Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 5644

Genetic Testing Summary

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

Last Updated: 06/13/22

Donor Reported Ancestry: German, Swedish, English 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	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by gene sequencing in the CFTR gene	1/1250
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Expanded Genetic Disease Testing Panel attached- 289 diseases by gene sequencing	Negative for genes sequenced	
Special Testing		
Non-Syndromic Hearing Loss (GJB6)	Negative by gene sequencing in the GJB6 gene	
Schwachman-Diamond Syndrome (SBDS)	Negative by gene sequencing in the SBDS gene	
Genes: GDF6, DYNC2LI1, FLNB, CDAN1	Negative by gene sequencing	
Phenylalanine Hydroxylase Deficiency (PAH)	Possible Carrier: Variant of Unknown Significance (VUS) in the PAH gene. - see attached	Partner testing is recommended before 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.

PREVENTION GENETICS

NAME	PATIENT ID
5644, Donor	[redacted]

PATIENT INFORMATION	SPECIMEN INFORMATION	PROVIDER INFORMATION
5644, Donor ID#: 5644 DOB: [redacted] Sex: Male	Type: DNA From Previous Test Previous PG ID: 2020-163-002 Requested: January 26, 2022 PG ID: 2022-026-320	Harvey Stern, MD, PhD Suzanne Seitz, MS, MPA, CGC Fairfax Cyrobank

MOLECULAR GENETICS REPORT:
Custom Panel with CNV Detection

SUMMARY OF RESULTS: Indeterminate

Gene, transcript	Mode of Inheritance, Gene OMIM	DNA Variants, predicted Effects, Zygosity	ClinVar ID	Highest Allele Frequency in a gnomAD Population	In Silico Missense Predictions	Interpretation
PAH, NM_000277.1	AR, 612349	c.244C>T, p.His82Tyr, Heterozygous	Not listed in ClinVar	Not Present	Conflicting	UNCERTAIN

Mode of Inheritance: Autosomal Dominant=AD, Autosomal Recessive=AR, X-Linked=XL
ClinVar ID: Variant accession (www.ncbi.nlm.nih.gov/clinvar)
GnomAD: Allele Frequency registered in a large population database (gnomad.broadinstitute.org). Value listed is the highest allele frequency reported within one of seven population categories recognized in gnomAD v.2.0 (The "Other" population is excluded).
Missense Predictions: Summarized output (Damaging, Conflicting, or Tolerated) via PolyPhen-2, SIFT, MutationTaster, and FATHMM (PMID: 26555599).

PAH VARIANT INFORMATION:

This patient is heterozygous in the *PAH* gene for a sequence variant defined as c.244C>T, which is predicted to result in the amino acid substitution p.His82Tyr. To our knowledge, this variant has not been reported in the literature or in a large population database (http://gnomad.broadinstitute.org), indicating this variant is rare. At this time, the clinical significance of this variant is uncertain due to the absence of conclusive functional and genetic evidence.

Pathogenic variants in *PAH* have been reported in individuals with autosomal recessive phenylketonuria (PKU) or non-PKU mild hyperphenylalanemia (HPA) (OMIM #261600). The clinical features in untreated individuals may include microcephaly, pigmentary issues of eyes, skin and hair, intellectual disability, seizures, defective myelin formation, hypotonia, and behavioral problems such as autism, aggression, anxiety disorders, obsessive-compulsive disorder, hyperactivity and attention deficit disorder (https://www.omim.org/clinicalSynopsis/261600 and https://gene.sfari.org/database/human-gene/*PAH*). Only one variant was detected in this gene. It is possible that the second pathogenic variant is not detectable by our sequencing test. This variant is not eligible for no-cost family follow-up testing.

This patient is apparently negative for copy number variants (CNVs) within the genomic regions of this test.

These results should be interpreted in the context of clinical findings, family history and other laboratory data.

All genetic tests have limitations. See limitations and other information for this test on the following page(s).

NOTES:

1) Since this test is performed using exome capture probes, a reflex to any of our exome-based tests is available (PGxome, PGxome Custom Panels).

2) Genetic counseling is recommended.

GENES ANALYZED: *DYNC2LI1*, *FLNB*, *GDF6*, *PAH*

SUMMARY STATISTICS:

Pipeline	Version	Average NGS Coverage	Fraction Bases Covered with NGS
Infinity_Pipeline	1.10.1	172x	97.9%

Minimum NGS coverage is ≥20x for all coding exons and +/-10bp of flanking DNA.

