

Usher Syndrome: Type 1F (PCDH15): Mutations (7): ♂ Genotyping | c.733C>T (p.R245X), c.2067C>A (p.Y684X), c.7C>T (p.R3X), c.1942C>T (p.R648X), c.1101delT (p.A367fsX), c.2800C>T (p.R934X), c.4272delA (p.L1425fs) Sequencing | NM_001142763:2-35

Usher Syndrome: Type 2A (USH2A): Mutations (22): ♂ Genotyping | c.14020A>G (p.R4674G), c.12067-2A>G, c.4338_4339delCT (p.C1447fs), c.2299delG (p.E767SfsX21), c.2209C>T (p.R737X), c.1256G>T (p.C419F), c.1000C>T (p.R334W), c.923_924insGCCA (p.H308fs), c.12708T>A (p.C4236X), c.13576C>T (p.R4526X), c.1840+1G>A, c.11328T>G (p.Y3776X), c.5329C>T (p.R1777W), c.9165_9168delCTAT (p.I3055MfsX2), c.9469C>T (p.Q3157X), c.1876C>T (p.R626X), c.7123delG (p.G2375fs), c.9492_9498delTGATGAG (p.D3165fs), c.6235A>T (p.K2079X), c.14403C>G (p.Y4801X), c.3788G>A (p.W1263X), c.11328T>A (p.Y3776X) Sequencing | NM_206933:2-72

Usher Syndrome: Type 3 (CLRN1): Mutations (5): ♂ Genotyping | c.144T>G (p.N48K), c.131T>A (p.M120K), c.567T>G (p.Y189X), c.634C>T (p.Q212X), c.221T>C (p.L74P) Sequencing | NM_001195794:1-4

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL): Mutations (29): ♂ Genotyping | c.779C>T (p.T260M), c.848T>C (p.V283A), c.1144A>C (p.K382Q), c.1226C>T (p.T409M), c.1322G>A (p.G441D), c.1372T>C (p.F458L), c.1405C>T (p.R469W), c.1837C>T (p.R613W), c.553G>A (p.G185S), c.739A>C (p.K247Q), c.37C>T (p.Q13X), c.265C>T (p.P89S), c.272C>A (p.P91Q), c.364A>G (p.N122D), c.388_391delGAGA (p.E130fs), c.520G>A (p.V174M), c.856A>G (p.R286G), c.1606_1609delGCAG (p.A536fs), c.1531C>T (p.R511W), c.1512G>T (p.E504D), c.664G>A (p.G222R), c.685C>T (p.R229X), c.577G>C (p.G193R), c.881G>A (p.G294E), c.753-2A>C (IVS8-2A>C), c.1349G>A (p.R450H), c.1358G>A (p.R453Q), c.790A>G (p.K264E), c.1246G>A (p.A416T) Sequencing | NM_000018:1-20

Walker-Warburg Syndrome (FKTN): Mutations (5): ♂ Genotyping | c.1167insA (p.F390fs), c.139C>T (p.R47X), c.748T>G (p.C250G), c.648-1243G>T (IVS5-1243G>T), c.515A>G (p.H172R) Sequencing | NM_006731:2-10

Werner Syndrome (WRN): Mutations (8): ♂ Genotyping | c.3139-1G>C (IVS25-1G>C), c.3913C>T (p.R1305X), c.3493C>T (p.Q1165X), c.1730A>T (p.K577M), c.1336C>T (p.R368X), c.3686A>T (p.Q1229L), c.3915_3916insA (p.R1306fs), c.2089-3024A>G Sequencing | NM_000553:2-35

Wilson Disease (ATP7B): Mutations (17): ♂ Genotyping | c.1340_1343delAAAC, c.2304delC (p.M769Cfs), c.2332C>G (p.R778G), c.3207C>A (p.H1069Q), c.2333G>T (p.R778L), c.2336G>A (p.W779X), c.2337G>A (p.W779X), c.2906G>A (p.R969Q), c.1934T>G (p.M645R), c.2123T>C (p.L708P), c.-370_-394delTGGCCGAGACCGCGG, c.3191A>C (p.E1064A), c.845delT (p.L282Pfs), c.3817C>T (p.P1273S), c.3683G>C (p.R1228T), c.3809A>G (p.N1270S), c.2293G>A (p.D765N) Sequencing | NM_000053:1-21

Wolcott-Rallison Syndrome (EIF2AK3): Mutations (5): ♂ Genotyping | c.1409C>G (p.S470X), c.1262delA (p.N421fs), c.1570delGAAA (p.E524fsX), c.478delG (p.A160fs), c.1047_1060delAGTCATTCCCATCA (p.V350Sfs) Sequencing | NM_004836:1-17

Wolman Disease (LIPA): Mutations (3): ♂ Genotyping | c.964C>T (p.Q322X), c.419G>A (p.W140X), c.260G>T (p.G87V) Sequencing | NM_001127605:2-10

Xeroderma Pigmentosum: Group A (XPA): Mutations (7): ♂ Genotyping | c.172+2T>G, c.323G>T (p.C108F), c.374delC (p.T125fs), c.682C>T (p.R228X), c.619C>T (p.R207X), c.348T>A (p.Y116X), c.390-1G>C Sequencing | NM_000380:1-6

Xeroderma Pigmentosum: Group C (XPC): Mutations (5): ♂ Genotyping | c.1735C>T (p.R579X), c.566_567delAT (p.Y189fs), c.413-9T>A, c.413-24A>G, c.1643_1644delTG (p.V548fs) Sequencing | NM_004628:1-16

Zellweger Spectrum Disorders: PEX1 Related (PEX1): Mutations (3): ♂ Genotyping | c.2528G>A (p.G843D), c.2916delA (p.G973fs), c.2097insT (p.I700fs) Sequencing | NM_000466:1-24

Zellweger Spectrum Disorders: PEX10 Related (PEX10): Mutations (2): ♂ Genotyping | c.764_765insA, c.874_875delCT Sequencing | NM_153818:2-6

Zellweger Spectrum Disorders: PEX2 Related (PEX2): Mutations (1): ♂ Genotyping | c.355C>T (p.R119X) Sequencing | NM_001172087:1-3

Zellweger Spectrum Disorders: PEX6 Related (PEX6): Mutations (8): ♂ Genotyping | c.1130+1G>A (IVS3+1G>A), c.1688+1G>A (IVS7+1G>A), c.1962-1G>A (p.L655fsX3), c.1301delC (p.S434Ffs), c.1601T>C (p.L534P), c.511insT (p.G171Wfs), c.802_815delGACGGACTGGCGCT (p.D268Cfs), c.1715C>T (p.T572I) Sequencing | NM_000287:1-17

Residual Risk Information

Detection rates are calculated from the primary literature and may not be available for all ethnic populations. The values listed below are for genotyping. Sequencing provides higher detection rates and lower residual risks for each disease. More precise values for sequencing may become available in the future.

Disease	Carrier Rate	Detection Rate	Residual Risk
11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	♂ Moroccan Jewish: 1/39	91.67%	1/468
17-Alpha-Hydroxylase Deficiency	♂ Brazilian: Unknown	54.55%	Unknown
	♂ Japanese: Unknown	45.45%	Unknown
17-Beta-Hydroxysteroid Dehydrogenase Deficiency	♂ Arab: 1/8	>99%	<1/800
	♂ Dutch: 1/192	13.89%	1/223
21-Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia	♂ European: 1/62	27.65%	1/86
	♂ General: 1/62	29.34%	1/88
21-Hydroxylase-Deficient Nonclassical Congenital Adrenal Hyperplasia	♂ Argentinian: 1/4	<10%	1/4
	♂ European: 1/16	<10%	1/16
3-Beta-Hydroxysteroid Dehydrogenase Deficiency	♂ General: Unknown	16.13%	Unknown
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related	♂ European: 1/146	26.32%	1/198
	♂ General: 1/112	37.50%	1/179
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related	♂ General: 1/112	35.29%	1/173
	♂ Japanese: 1/112	33.33%	1/168
	♂ Korean: 1/141	66.67%	1/423
	♂ Turkish: 1/112	24.07%	1/148
3-Methylglutaconic Aciduria: Type 3	♂ Iraqi Jewish: 1/10	>99%	<1/1,000
3-Phosphoglycerate Dehydrogenase Deficiency	♂ Ashkenazi Jewish: 1/400	>99%	<1/40,000
5-Alpha Reductase Deficiency	♂ Dominican: Unknown	>99%	Unknown
	♂ Mexican: Unknown	68.75%	Unknown
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	♂ Chinese: 1/183	78.95%	1/869
	♂ East Asian: 1/180	64.20%	1/503
ARSACS	♂ French Canadian: 1/22	95.45%	1/484
Abetalipoproteinemia	♂ Ashkenazi Jewish: 1/131	>99%	<1/13,100
Acrodermatitis Enteropathica	♂ Arab: Unknown	40.00%	Unknown
	♂ Egyptian: Unknown	33.33%	Unknown
	♂ French: Unknown	27.78%	Unknown
	♂ Tunisian: Unknown	77.78%	Unknown
Acute Infantile Liver Failure: TRMU Related	♂ Yemenite Jewish: 1/40	71.43%	1/140
Acyl-CoA Oxidase I Deficiency	♂ General: Unknown	35.00%	Unknown
	♂ Japanese: Unknown	42.86%	Unknown
Adenosine Deaminase Deficiency	♂ General: 1/388	36.96%	1/615
Alkaptonuria	♂ Dominican: Unknown	>99%	Unknown
	♂ Finnish: 1/251	60.00%	1/628
	♂ Slovak: 1/69	59.38%	1/170
Alpha Thalassemia	♂ General: 1/48	50.67%	1/97
Alpha-1-Antitrypsin Deficiency	♂ European: 1/35	95.00%	1/700
	♂ General: Unknown	95.00%	Unknown
Alpha-Mannosidosis	♂ European: 1/354	30.23%	1/507
	♂ General: 1/354	35.19%	1/546
Alport Syndrome: COL4A3 Related	♂ Dutch: 1/409	22.73%	1/529
Alport Syndrome: COL4A4 Related	♂ General: 1/409	23.33%	1/533
Amegakaryocytic Thrombocytopenia	♂ Ashkenazi Jewish: 1/76	>99%	<1/7,600
	♂ General: Unknown	64.81%	Unknown
Andermann Syndrome	♂ French Canadian: 1/24	99.38%	1/3,888
Antley-Bixler Syndrome	♂ General: Unknown	45.65%	Unknown
	♂ Japanese: Unknown	60.47%	Unknown
Argininemia	♂ Chinese: Unknown	40.00%	Unknown
	♂ French Canadian: Unknown	75.00%	Unknown
	♂ Japanese: Unknown	>99%	Unknown
Argininosuccinate Lyase Deficiency	♂ European: 1/133	57.41%	1/312
	♂ Saudi Arabian: 1/80	51.72%	1/166
Aromatase Deficiency	♂ General: Unknown	25.00%	Unknown
Arthrogryposis, Mental Retardation, & Seizures	♂ Ashkenazi Jewish: 1/205	>99%	<1/20,500
Asparagine Synthetase Deficiency	♂ Iranian Jewish: 1/80	>99%	<1/8,000
Aspartylglycosaminuria	♂ Finnish: 1/69	96.12%	1/1,780
Ataxia with Vitamin E Deficiency	♂ European: 1/274	80.00%	1/1,370
	♂ Italian: 1/224	97.73%	1/9,856
	♂ North African: 1/159	>99%	<1/15,900
Ataxia-Telangiectasia	♂ Costa Rican: 1/100	68.52%	1/318
	♂ North African Jewish: 1/81	96.97%	1/2,673
	♂ Norwegian: 1/197	50.00%	1/394
	♂ Sardinians: Unknown	85.71%	Unknown
	♂ US Amish: Unknown	>99%	Unknown
Autosomal Recessive Polycystic Kidney Disease	♂ Finnish: 1/45	84.21%	1/285
	♂ French: 1/71	62.50%	1/189
	♂ General: 1/71	37.11%	1/113
Bardet-Biedl Syndrome: BBS1 Related	♂ General: 1/376	70.27%	1/1,265
	♂ Northern European: 1/376	85.90%	1/2,666
	♂ Puerto Rican: Unknown	90.00%	Unknown
Bardet-Biedl Syndrome: BBS10 Related	♂ General: 1/404	47.79%	1/774
Bardet-Biedl Syndrome: BBS11 Related	♂ Bedouin: 1/59	>99%	<1/5,900
Bardet-Biedl Syndrome: BBS12 Related	♂ General: Unknown	50.00%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Bardet-Biedl Syndrome: BBS2 Related	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: 1/638	38.46%	1/1,037
	♂ Middle Eastern: Unknown	>99%	Unknown
Bare Lymphocyte Syndrome: Type II	♂ General: Unknown	66.67%	Unknown
Bartter Syndrome: Type 4A	♂ General: 1/457	81.82%	1/2,514
Beta Thalassemia	♂ African American: 1/75	84.21%	1/475
	♂ Indian: 1/24	74.12%	1/93
	♂ Sardinians: 1/23	97.14%	1/804
	♂ Spaniard: 1/51	93.10%	1/739
Beta-Hexosaminidase Pseudodeficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: Unknown	>99%	Unknown
Beta-Ketothiolase Deficiency	♂ Japanese: Unknown	58.33%	Unknown
	♂ Spaniard: Unknown	90.00%	Unknown
Biotinidase Deficiency	♂ General: 1/123	78.32%	1/567
Bloom Syndrome	♂ Ashkenazi Jewish: 1/134	96.67%	1/4,020
	♂ European: Unknown	66.22%	Unknown
	♂ Japanese: Unknown	50.00%	Unknown
Canavan Disease	♂ Ashkenazi Jewish: 1/55	98.86%	1/4,840
	♂ European: Unknown	53.23%	Unknown
Carnitine Palmitoyltransferase IA Deficiency	♂ General: Unknown	38.89%	Unknown
	♂ Hutterite: 1/16	>99%	<1/1,600
	♂ Japanese: 1/101	66.67%	1/303
Carnitine Palmitoyltransferase II Deficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: Unknown	71.43%	Unknown
Carnitine-Acylcarnitine Translocase Deficiency	♂ Asian: Unknown	95.45%	Unknown
	♂ General: Unknown	18.75%	Unknown
Carpenter Syndrome	♂ Brazilian: Unknown	40.00%	Unknown
	♂ Northern European: Unknown	85.00%	Unknown
Cartilage-Hair Hypoplasia	♂ Finnish: 1/76	93.33%	1/1,140
	♂ US Amish: 1/19	>99%	<1/1,900
Cerebrotendinous Xanthomatosis	♂ Dutch: Unknown	78.57%	Unknown
	♂ Italian: Unknown	45.95%	Unknown
	♂ Japanese: Unknown	92.86%	Unknown
	♂ Moroccan Jewish: 1/6	87.50%	1/48
Chediak-Higashi Syndrome	♂ General: Unknown	19.64%	Unknown
Cholesteryl Ester Storage Disease	♂ General: 1/101	68.97%	1/325
Choreoacanthocytosis	♂ Ashkenazi Jewish: Unknown	66.67%	Unknown
Chronic Granulomatous Disease: CYBA Related	♂ Iranian: Unknown	71.43%	Unknown
	♂ Japanese: 1/274	>99%	<1/27,400
	♂ Korean: 1/105	>99%	<1/10,500
	♂ Moroccan Jewish: 1/234	>99%	<1/23,400
Citrin Deficiency	♂ Japanese: 1/70	>99%	<1/7,000
Citrullinemia: Type I	♂ European: 1/120	18.18%	1/147
	♂ General: 1/120	52.27%	1/251
	♂ Japanese: Unknown	64.71%	Unknown
	♂ Mediterranean: 1/120	50.00%	1/240
Classical Galactosemia	♂ African American: 1/78	73.13%	1/290
	♂ Ashkenazi Jewish: 1/127	>99%	<1/12,700
	♂ Dutch: 1/91	75.47%	1/371
	♂ European: 1/112	88.33%	1/960
	♂ General: 1/125	80.00%	1/625
	♂ Irish: 1/76	91.30%	1/874
	♂ Irish Travellers: 1/14	>99%	<1/1,400
Cockayne Syndrome: Type A	♂ Christian Arab: Unknown	50.00%	Unknown
Cockayne Syndrome: Type B	♂ General: 1/378	19.30%	1/468
Cohen Syndrome	♂ European: Unknown	19.05%	Unknown
	♂ Finnish: 1/140	67.24%	1/427
	♂ US Amish: 1/12	>99%	<1/1,200
Combined Pituitary Hormone Deficiency: PROP1 Related	♂ European: 1/45	93.29%	1/671
	♂ General: 1/45	82.35%	1/255
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related	♂ Danish: 1/71	90.00%	1/710
	♂ Dutch: 1/68	39.29%	1/112
	♂ European: 1/71	55.33%	1/159
Congenital Disorder of Glycosylation: Type 1B: MPI Related	♂ French: Unknown	54.17%	Unknown
Congenital Disorder of Glycosylation: Type 1C: ALG6 Related	♂ French: Unknown	59.09%	Unknown
	♂ General: Unknown	86.21%	Unknown
Congenital Ichthyosis: ABCA12 Related	♂ North African: Unknown	>99%	Unknown
	♂ South Asian: Unknown	66.67%	Unknown
Congenital Insensitivity to Pain with Anhidrosis	♂ Japanese: Unknown	56.52%	Unknown
	♂ Moroccan Jewish: Unknown	>99%	Unknown
Congenital Lipoid Adrenal Hyperplasia	♂ Japanese: 1/201	51.11%	1/411
	♂ Korean: 1/251	63.64%	1/690
Congenital Myasthenic Syndrome: CHRNE Related	♂ European Gypsy: 1/26	>99%	<1/2,600
	♂ North African: Unknown	60.87%	Unknown
Congenital Myasthenic Syndrome: DOK7 Related	♂ European: 1/472	19.05%	1/583
	♂ General: 1/472	18.75%	1/581
Congenital Myasthenic Syndrome: RAPSN Related	♂ General: 1/437	88.57%	1/3,824
	♂ Non-Ashkenazi Jewish: Unknown	>99%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Congenital Neutropenia: Recessive	♂ English: Unknown	11.76%	Unknown
	♂ Japanese: Unknown	22.22%	Unknown
	♂ Turkish: Unknown	89.47%	Unknown
Corneal Dystrophy and Perceptive Deafness	♂ General: Unknown	71.43%	Unknown
Corticosterone Methyloxidase Deficiency	♂ Iranian Jewish: 1/32	>99%	<1/3,200
Crigler-Najjar Syndrome	♂ Sardinians: Unknown	80.00%	Unknown
	♂ Tunisian: Unknown	>99%	Unknown
Cystic Fibrosis	♂ African American: 1/62	69.99%	1/207
	♂ Ashkenazi Jewish: 1/23	96.81%	1/721
	♂ Asian: 1/94	65.42%	1/272
	♂ European: 1/25	94.96%	1/496
	♂ Hispanic American: 1/48	77.32%	1/212
	♂ Native American: 1/53	84.34%	1/338
Cystinosis	♂ Dutch: 1/194	73.08%	1/721
	♂ French Canadian: 1/40	75.00%	1/160
	♂ General: 1/194	54.51%	1/426
Cystinuria: Non-Type I	♂ European: 1/42	61.11%	1/108
	♂ General: 1/42	37.50%	1/67
	♂ Libyan Jewish: 1/26	93.48%	1/399
	♂ United States: 1/42	56.25%	1/96
Cystinuria: Type I	♂ European: 1/42	46.67%	1/79
	♂ Swedish: 1/159	55.88%	1/360
D-Bifunctional Protein Deficiency	♂ General: 1/159	38.64%	1/259
Diabetes: Recessive Permanent Neonatal	♂ General: Unknown	25.00%	Unknown
Du Pan Syndrome	♂ Pakistani: Unknown	>99%	Unknown
Dyskeratosis Congenita: RTEL1 Related	♂ Ashkenazi Jewish: 1/203	>99%	<1/20,300
	♂ General: 1/501	50.00%	1/1,002
Dystrophic Epidermolysis Bullosa: Recessive	♂ Italian: Unknown	45.00%	Unknown
	♂ Mexican American: 1/345	56.25%	1/789
Ehlers-Danlos Syndrome: Type VIIC	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
Ellis-van Creveld Syndrome: EVC Related	♂ General: 1/123	32.14%	1/181
Ellis-van Creveld Syndrome: EVC2 Related	♂ General: Unknown	<10%	Unknown
Enhanced S-Cone	♂ Ashkenazi Jewish: Unknown	90.48%	Unknown
	♂ General: Unknown	52.50%	Unknown
Ethylmalonic Aciduria	♂ Arab/Mediterranean: Unknown	29.17%	Unknown
	♂ General: Unknown	38.24%	Unknown
Familial Chloride Diarrhea	♂ Finnish: 1/51	>99%	<1/5,100
	♂ Kuwaiti: 1/38	90.00%	1/380
	♂ Polish: 1/224	45.24%	1/409
	♂ Saudi Arabian: 1/38	>99%	<1/3,800
Familial Dysautonomia	♂ Ashkenazi Jewish: 1/31	>99%	<1/3,100
Familial Hyperinsulinism: Type 1: ABCC8 Related	♂ Ashkenazi Jewish: 1/52	98.75%	1/4,160
	♂ Finnish: 1/101	45.16%	1/184
Familial Hyperinsulinism: Type 2: KCNJ11 Related	♂ Arab: Unknown	40.00%	Unknown
Familial Mediterranean Fever	♂ Arab: 1/4	51.27%	1/8
	♂ Armenian: 1/5	94.51%	1/91
	♂ Ashkenazi Jewish: 1/81	40.95%	1/137
	♂ Iraqi Jewish: 1/4	76.92%	1/17
	♂ Israeli Jewish: 1/5	62.67%	1/13
	♂ Lebanese: 1/6	91.67%	1/72
	♂ North African Jewish: 1/5	95.69%	1/116
	♂ Syrian: 1/6	85.14%	1/40
	♂ Turkish: 1/5	74.43%	1/20
Fanconi Anemia: Type A	♂ Moroccan Jewish: 1/100	>99%	<1/10,000
	♂ Spanish Gypsy: 1/67	>99%	<1/6,700
Fanconi Anemia: Type C	♂ Ashkenazi Jewish: 1/101	>99%	<1/10,100
	♂ General: Unknown	30.00%	Unknown
Fanconi Anemia: Type G	♂ Black South African: 1/101	81.82%	1/556
	♂ French Canadian: Unknown	87.50%	Unknown
	♂ Japanese: Unknown	75.00%	Unknown
	♂ Korean: Unknown	66.67%	Unknown
Fanconi Anemia: Type J	♂ General: Unknown	86.36%	Unknown
Fumarase Deficiency	♂ General: Unknown	30.00%	Unknown
GM1-Gangliosidoses	♂ Eurodescent Brazilian: 1/66	62.15%	1/174
	♂ European: 1/194	50.00%	1/388
	♂ General: 1/194	20.00%	1/243
	♂ Hispanic American: 1/194	58.33%	1/466
	♂ Japanese: Unknown	62.82%	Unknown
GRACILE Syndrome	♂ Finnish: 1/109	97.22%	1/3,924
Galactokinase Deficiency	♂ Japanese: 1/501	50.00%	1/1,002
	♂ Roma: 1/51	>99%	<1/5,100
Gaucher Disease	♂ Ashkenazi Jewish: 1/15	87.16%	1/117
	♂ General: 1/112	31.60%	1/164
	♂ Spaniard: Unknown	44.29%	Unknown
	♂ Turkish: 1/236	59.38%	1/581
Gitelman Syndrome	♂ European: 1/100	35.00%	1/154
	♂ European Gypsy: Unknown	>99%	Unknown
	♂ General: 1/101	30.00%	1/144
	♂ Taiwanese: Unknown	64.29%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Globoid Cell Leukodystrophy	♂ Dutch: 1/137	60.98%	1/351
	♂ European: 1/150	26.47%	1/204
	♂ Japanese: 1/150	36.00%	1/234
Glutaric Acidemia: Type I	♂ European: 1/164	57.78%	1/388
	♂ General: 1/164	25.51%	1/220
	♂ US Amish: 1/12	>99%	<1/1,200
Glutaric Acidemia: Type IIA	♂ General: Unknown	71.43%	Unknown
Glutaric Acidemia: Type IIB	♂ General: Unknown	33.33%	Unknown
Glutaric Acidemia: Type IIC	♂ Taiwanese: Unknown	>99%	Unknown
	♂ Turkish: Unknown	80.00%	Unknown
Glycine Encephalopathy: AMT Related	♂ General: Unknown	40.91%	Unknown
Glycine Encephalopathy: GLDC Related	♂ Finnish: 1/118	78.00%	1/536
	♂ General: 1/280	12.50%	1/320
Glycogen Storage Disease: Type IA	♂ Ashkenazi Jewish: 1/71	>99%	<1/7,100
	♂ Chinese: 1/159	80.00%	1/795
	♂ European: 1/177	76.88%	1/765
	♂ Hispanic American: 1/177	27.78%	1/245
	♂ Japanese: 1/177	89.22%	1/1,641
Glycogen Storage Disease: Type IB	♂ Australian: 1/354	50.00%	1/708
	♂ European: 1/354	45.74%	1/652
	♂ Japanese: 1/354	39.13%	1/582
Glycogen Storage Disease: Type II	♂ African American: 1/60	45.83%	1/111
	♂ Chinese: 1/112	72.00%	1/400
	♂ European: 1/97	51.76%	1/201
	♂ North African: Unknown	60.00%	Unknown
Glycogen Storage Disease: Type III	♂ Faroese: 1/30	>99%	<1/3,000
	♂ General: 1/159	39.81%	1/264
	♂ North African Jewish: 1/35	>99%	<1/3,500
Glycogen Storage Disease: Type IV	♂ Ashkenazi Jewish: 1/35	>99%	<1/3,500
	♂ General: 1/461	18.60%	1/566
Glycogen Storage Disease: Type V	♂ Caucasus Jewish: Unknown	>99%	Unknown
	♂ European: 1/159	60.71%	1/405
	♂ General: Unknown	74.10%	Unknown
	♂ Spaniard: 1/159	67.11%	1/483
	♂ Yemenite Jewish: Unknown	75.00%	Unknown
Glycogen Storage Disease: Type VII	♂ Ashkenazi Jewish: 1/250	>99%	<1/25,000
Guanidinoacetate Methyltransferase Deficiency	♂ General: Unknown	29.41%	Unknown
HMG-CoA Lyase Deficiency	♂ General: 1/159	40.00%	1/265
	♂ Japanese: Unknown	30.00%	Unknown
	♂ Portuguese: Unknown	86.36%	Unknown
	♂ Saudi Arabian: Unknown	93.33%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Hemochromatosis: Type 2A: HFE2 Related	♂ European: Unknown	69.23%	Unknown
	♂ Mediterranean: Unknown	72.73%	Unknown
Hemochromatosis: Type 3: TFR2 Related	♂ Italian: Unknown	73.21%	Unknown
Hemoglobinopathy: Hb C	♂ African American: 1/51	>99%	<1/5,100
Hemoglobinopathy: Hb D	♂ Canadian: 1/64	>99%	<1/6,400
	♂ Indian: 1/16	>99%	<1/1,600
	♂ Iranian: 1/11	>99%	<1/1,100
Hemoglobinopathy: Hb E	♂ Cambodia: 1/4	>99%	<1/400
	♂ Chinese: 1/13	>99%	<1/1,300
	♂ Indian: 1/10	>99%	<1/1,000
	♂ Thai: 1/9	>99%	<1/900
Hemoglobinopathy: Hb O	♂ African American: 1/87	>99%	<1/8,700
	♂ Middle Eastern: Unknown	>99%	Unknown
Hereditary Fructose Intolerance	♂ European: 1/81	72.73%	1/297
	♂ Italian: 1/81	90.91%	1/891
	♂ Slavic: 1/81	>99%	<1/8,100
Hereditary Spastic Paraplegia: TECPR2 Related	♂ Bukharan Jewish: 1/75	>99%	<1/7,500
Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related	♂ Pakistani: Unknown	>99%	Unknown
Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	♂ European: Unknown	70.00%	Unknown
	♂ General: 1/781	52.27%	1/1,636
Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related	♂ Italian: Unknown	28.57%	Unknown
Hermansky-Pudlak Syndrome: Type 1	♂ Puerto Rican: 1/22	94.95%	1/436
Hermansky-Pudlak Syndrome: Type 3	♂ Ashkenazi Jewish: 1/235	>99%	<1/23,500
	♂ European: 1/434	12.50%	1/496
Hermansky-Pudlak Syndrome: Type 4	♂ European: Unknown	54.17%	Unknown
Holocarboxylase Synthetase Deficiency	♂ European: 1/148	83.33%	1/888
	♂ Japanese: 1/159	76.92%	1/689
Homocystinuria Caused by CBS Deficiency	♂ European: 1/224	64.29%	1/627
	♂ Irish: 1/128	70.59%	1/435
	♂ Italian: 1/224	35.71%	1/348
	♂ Norwegian: 1/41	84.38%	1/262
	♂ Qatari: 1/22	>99%	<1/2,200
	♂ Saudi Arabian: Unknown	92.31%	Unknown
Hurler Syndrome	♂ Czech: 1/190	52.50%	1/400
	♂ European: 1/194	81.71%	1/1,061
	♂ General: 1/194	62.50%	1/517
	♂ Italian: 1/194	61.11%	1/499
	♂ Japanese: 1/194	23.68%	1/254
	♂ Moroccan Jewish: 1/194	92.31%	1/2,522
	♂ Scandinavian: 1/194	79.41%	1/942