Electronically signed on February 14, 2022 by:
Hannah Cox, PhD, HCLD(ABB)
Laboratory Director

Electronically signed and reported on February 15, 2022 by:
Anthony Krentz, PhD, HCLD(ABB)
Laboratory Director

SUPPLEMENTAL INFORMATION V.21.07

Limitations and Other Test Notes: Interpretation of the test results is limited by the information that is currently available. Better interpretation should be possible in the future as our knowledge about human genetics and genetic disorders improves.

When Next Generation Sequencing (NGS) or Sanger sequencing does not reveal any difference from the reference sequence, or when a sequence variant is homozygous, we cannot be certain that we were able to detect both patient alleles. Occasionally, a patient may carry an allele which does not capture or amplify due for example to a large deletion or insertion.

Copy number variants (CNVs) of four exons or more in size are detected with sensitivity approaching 100% through analysis of NGS data. However, sensitivity for detection of CNVs smaller than four exons is lower (~75%).

Unless otherwise indicated, coverage includes all coding exons of the gene(s) analyzed plus 10 bases of flanking noncoding DNA in all available transcripts along with other non-coding regions in which pathogenic variants have been identified at PreventionGenetics or reported elsewhere.

In most cases, we are unable to determine the phase of sequence variants.

Our ability to detect minor sequence variants due to somatic mosaicism is limited. Sequence variants that are present in less than 15% of the patient's nucleated cells may not be detected.

Unless present within coding regions, runs of mononucleotide repeats (eg (A)n or (T)n) with n >8 in the reference sequence) are generally not analyzed because of strand slippage during amplification.

Unless otherwise indicated, DNA sequence data is obtained from a specific cell type (often leukocytes from whole blood). Test reports contain no information about the DNA sequence in other cell types.

We cannot be certain that the reference sequences are correct. Genome build hg19, GRCh37 (Feb2009) is currently used as our reference in nearly all cases.

We have confidence in our ability to track a specimen once it has been received by PreventionGenetics. However, we take no responsibility for any specimen labeling errors that occur before the sample arrives at PreventionGenetics.

Genetic counseling to help to explain test results to the patients and to discuss reproductive options is recommended.

Test Methods: We use NGS technologies to cover the coding regions of the targeted genes plus 10 bases of non-coding DNA flanking each exon. As required, genomic DNA is extracted from the specimen. The DNA corresponding to these regions is captured using hybridization probes. Captured DNA is sequenced using Illumina's Reversible Dye Terminator (RDT) platform NovaSeq 6000 using 150 by 150 bp paired end reads (Illumina, San Diego, CA, USA).

The following quality control metrics are generally achieved: >98% of target bases are covered at >20x, and mean coverage of target bases >100x. Data analysis is performed using internally developed software. Where available, specified genes for which the enhance option is selected are backfilled with Sanger sequencing to achieve 100% coverage.

For Sanger sequencing, Polymerase Chain Reaction (PCR) is used to amplify the necessary exons plus additional flanking non-coding sequence. After purification of the PCR products, cycle sequencing is carried out using the

Applied Biosystems Incorporated (ABI) Big Dye Terminator v.3.1 kit. PCR products are resolved by electrophoresis on an ABI 3730xl capillary sequencer. In most cases, cycle sequencing is performed separately in both the forward and reverse directions; in some cases, sequencing is performed twice in either the forward or reverse directions.

CNVs are also detected from NGS data. We utilize a CNV calling algorithm that compares mean read depth and distribution for each target in the test sample against multiple matched controls. Neighboring target read depth and distribution and zygosity of any variants within each target region are used to reinforce CNV calls. All reported CNVs are confirmed using another technology such as microarray-based Comparative Genomic Hybridization (aCGH), Chromosomal Microarray Analysis (CMA), Multiplex Ligation-dependent Probe Amplification (MLPA), or PCR. On occasion, it will not be technically possible to confirm a smaller CNV called by NGS. In these instances, the CNV will not be included on the report. Exome-wide CNV is available as an add-on order for tests performed on an exome-backbone.