Disease	Carrier Rate	Detection Rate	Residual Risk
	♂ Spaniard: 1/194	52.50%	1/408
Hypophosphatasia	♂ Canadian Amish: 1/26	>99%	<1/2,600
	♂ European: 1/159	19.23%	1/197
	♂ Japanese: Unknown	54.55%	Unknown
Inclusion Body Myopathy: Type 2	♂ General: Unknown	85.83%	Unknown
	♂ Iranian Jewish: 1/16	>99%	<1/1,600
	♂ Japanese: Unknown	71.88%	Unknown
	♂ Korean: Unknown	72.50%	Unknown
Infantile Cerebral and Cerebellar Atrophy	♂ Caucasus Jewish: 1/20	>99%	<1/2,000
Isolated Microphthalmia: VSX2 Related	♂ Middle Eastern: Unknown	71.43%	Unknown
Isovaleric Acidemia	♂ General: 1/251	47.37%	1/477
Joubert Syndrome	♂ Ashkenazi Jewish: 1/92	>99%	<1/9,200
Lamellar Ichthyosis: Type 1	♂ Norwegian: 1/151	81.40%	1/812
Laryngoonychocutaneous Syndrome	♂ Pakistani: Unknown	>99%	Unknown
Leber Congenital Amaurosis: CEP290 Related	♂ European: 1/251	47.32%	1/476
Leber Congenital Amaurosis: GUCY2D Related	♂ Finnish: Unknown	>99%	Unknown
Leber Congenital Amaurosis: LCA5 Related	♂ Pakistani: Unknown	83.33%	Unknown
Leber Congenital Amaurosis: RDH12 Related	♂ General: 1/560	38.37%	1/909
Leigh Syndrome: French-Canadian	♂ French Canadian: 1/23	95.45%	1/506
Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related	♂ Cree: Unknown	>99%	Unknown
	♂ European: Unknown	65.22%	Unknown
Leydig Cell Hypoplasia (Luteinizing Hormone Resistance)	♂ Brazilian: Unknown	>99%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2A	♂ Basque: 1/61	61.46%	1/158
	♂ Croatian: 1/133	76.00%	1/554
	♂ European: 1/103	17.23%	1/124
	♂ General: 1/103	26.47%	1/140
	♂ Italian: 1/162	35.71%	1/252
	♂ Russian: 1/103	53.33%	1/221
	♂ US Amish: Unknown	>99%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2B	♂ Caucasus Jewish: 1/25	>99%	<1/2,500
	♂ Libyan Jewish: 1/19	>99%	<1/1,900
Limb-Girdle Muscular Dystrophy: Type 2C	♂ European Gypsy: 1/50	>99%	<1/5,000
	♂ General: Unknown	60.00%	Unknown
	♂ Tunisian: Unknown	>99%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2D	♂ Brazilian: Unknown	64.29%	Unknown
	♂ European: 1/288	22.22%	1/370
	♂ Finnish: 1/150	95.45%	1/3,300
	♂ General: Unknown	26.09%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Limb-Girdle Muscular Dystrophy: Type 2E	♂ Brazilian: Unknown	57.14%	Unknown
	♂ European: 1/539	25.00%	1/719
	♂ General: Unknown	12.50%	Unknown
	♂ US Amish: Unknown	>99%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2F	♂ Brazilian: Unknown	>99%	Unknown
	♂ General: Unknown	83.33%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2I	♂ Brazilian: Unknown	34.62%	Unknown
	♂ Danish: 1/100	85.53%	1/691
	♂ General: Unknown	43.18%	Unknown
	♂ German: 1/300	82.50%	1/1,714
Lipoprotein Lipase Deficiency	♂ French Canadian: 1/44	28.95%	1/62
	♂ General: Unknown	20.00%	Unknown
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	♂ European: 1/126	88.98%	1/1,144
	♂ General: 1/126	56.25%	1/288
Lysinuric Protein Intolerance	♂ Finnish: 1/123	>99%	<1/12,300
	♂ Italian: 1/120	45.45%	1/220
	♂ Japanese: 1/115	37.93%	1/185
	♂ North African: Unknown	>99%	Unknown
MTHFR Deficiency: Severe	♂ Bukharan Jewish: 1/39	>99%	<1/3,900
Malonyl-CoA Decarboxylase Deficiency	♂ General: Unknown	33.33%	Unknown
Maple Syrup Urine Disease: Type 1A	♂ US Amish: 1/10	97.73%	1/440
Maple Syrup Urine Disease: Type 1B	♂ Ashkenazi Jewish: 1/97	>99%	<1/9,700
Maple Syrup Urine Disease: Type 2	♂ General: 1/481	42.31%	1/834
	♂ Norwegian: 1/481	50.00%	1/962
	♂ Turkish: 1/112	58.33%	1/269
Maple Syrup Urine Disease: Type 3	♂ Ashkenazi Jewish: 1/94	>99%	<1/9,400
	♂ General: Unknown	68.75%	Unknown
Maroteaux-Lamy Syndrome	♂ Argentinian: 1/274	75.00%	1/1,096
	♂ General: 1/388	61.54%	1/1,009
	♂ Spaniard: 1/274	29.17%	1/387
Meckel Syndrome: Type 1	♂ European: 1/212	72.22%	1/763
	♂ Finnish: 1/48	>99%	<1/4,800
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	♂ European: 1/50	90.91%	1/550
	♂ Saudi Arabian: 1/68	95.00%	1/1,360
	♂ United Kingdom: 1/51	90.00%	1/510
Megalencephalic Leukoencephalopathy	♂ Japanese: Unknown	50.00%	Unknown
	♂ Libyan Jewish: 1/40	>99%	<1/4,000
	♂ Turkish: Unknown	20.00%	Unknown
Metachromatic Leukodystrophy	♂ European: 1/150	43.88%	1/267
	♂ Habbanite Jewish: 1/5	50.00%	1/10

Disease	Carrier Rate	Detection Rate	Residual Risk
Methylmalonic Acidemia: MMAA Related	♂ General: 1/274	63.51%	1/751
Methylmalonic Acidemia: MMAB Related	♂ General: 1/396	71.25%	1/1,377
Methylmalonic Acidemia: MUT Related	♂ General: 1/177	43.62%	1/314
Methylmalonic Aciduria and Homocystinuria: Type cblC	♂ Chinese: Unknown	61.39%	Unknown
	♂ General: 1/159	65.74%	1/464
	♂ Italian: Unknown	75.00%	Unknown
	♂ Portuguese: Unknown	91.18%	Unknown
Mitochondrial Complex I Deficiency: NDUFS6 Related	♂ Caucasus Jewish: 1/24	>99%	<1/2,400
Mitochondrial DNA Depletion Syndrome: MNGIE Type	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: Unknown	47.37%	Unknown
	♂ Iranian Jewish: Unknown	>99%	Unknown
Mitochondrial Myopathy and Sideroblastic Anemia	♂ Iranian Jewish: Unknown	>99%	Unknown
Mitochondrial Trifunctional Protein Deficiency: HADHB Related	♂ Japanese: Unknown	60.00%	Unknown
Morquio Syndrome: Type A	♂ Colombian: 1/257	85.00%	1/1,713
	♂ European: 1/257	20.97%	1/325
	♂ Finnish: 1/257	50.00%	1/514
	♂ Latin American: 1/257	36.11%	1/402
Morquio Syndrome: Type B	♂ European: Unknown	83.33%	Unknown
Mucolipidosis: Type II/III	♂ General: 1/158	24.60%	1/210
	♂ Japanese: 1/252	51.25%	1/517
	♂ Korean: Unknown	30.00%	Unknown
	♂ Portuguese: 1/176	50.00%	1/352
Mucolipidosis: Type IV	♂ Ashkenazi Jewish: 1/97	96.15%	1/2,522
Multiple Pterygium Syndrome	♂ European: Unknown	41.67%	Unknown
	♂ Middle Eastern: Unknown	60.00%	Unknown
	♂ Pakistani: Unknown	50.00%	Unknown
Multiple Sulfatase Deficiency	♂ Ashkenazi Jewish: 1/320	95.00%	1/6,400
	♂ General: 1/501	18.18%	1/612
Muscle-Eye-Brain Disease	♂ European: Unknown	54.17%	Unknown
	♂ Finnish: 1/112	97.37%	1/4,256
	♂ General: Unknown	23.53%	Unknown
	♂ United States: Unknown	25.00%	Unknown
Navajo Neurohepatopathy	♂ Navajo: 1/39	>99%	<1/3,900
Nemaline Myopathy: NEB Related	♂ Ashkenazi Jewish: 1/108	>99%	<1/10,800
Nephrotic Syndrome: Type 1	♂ Finnish: 1/45	76.84%	1/194
	♂ US Amish: 1/12	50.00%	1/24
Nephrotic Syndrome: Type 2	♂ Israeli-Arab: Unknown	55.56%	Unknown
	♂ Pakistani: Unknown	20.00%	Unknown
	♂ Polish: Unknown	16.18%	Unknown
	♂ Saudi Arabian: Unknown	72.73%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Neuronal Ceroid-Lipofuscinosis: CLN5 Related	♂ Finnish: 1/101	>99%	<1/10,100
Neuronal Ceroid-Lipofuscinosis: CLN6 Related	♂ European: 1/159	36.36%	1/250
	♂ General: 1/159	59.52%	1/393
	♂ Portuguese: 1/128	81.00%	1/674
Neuronal Ceroid-Lipofuscinosis: CLN8 Related	♂ Finnish: 1/135	>99%	<1/13,500
	♂ Italian: 1/212	33.33%	1/318
	♂ Turkish: Unknown	77.78%	Unknown
Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	♂ General: 1/159	56.25%	1/363
Neuronal Ceroid-Lipofuscinosis: PPT1 Related	♂ Finnish: 1/58	97.62%	1/2,436
	♂ General: 1/159	72.50%	1/578
Neuronal Ceroid-Lipofuscinosis: TPP1 Related	♂ Canadian: 1/159	67.50%	1/489
	♂ European: 1/159	75.00%	1/636
	♂ General: 1/159	50.00%	1/318
	♂ Newfoundlander: 1/43	85.29%	1/292
Niemann-Pick Disease: Type A	♂ Ashkenazi Jewish: 1/101	95.00%	1/2,020
Niemann-Pick Disease: Type B	♂ Czech: 1/276	83.33%	1/1,656
	♂ General: Unknown	19.82%	Unknown
	♂ North African: Unknown	86.67%	Unknown
	♂ Spaniard: Unknown	38.10%	Unknown
Niemann-Pick Disease: Type C1	♂ Acadian: Unknown	>99%	Unknown
	♂ General: 1/194	15.60%	1/230
	♂ Japanese: Unknown	18.18%	Unknown
	♂ Portuguese: 1/194	25.00%	1/259
Niemann-Pick Disease: Type C2	♂ General: 1/194	75.00%	1/776
Nijmegen Breakage Syndrome	♂ Eastern European: 1/155	>99%	<1/15,500
Nonsyndromic Hearing Loss and Deafness: GJB2 Related	♂ Ashkenazi Jewish: 1/20	95.83%	1/480
	♂ Chinese: 1/100	82.26%	1/564
	♂ European: 1/53	82.47%	1/302
	♂ Ghanaian: Unknown	90.91%	Unknown
	♂ Indian: Unknown	66.98%	Unknown
	♂ Israeli: 1/16	93.10%	1/232
	♂ Japanese: 1/75	75.00%	1/300
	♂ Roma: Unknown	>99%	Unknown
	♂ United States: 1/34	45.22%	1/62
Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related	♂ Ashkenazi Jewish: 1/180	>99%	<1/18,000
Nonsyndromic Hearing Loss and Deafness: MYO15A Related	♂ Balinese: 1/6	>99%	<1/600
	♂ Pakistani: 1/77	24.00%	1/101
Oculocutaneous Albinism: Type 1	♂ European: 1/101	26.32%	1/137
	♂ Hutterite: 1/7	>99%	<1/700
	♂ Moroccan Jewish: 1/30	71.88%	1/107