All differences from the reference sequences (sequence variants) are assigned to one of seven interpretation categories (Pathogenic, Likely Pathogenic, Variant of Uncertain Significance, Likely Benign, Benign, Risk, and Pseudodeficiency) per ACMG Guidelines (Richards et al. 2015. PubMed ID: 25741868). Rare and undocumented synonymous variants are nearly always classified as likely benign if there is no indication that they alter protein sequence or disrupt splicing. Benign and Likely Benign variants are not listed in the reports but are available upon request. Risk and pseudodeficiency variants may not be listed on the report but are available upon request.

Human Genome Variation Society (HGVS) recommendations are used to describe sequence variants (http://www.hgvs.org).

FDA Notes: These results should be used in the context of available clinical findings, and should not be used as the sole basis for treatment. This test was developed and its performance characteristics determined by PreventionGenetics. US Food and Drug Administration (FDA) does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing.

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank - [redacted]
[redacted]
Physician: [redacted]
Report Generated: 2018-03-27

Donor 5644

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 114623
Kit Barcode: [redacted]
Specimen: Blood, #117105
Specimen Collection: 2018-03-15
Specimen Received: 2018-03-16
Specimen Analyzed: 2018-03-27

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: NO MUTATIONS IDENTIFIED

Donor 5644 was not identified to carry any pathogenic mutations in the gene(s) tested.

No 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 5644	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.

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_4424delCTTTT, 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, 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 (10): ♂ Genotyping | c.2076_2078delAAT (p.692delI), c.2080A>G (p.M694V), c.1437C>G (p.F479L), c.800C>T (p.T267I), 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.1829_1831delGCC (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 (p.G12fs), 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 (p.G12V), 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 (6): ♂ Genotyping | c.496+1G>A, c.238_239insC, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.36+1G>T, c.496+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 (23): ♂ Genotyping | c.14020A>G (p.R4674G), c.12067-2A>G, c.4338_4339delCT (p.C1447fs), c.2299delG (p.E767SfsX21), c.2276G>T (p.C759F), 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	51.18%	1/8
	♂ Armenian: 1/5	94.51%	1/91
	♂ Ashkenazi Jewish: 1/81	39.52%	1/134
	♂ Iraqi Jewish: 1/4	76.92%	1/17
	♂ Israeli Jewish: 1/5	62.26%	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	74.25%	1/19
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
	♂ 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	47.69%	1/260
	♂ Japanese: Unknown	55.56%	Unknown
	♂ Non-Ashkenazi Jewish: Unknown	61.11%	Unknown
	♂ Scandinavian: 1/125	40.52%	1/210
	♂ Spaniard: 1/133	53.66%	1/287
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

PREVENTION GENETICS

NAME	PATIENT ID
5644, Donor	[redacted]

PATIENT INFORMATION	SPECIMEN INFORMATION	PROVIDER INFORMATION
5644, Donor ID#: [redacted] DOB: [redacted] Sex: Male	Type: Whole Blood Collected: September 17, 2019 Received: September 19, 2019 PG ID: 2019-262-287	Harvey Stern, MD, PhD Suzanne Seitz, MS Fairfax Cyrobank

MOLECULAR GENETICS REPORT:
GJB6 Gene Sequencing with CNV Detection

SUMMARY OF RESULTS: NEGATIVE

RESULTS AND INTERPRETATIONS: In this patient, for the *GJB6* gene, we found no sequence variants.

This patient is apparently negative for copy number variants (CNVs) within the genomic region encompassing the *GJB6* gene.

These results should be interpreted in the context of clinical findings, family history and other laboratory data. All genetic tests have limitations. Please see limitations and other information for this test on the following pages.

NOTE: Since this test is performed using exome capture probes, a reflex to any of our exome-based tests is available (PGxome, PGxome Custom Panels).

GENE ANALYZED: *GJB6*

SUMMARY STATISTICS:

Pipeline	Version	Average NGS Coverage	Fraction Bases Covered with NGS
Titanium	1.1.0	113x	100.0%

Minimum NGS coverage is ≥20x for all exons and +/-10bp of flanking DNA, and ≥10x from 11-20bp of flanking DNA.