Disease	Carrier Rate	Detection Rate	Residual Risk
	♂ Puerto Rican: Unknown	91.67%	Unknown
Oculocutaneous Albinism: Type 3	♂ Black South African: 1/47	94.74%	1/893
Oculocutaneous Albinism: Type 4	♂ Japanese: 1/146	58.33%	1/350
Omenn Syndrome: DCLRE1C Related	♂ Apache: 1/29	>99%	<1/2,900
	♂ Navajo: 1/29	97.22%	1/1,044
Omenn Syndrome: RAG2 Related	♂ Arab: Unknown	40.00%	Unknown
	♂ Non-Ashkenazi Jewish: Unknown	70.00%	Unknown
Ornithine Translocase Deficiency	♂ French Canadian: 1/20	95.00%	1/400
	♂ Italian: Unknown	18.75%	Unknown
	♂ Japanese: Unknown	60.00%	Unknown
Osteopetrosis: TCIRG1 Related	♂ Ashkenazi Jewish: 1/350	>99%	<1/35,000
	♂ Costa Rican: Unknown	>99%	Unknown
	♂ General: 1/251	25.00%	1/335
POLG Related Disorders: Autosomal Recessive	♂ Belgian: Unknown	85.00%	Unknown
	♂ Finnish: 1/140	>99%	<1/14,000
	♂ General: Unknown	93.10%	Unknown
	♂ Norwegian: Unknown	>99%	Unknown
Papillon-Lefevre Syndrome	♂ General: Unknown	35.29%	Unknown
	♂ Indian Jewish: Unknown	>99%	Unknown
	♂ Turkish: Unknown	50.00%	Unknown
Pendred Syndrome	♂ European: 1/58	42.11%	1/100
	♂ Japanese: Unknown	45.83%	Unknown
	♂ Pakistani: Unknown	29.82%	Unknown
Persistent Mullerian Duct Syndrome: Type I	♂ General: Unknown	28.12%	Unknown
Persistent Mullerian Duct Syndrome: Type II	♂ General: Unknown	78.12%	Unknown
Phenylalanine Hydroxylase Deficiency	♂ Arab: Unknown	46.08%	Unknown
	♂ Ashkenazi Jewish: 1/224	44.44%	1/403
	♂ Brazilian: 1/71	56.41%	1/163
	♂ Chinese: 1/51	76.57%	1/218
	♂ Cuban: 1/71	69.64%	1/234
	♂ European: 1/51	73.00%	1/189
	♂ French Canadian: 1/80	76.27%	1/337
	♂ Iranian: 1/31	66.94%	1/94
	♂ Korean: 1/51	51.52%	1/105
	♂ Non-Ashkenazi Jewish: Unknown	63.64%	Unknown
	♂ Slovakian Gypsy: 1/39	>99%	<1/3,900
	♂ Spanish Gypsy: 1/4	93.75%	1/64
	♂ Taiwanese: Unknown	83.10%	Unknown
	♂ US Amish: 1/16	86.84%	1/122
Polyglandular Autoimmune Syndrome: Type I	♂ Finnish: 1/80	90.48%	1/840
	♂ Iranian Jewish: 1/48	>99%	<1/4,800

Disease	Carrier Rate	Detection Rate	Residual Risk
	♂ Italian: Unknown	27.78%	Unknown
	♂ Norwegian: 1/142	47.92%	1/273
	♂ Sardinians: 1/61	81.82%	1/336
	♂ United Kingdom: Unknown	70.00%	Unknown
	♂ United States: Unknown	65.62%	Unknown
Pontocerebellar Hypoplasia: EXOSC3 Related	♂ General: Unknown	83.33%	Unknown
Pontocerebellar Hypoplasia: RARS2 Related	♂ Sephardic Jewish: Unknown	>99%	Unknown
Pontocerebellar Hypoplasia: SEPSECS Related	♂ Iraqi Jewish: 1/42	>99%	<1/4,200
Pontocerebellar Hypoplasia: TSEN54 Related	♂ European: 1/250	95.65%	1/5,750
Pontocerebellar Hypoplasia: VPS53 Related	♂ Moroccan Jewish: 1/37	>99%	<1/3,700
Pontocerebellar Hypoplasia: VRK1 Related	♂ Ashkenazi Jewish: 1/225	>99%	<1/22,500
Primary Carnitine Deficiency	♂ European: 1/101	58.33%	1/242
	♂ Faroese: 1/9	53.95%	1/20
	♂ General: Unknown	20.22%	Unknown
Primary Ciliary Dyskinesia: DNAI1 Related	♂ European: 1/211	52.38%	1/443
Primary Ciliary Dyskinesia: DNAI2 Related	♂ Ashkenazi Jewish: 1/200	>99%	<1/20,000
Primary Congenital Glaucoma	♂ Moroccan: Unknown	>99%	Unknown
	♂ Saudi Arabian: 1/23	91.67%	1/276
	♂ Turkish: 1/51	70.59%	1/173
Primary Hyperoxaluria: Type 1	♂ Dutch: 1/174	62.12%	1/459
	♂ General: 1/189	52.68%	1/399
Primary Hyperoxaluria: Type 2	♂ General: Unknown	70.31%	Unknown
Primary Hyperoxaluria: Type 3	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ European: Unknown	25.00%	Unknown
Progressive Familial Intrahepatic Cholestasis: Type 2	♂ European: Unknown	33.33%	Unknown
Propionic Acidemia: PCCA Related	♂ Japanese: 1/102	86.67%	1/765
Propionic Acidemia: PCCB Related	♂ General: 1/182	42.86%	1/319
	♂ Greenlandic Inuit: 1/16	58.33%	1/38
	♂ Japanese: 1/102	78.00%	1/464
	♂ Korean: Unknown	56.25%	Unknown
	♂ Latin American: 1/182	75.00%	1/728
	♂ Spaniard: 1/182	52.38%	1/382
Pseudocholinesterase Deficiency	♂ General: 1/33	65.00%	1/94
	♂ Iranian Jewish: 1/9	>99%	<1/900
Pycnodysostosis	♂ Danish: Unknown	87.50%	Unknown
Pyruvate Carboxylase Deficiency	♂ General: 1/251	62.50%	1/669
	♂ Native American: 1/10	>99%	<1/1,000
Pyruvate Dehydrogenase Deficiency	♂ General: Unknown	50.00%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Renal Tubular Acidosis and Deafness	♂ Colombian (Antioquia): Unknown	92.86%	Unknown
Retinal Dystrophies: RLBP1 Related	♂ Newfoundlander: 1/106	>99%	<1/10,600
	♂ Swedish: 1/84	>99%	<1/8,400
Retinal Dystrophies: RPE65 Related	♂ Dutch: 1/32	>99%	<1/3,200
	♂ North African Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: CERKL Related	♂ Yemenite Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: DHDDS Related	♂ Ashkenazi Jewish: 1/91	>99%	<1/9,100
Retinitis Pigmentosa: FAM161A Related	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ Non-Ashkenazi Jewish: 1/32	>99%	<1/3,200
Rhizomelic Chondrodysplasia Punctata: Type I	♂ General: 1/159	72.68%	1/582
Salla Disease	♂ European: Unknown	33.33%	Unknown
	♂ Scandinavian: 1/200	94.27%	1/3,491
Sandhoff Disease	♂ Argentinian: Unknown	95.45%	Unknown
	♂ Cypriot: 1/7	80.00%	1/35
	♂ Italian: Unknown	29.17%	Unknown
	♂ Spaniard: Unknown	64.29%	Unknown
Sanfilippo Syndrome: Type A	♂ Australasian: 1/119	44.12%	1/213
	♂ Dutch: 1/78	63.10%	1/211
	♂ European: 1/159	35.16%	1/245
	♂ United States: 1/159	32.14%	1/234
Sanfilippo Syndrome: Type B	♂ Australasian: 1/230	28.00%	1/319
	♂ Dutch: Unknown	42.31%	Unknown
	♂ European: Unknown	52.38%	Unknown
	♂ Japanese: 1/200	81.82%	1/1,100
Sanfilippo Syndrome: Type C	♂ Dutch: 1/346	75.00%	1/1,384
	♂ Greek: 1/415	25.00%	1/553
	♂ Moroccan: Unknown	80.00%	Unknown
	♂ Spaniard: Unknown	64.29%	Unknown
Sanfilippo Syndrome: Type D	♂ General: 1/501	83.33%	1/3,006
Short-Chain Acyl-CoA Dehydrogenase Deficiency	♂ Ashkenazi Jewish: 1/15	65.00%	1/43
Sickle-Cell Anemia	♂ African American: 1/10	>99%	<1/1,000
	♂ Hispanic American: 1/95	>99%	<1/9,500
Sjogren-Larsson Syndrome	♂ Dutch: Unknown	25.86%	Unknown
	♂ Swedish: 1/205	>99%	<1/20,500
Sly Syndrome	♂ General: 1/251	35.71%	1/390
Smith-Lemli-Opitz Syndrome	♂ Brazilian: 1/94	79.17%	1/451
	♂ European: 1/71	84.72%	1/465
	♂ Japanese: Unknown	71.43%	Unknown
	♂ United States: 1/70	95.00%	1/1,400
Stargardt Disease	♂ General: 1/51	17.51%	1/62
Stuve-Wiedemann Syndrome	♂ Emirati: 1/70	>99%	<1/7,000