Electronically signed on October 06, 2019 by:
Wuyan Chen, PhD
Human Molecular Geneticist

Electronically signed and reported on October 07, 2019 by:
Diane Allingham-Hawkins, PhD, FCCMG, FACMG
Clinical Molecular Geneticist

SUPPLEMENTAL INFORMATION V.19.04
SEQUENCING WITH CNV DETECTION

Limitations and Other Test Notes
Interpretation of the test results is limited by the information that is currently available. Better interpretation should be possible in the future as our knowledge about human genetics and the patient's condition improve.

When Next Gen or Sanger sequencing does not reveal any difference from the reference sequence, or when a sequence variant is homozygous, we cannot be certain that we were able to detect both patient alleles. Occasionally, a patient may carry an allele which does not capture or amplify due for example to a large deletion or insertion.

Copy number variants (CNVs) of four exons or more in size are detected with sensitivity approaching 100% through analysis of Next Gen sequence data. However, sensitivity for detection of CNVs smaller than four exons is lower (we estimate ~75%).

Coverage includes all coding exons of the gene(s) analyzed plus 10 bases of flanking noncoding DNA in all available transcripts along with other non-coding regions in which pathogenic variants have been identified at PreventionGenetics or reported elsewhere.

In most cases, we are unable to determine the phase of sequence variants. In particular, when we find two likely causative variants for recessive disorders, we cannot be certain that the variants are on different chromosomes.

Our ability to detect minor sequence variants due to somatic mosaicism is limited. Sequence variants that are present in less than 50% of the patient's nucleated cells may not be detected.

Unless present within coding regions, runs of mononucleotide repeats (eg $(A)_n$ or $(T)_n$) with n >8 in the reference sequence) are generally not analyzed because of strand slippage during amplification.

Unless otherwise indicated, DNA sequence data is obtained from a specific cell type (often leukocytes from whole blood). Test reports contain no information about the DNA sequence in other cell types.

We cannot be certain that the reference sequences are correct. Genome build hg19, GRCh37 (Feb2009) is currently used as our reference in nearly all cases.

We have confidence in our ability to track a specimen once it has been received by PreventionGenetics. However, we take no responsibility for any specimen labeling errors that occur before the sample arrives at PreventionGenetics.

Genetic counseling to help to explain test results to the patients and to discuss reproductive options is recommended.

Reported results will typically not contain any additional information regarding pharmacogenetic analysis of genes, nor are these tests designed to help guide dosage requirements. Pharmacogenetic variant analysis is available, for a select list of genes, as an opt-in with PGxome® tests.

Test Methods
We use Next Generation Sequencing (NGS) technologies to cover the coding regions of the targeted genes plus 10 bases of non-coding DNA flanking each exon. As required, genomic DNA is extracted from the specimen. The DNA corresponding to these regions is captured using Agilent Clinical Research Exome hybridization

probes. Captured DNA is sequenced using Illumina's Reversible Dye Terminator (RDT) platform NovaSeq 6000 using 150 by 150 bp paired end reads (Illumina, San Diego, CA, USA).

The following quality control metrics are generally achieved: >98% of target bases are covered at >20x, and mean coverage of target bases >120x. Data analysis is performed using the internally developed software Titanium-Exome. Specified genes for which the enhance option is selected are backfilled with Sanger sequencing to achieve 100% coverage.

For Sanger sequencing, Polymerase Chain Reaction (PCR) is used to amplify the necessary exons plus additional flanking non-coding sequence. After purification of the PCR products, cycle sequencing is carried out using the ABI Big Dye Terminator v.3.1 kit. PCR products are resolved by electrophoresis on an ABI 3730xl capillary sequencer. In most cases, cycle sequencing is performed separately in both the forward and reverse directions; in some cases, sequencing is performed twice in either the forward or reverse directions.

Copy number variants (CNVs) are also detected from NGS data. We utilize a CNV calling algorithm that compares mean read depth and distribution for each target in the test sample against multiple matched controls. Neighboring target read depth and distribution and zygosity of any variants within each target region are used to reinforce CNV calls. All reported CNVs are confirmed using another technology such as aCGH, MLPA, or PCR. On occasion, it will not be technically possible to confirm a smaller CNV called by NGS. In these instances, the CNV will not be included on the report.

All differences from the reference sequences (sequence variants) are assigned to one of five interpretation categories (Pathogenic, Likely Pathogenic, Variant of Uncertain Significance, Likely Benign and Benign) per ACMG Guidelines (Richards et al. 2015). Rare and undocumented synonymous variants are nearly always classified as likely benign if there is no indication that they alter protein sequence or disrupt splicing. Benign variants are not listed in the reports, but are available upon request.