Disease	Carrier Rate	Detection Rate	Residual Risk
	♂ General: Unknown	75.00%	Unknown
Sulfate Transporter-Related Osteochondrodysplasia	♂ Finnish: 1/51	95.83%	1/1,224
	♂ General: 1/100	70.00%	1/333
Tay-Sachs Disease	♂ Argentinian: 1/280	82.35%	1/1,587
	♂ Ashkenazi Jewish: 1/29	99.53%	1/6,177
	♂ Cajun: 1/30	>99%	<1/3,000
	♂ European: 1/280	25.35%	1/375
	♂ General: 1/280	32.09%	1/412
	♂ Indian: Unknown	85.71%	Unknown
	♂ Iraqi Jewish: 1/140	56.25%	1/320
	♂ Japanese: 1/127	82.81%	1/739
	♂ Moroccan Jewish: 1/110	22.22%	1/141
	♂ Portuguese: 1/280	92.31%	1/3,640
	♂ Spaniard: 1/280	67.65%	1/865
	♂ United Kingdom: 1/161	71.43%	1/564
Trichohepatoenteric Syndrome: Type 1	♂ European: 1/434	42.86%	1/760
	♂ South Asian: 1/434	66.67%	1/1,302
Tyrosine Hydroxylase Deficiency	♂ General: Unknown	36.11%	Unknown
Tyrosinemia: Type I	♂ Ashkenazi Jewish: 1/158	>99%	<1/15,800
	♂ European: 1/166	57.14%	1/387
	♂ Finnish: 1/123	97.22%	1/4,428
	♂ French Canadian: 1/64	96.30%	1/1,728
	♂ Pakistani: Unknown	92.86%	Unknown
Tyrosinemia: Type II	♂ General: 1/251	40.00%	1/418
Usher Syndrome: Type 1B	♂ European: 1/166	39.29%	1/273
	♂ General: 1/143	12.89%	1/164
	♂ North African: Unknown	66.67%	Unknown
	♂ Spaniard: 1/152	12.16%	1/173
Usher Syndrome: Type 1C	♂ Acadian: 1/82	98.86%	1/7,216
	♂ French Canadian: 1/227	83.33%	1/1,362
Usher Syndrome: Type 1D	♂ General: 1/296	23.17%	1/385
Usher Syndrome: Type 1F	♂ Ashkenazi Jewish: 1/126	93.75%	1/2,016
Usher Syndrome: Type 2A	♂ Chinese: Unknown	83.33%	Unknown
	♂ European: 1/136	40.00%	1/227
	♂ French Canadian: Unknown	66.67%	Unknown
	♂ General: 1/136	46.92%	1/256
	♂ Japanese: Unknown	55.56%	Unknown
	♂ Non-Ashkenazi Jewish: Unknown	61.11%	Unknown
	♂ Scandinavian: 1/125	39.22%	1/206
	♂ Spaniard: 1/133	39.02%	1/218
Usher Syndrome: Type 3	♂ Ashkenazi Jewish: 1/120	>99%	<1/12,000
	♂ Finnish: 1/134	>99%	<1/13,400

Disease	Carrier Rate	Detection Rate	Residual Risk
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	♂ General: 1/87	65.28%	1/251
Walker-Warburg Syndrome	♂ Ashkenazi Jewish: 1/150	>99%	<1/15,000
Werner Syndrome	♂ General: 1/224	31.25%	1/326
	♂ Japanese: 1/87	65.62%	1/253
Wilson Disease	♂ Ashkenazi Jewish: 1/100	>99%	<1/10,000
	♂ Canarian: 1/26	68.75%	1/83
	♂ Chinese: 1/51	55.97%	1/116
	♂ Cuban: Unknown	22.22%	Unknown
	♂ European: 1/93	41.64%	1/159
	♂ Greek: 1/90	44.94%	1/163
	♂ Korean: 1/88	51.53%	1/182
	♂ Spaniard: 1/93	38.18%	1/150
Wolcott-Rallison Syndrome	♂ Saudi Arabian: Unknown	66.67%	Unknown
Wolman Disease	♂ Iranian Jewish: 1/33	>99%	<1/3,300
Xeroderma Pigmentosum: Group A	♂ Japanese: 1/75	97.62%	1/3,150
	♂ North African: Unknown	87.50%	Unknown
	♂ Tunisian: 1/112	90.91%	1/1,232
Xeroderma Pigmentosum: Group C	♂ Moroccan: 1/71	76.19%	1/298
	♂ Tunisian: 1/51	>99%	<1/5,100
Zellweger Spectrum Disorders: PEX1 Related	♂ European: 1/139	70.27%	1/468
	♂ General: 1/139	67.84%	1/432
Zellweger Spectrum Disorders: PEX10 Related	♂ Japanese: Unknown	40.74%	Unknown
Zellweger Spectrum Disorders: PEX2 Related	♂ Ashkenazi Jewish: 1/123	>99%	<1/12,300
Zellweger Spectrum Disorders: PEX6 Related	♂ General: 1/288	30.00%	1/411