Human Genome Variation Society (HGVS) recommendations are used to describe sequence variants (http://www.hgvs.org).

FDA Notes

These results should be used in the context of available clinical findings, and should not be used as the sole basis for treatment. This test was developed and its performance characteristics determined by PreventionGenetics. US Food and Drug Administration (FDA) does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing.

PREVENTION GENETICS

NAME	PATIENT ID
5644, Donor	[redacted]

PATIENT INFORMATION	SPECIMEN INFORMATION	PROVIDER INFORMATION
5644, Donor ID#: [redacted] DOB: [redacted] Sex: Male	[redacted] Previous PG ID: 2019-262-287 Requested: June 11, 2020 PG ID: [redacted]	Harvey Stern, MD, PhD Suzanne Seitz, MS, MPA Fairfax Cyrobank

MOLECULAR GENETICS REPORT:
Shwachman-Diamond Syndrome via the *SBDS* Gene

SUMMARY OF RESULTS **NEGATIVE**

RESULTS AND INTERPRETATIONS: In this patient, for the *SBDS* gene, we found no sequence variants.

These results should be interpreted in context of clinical findings, family history and other laboratory data. All genetic tests have limitations. See limitations and other information for this test on the following page(s).

GENE(S) ANALYZED: *SBDS* (NM_016038.2)

Electronically signed on June 17, 2020 by:
Michael Chicka, PhD
Human Molecular Geneticist

Electronically signed and reported on June 17, 2020 by:
Juan Dong, PhD, FACMG
Clinical Molecular Geneticist

SUPPLEMENTAL INFORMATION V.18.09
SANGER SEQUENCING

Limitations of Test and Other Test Notes: Interpretation of the test results is limited by the information that is currently available. Better interpretation should be possible in the future as more data and knowledge about human genetics and this specific disorder are accumulated.

In exons where our sequencing did not reveal any variation between the two alleles, we cannot be certain that we were able to PCR amplify both of the patient's alleles. Occasionally, a patient may carry an allele which does not amplify, due for example to a deletion or a large insertion. In these cases, the report contains no information about the second allele. Similarly, our Sanger sequencing tests have almost no power to detect duplications, triplications, etc. of the gene sequences.

Only the indicated exons and roughly 10 bp of flanking non-coding sequence on each side were analyzed. Test reports contain no information about other portions of the gene, including many regulatory regions.

In nearly all cases, we are unable to determine the phase of sequence variants. In particular, when we find two likely causative variants for recessive disorders, we cannot be certain that the variants are on different alleles.

The ability to detect low-level mosaicism of variants is limited.

Runs of mononucleotide repeats (eg $(A)_n$ or $(T)_n$) with n >8 in the reference sequence are generally not analyzed because of strand slippage during PCR and cycle sequencing.

Unless otherwise indicated, the sequence data that we report are based on DNA isolated from a specific tissue (usually leukocytes). Test reports contain no information about gene sequences in other tissues.

We cannot be certain that the reference sequence(s) are correct. Exons, for example, may be misidentified. In cases where the genomic and mRNA sequences disagree, we use the genomic sequence as our reference.

We have confidence in our ability to track a specimen once it has been received by PreventionGenetics. However, we take no responsibility for specimen labeling errors that occur before the sample arrives at PreventionGenetics.

Genetic counseling to help to explain test results to the patients and to discuss reproductive or medical options is recommended.

Methodology: As required, DNA was extracted from the patient specimen. PCR was used to amplify the indicated exons plus additional flanking non-coding sequence. After cleaning of the PCR products, cycle sequencing was carried out using the ABI Big Dye Terminator v.3.1 kit. Products were resolved by electrophoresis on an ABI 3730xl capillary sequencer. In most cases, sequencing was performed in both forward and reverse directions.

Human Genome Variation Society (HGVS) recommendations are used to describe sequence variants (http://www.hgvs.org). All differences from the reference sequences are assigned to one of five interpretation categories (Pathogenic, Likely Pathogenic, Variant of Uncertain Significance, Likely Benign and Benign) per ACMG Guidelines (Richards et al. 2015. PubMed ID: 25741868). Rare variants and undocumented variants are nearly always classified as likely benign if there is no indication that they alter protein sequence or disrupt splicing. Benign variants are not listed in the reports, but are available upon request.

Data Transfer: PreventionGenetics recommends that DNA sequence information from this test be stored in the patient's electronic medical record. This will facilitate reinterpretation of the sequence in future, and will best benefit the patient and family members. Upon request, we will be pleased to transfer the sequence data.

FDA Notes: These results should be used in the context of available clinical findings, and should not be used as the sole basis for treatment. This test was developed and its performance characteristics determined by PreventionGenetics. US Food and Drug Administration (FDA) does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing.

SUPPLEMENTAL INFORMATION V.18.09
SHWACHMAN-DIAMOND SYNDROME VIA THE *SBDS* GENE

Clinical Features: Shwachman-Diamond Syndrome (SDS), also known as Shwachman-Bodian syndrome, is characterized by exocrine pancreatic dysfunction, bone marrow failure and skeletal abnormalities (Dall'Oca et al. 2012). The pancreatic dysfunction causes malabsorption, malnutrition and growth failure. Other features include short stature, hepatomegaly, recurrent infections, and bone abnormalities such as bone age delay, metaphyseal chondrodysplasia, and generalized osteopenia. Hematologic abnormalities cause single or multiple lineage cytopenia, such as neutropenia, thrombocytopenia, anemia or pancytopenia (Rommens and Durie 2008). A diagnosis of SDS also increases an individual's susceptibility to myleodysplasia syndrome (MDS), and acute myelogenous leukemia (Burroughs et al. 2009). Its prevalence is 1:76,000 births with no specific ethnic predilection (Rommens and Durie 2008).

Genetics: Shwachman-Diamond Syndrome is inherited in an autosomal recessive manner. It is caused by pathogenic variants in the *SBDS* gene. *SBDS* encodes a protein that is involved in ribosome biogenesis and mitotic spindle stabilization (Burroughs et al. 2009). Most parents of an affected individual are carriers of an *SBDS* mutation, however *de novo* mutations have been reported (Rommens and Durie 2008). Approximately 90% of the reported mutations are the result of gene conversion from an adjacent pseudogene, *SBDSP*, which shares 97% homology with *SBDS* but does not generate a functional protein (Boocock et al. 2003). Two variants within exon 2 (c.183_184delinsCT and c.258+2T>C) account for 76% of these mutations. Other mutations have been reported, but no genotype-phenotype correlations are known to exist (Rommens and Durie 2008). Up to 10% of patients lack an identifiable *SBDS* mutation, but can still be diagnosed clinically with SDS (Dall'Oca et al. 2012).

Testing Strategy: This test involves bidirectional DNA Sanger sequencing of all coding exons and ~10 bp of flanking noncoding sequence of the *SBDS* gene. We will also sequence any single exon (Test #100) or pair of exons (Test #200) in family members of patients with known mutations or to confirm research results.

Indications for Testing: Individuals who are suspected of Shwachman-Diamond Syndrome or individuals who have a family history of SDS and want to know their *SBDS* mutation carrier status.

Sensitivity of Test: The clinical sensitivity of this test is >90% (Rommens and Durie 2008).

The clinical sensitivity of large deletions and duplications is unknown, but gross deletions have been reported (Donadieu et al. 2012; Costa et al. 2007).

References: Please see the test description for this test on our website (www.preventiongenetics.com) for a full list of complete citations.

Patient Information
Name: Donor 5644
Date of Birth: [redacted]
Sema4 ID: [redacted]
Client ID: [redacted]
Indication: Carrier Screening

Specimen Information
Specimen Type: Purified DNA
Date Collected: 05/26/2022
Date Received: 06/03/2022
Final Report: 06/12/2022

Referring Provider
[redacted]
Fairfax Cryobank, Inc.
[redacted]

Custom Carrier Screen (1 gene)

with Personalized Residual Risk

SUMMARY OF RESULTS AND RECOMMENDATIONS

Negative

Negative for all genes tested: *CDAN1*
To view a full list of genes and diseases tested
please see Table 1 in this report

AR=Autosomal recessive; XL=X-linked

Recommendations

- Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder.

Test description

This patient was tested for the genes listed above using one or more of the following methodologies: target capture and short-read sequencing, long-range PCR followed by short-read sequencing, targeted genotyping, and/or copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see Table 1 for a list of genes and diseases tested with the patient's personalized residual risk. If personalized residual risk is not provided, please see the complete residual risk table at **go.sema4.com/residualrisk.** Only known pathogenic or likely pathogenic variants are reported. This carrier screening test does not report likely benign variants and variants of uncertain significance (VUS). If reporting of likely benign variants and VUS are desired in this patient, please contact the laboratory at 800-298-6470, option 2 to request an amended report.

Anastasia Larmore, Ph.D., Associate Laboratory Director
Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D

Genes and diseases tested

The personalized residual risks listed below are specific to this individual. The complete residual risk table is available at **go.sema4.com/residualrisk**

Table 1: List of genes and diseases tested with detailed results

Disease	Gene	Inheritance Pattern	Status	Detailed Summary
Negative				
Congenital Dyserythropoietic Anemia, Type Ia	*CDAN1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 470

AR=Autosomal recessive; XL=X-linked

Test methods and comments

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

Fragile X CGG Repeat Analysis (Analytical Detection Rate >99%)

PCR amplification using Asuragen, Inc. AmplideX® *FMR1* PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for *FMR1* CGG repeats in the premutation and full mutation size range were further analyzed by Southern blot analysis to assess the size and methylation status of the *FMR1* CGG repeat.

Genotyping (Analytical Detection Rate >99%)

Multiplex PCR amplification and allele specific primer extension analyses using the MassARRAY® System were used to identify certain recurrent variants that are complex in nature or are present in low copy repeats. Rare sequence variants may interfere with assay performance.

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%.

For alpha thalassemia, 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.

For Duchenne muscular dystrophy, the copy numbers of all *DMD* exons were analyzed. Potentially pathogenic single exon deletions and duplications are confirmed by a second method. Analysis of *DMD* is performed in association with sequencing of the coding regions.

For congenital adrenal hyperplasia, the copy number of the *CYP21A2* gene was analyzed. This analysis can detect large deletions typically due to unequal meiotic crossing-over between *CYP21A2* and the pseudogene *CYP21A1P*. Classic 30-kb deletions make up approximately 20% of *CYP21A2* pathogenic alleles. This test may also identify certain point mutations in *CYP21A2* caused by gene conversion events between *CYP21A2* and *CYP21A1P*. Some carriers may not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *CYP21A2* gene on one chromosome and loss of *CYP21A2* (deletion) on the other chromosome. Analysis of *CYP21A2* is performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For spinal muscular atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or individuals that carry an intragenic mutation in *SMN1*. Please also note that 2% of individuals diagnosed with SMA have a causative *SMN1* variant that occurred *de novo*, and therefore cannot be picked up by carrier screening in the parents. Analysis of *SMN1* is performed in association with short-read sequencing of exons 2a-7, followed by confirmation using long-range PCR (described below).

The presence of the c.*3+80T>G (chr5:70,247,901T>G) variant allele in an individual with Ashkenazi Jewish or Asian ancestry is typically indicative of a duplication of *SMN1*. When present in an Ashkenazi Jewish or Asian individual with two copies of *SMN1*, c.*3+80T>G is likely indicative of a silent (2+0) carrier. In individuals with two copies of *SMN1* with African American, Hispanic or Caucasian ancestry, the presence or absence of c.*3+80T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

MLPA for Gaucher disease (*GBA*), cystic fibrosis (*CFTR*), and non-syndromic hearing loss (*GJB2/GJB6*) will only be performed if indicated for confirmation of detected CNVs. If *GBA* analysis was performed, the copy numbers of exons 1, 3, 4, and 6 - 10 of the *GBA* gene (of 11 exons total) were analyzed. If *CFTR* analysis was performed, the copy numbers of all 27 *CFTR* exons were analyzed. If *GJB2/GJB6* analysis was performed, the copy number of the two *GJB2* exons were analyzed, as well as the presence or absence of the two upstream deletions of the *GJB2* regulatory region, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854).

Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)

NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelect^{TM}XT Low Input technology was used with a custom capture library to target the exonic regions and intron/exon splice junctions of the relevant genes, as well as a number of UTR, intronic or promoter regions that contain previously reported mutations. Libraries were pooled and sequenced on the Illumina NovaSeq 9000 platform, using paired-end 100 bp reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house.

The coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Most exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing. Please note that several genomic regions present difficulties in mapping or obtaining read depth >20X. These regions, which are described below, will not be reflexed to Sanger sequencing if the mapping quality or coverage is poor. Any variants identified during testing in these regions are confirmed by a second method and reported if determined to be pathogenic or likely pathogenic. However, as there is a possibility of false negative results within these regions, detection rates and residual risks for these genes have been calculated with the presumption that variants in these exons will not be detected, unless included in the MassARRAY® genotyping platform.

Exceptions: *ABCD1* (NM_000033.3) exons 8 and 9; *ACADSB* (NM_ 001609.3) chr10:124,810,695-124,810,707 (partial exon 9); *ADA* (NM_000022.2) exon 1; *ADAMTS2* (NM_014244.4) exon 1; *AGPS* (NM_003659.3) chr2:178,257,512-178,257,649 (partial exon 1); *ALDH7A1* (NM_001182.4) chr5:125,911,150-125,911,163 (partial exon 7) and chr5:125,896,807-125,896,821 (partial exon 10); *ALMS1* (NM_015120.4) chr2:73,612,990-73,613,041 (partial exon 1); *APOPT1* (NM_ 032374.4) chr14:104,040,437-104,040,455 (partial exon 3); *CDAN1* (NM_138477.2) exon 2; *CEP152* (NM_014985.3) chr15:49,061,146-49,061,165 (partial exon 14) and exon 22; *CEP290* (NM_025114.3) exon 5, exon 7, chr12:88,519,017-88,519,039 (partial exon 13), chr12:88,514,049-88,514,058 (partial exon 15), chr12:88,502,837-88,502,841 (partial exon 23), chr12:88,481,551-88,481,589 (partial exon 32), chr12:88,471,605-88,471,700 (partial exon 40); *CFTR* (NM_000492.3) exon 10; *COL4A4* (NM_000092.4) chr2:227,942,604-227,942,619 (partial exon 25); *COX10* (NM_001303.3) exon 6; *CYP11B1* (NM_000497.3) exons 3-7; *CYP11B2* (NM_000498.3) exons 3-7; *DNAI2* (NM_023036.4) chr17:72,308,136-72,308,147 (partial exon 12); *DOK7* (NM_173660.4) chr4:3,465,131-3,465,161 (partial exon 1) and exon 2; *DUOX2* (NM_014080.4) exons 6-8; *EIF2AK3* (NM_004836.5 exon 8; *EVC* (NM_153717.2) exon 1; *F5* (NM_000130.4) chr1:169,551,662-169,551,679 (partial exon 2); *FH* (NM_000143.3) exon 1; *GAMT* (NM_000156.5 exon 1; *GLDC* (NM_000170.2) exon 1; *GNPTAB* (NM_024312.4) chr17:4,837,000-4,837,400 (partial exon 2); *GNPTG* (NM_032520.4) exon 1; *GHR* (NM_000163.4) exon 3; *GYS2* (NM_021957.3) chr12:21,699,370-21,699,409 (partial exon 12); *HGSNAT* (NM_152419.2) exon 1; *IDS* (NM_000202.6 exon 3; *ITGB4* (NM_000213.4) chr17:73,749,976-73,750,060 (partial exon 33); *JAK3* (NM_000215.3) chr19:17,950,462-17,950,483 (partial exon 10); *LIFR* (NM_002310.5 exon 19; *LMBRD1* (NM_018368.3) chr6:70,459,226-70,459,257 (partial exon 5), chr6:70,447,828-70,447,836 (partial exon 7) and exon 12; *LYST* (NM_000081.3) chr1:235,944,158-235,944,176 (partial exon 16) and chr1:235,875,350-235,875,362 (partial exon 43); *MLYCD* (NM_012213.2) chr16:83,933,242-83,933,282 (partial exon 1); *MTR* (NM_000254.2) chr1 237,024,418-237,024,439 (partial exon 20) and chr1:237,038,019-237,038,029 (partial exon 24); *NBEAL2* (NM_015175.2) chr3 47,021,385-47,021,407 (partial exon 1); *NEB* (NM_001271208.1 exons 82-105; *NPC1* (NM_000271.4) chr18:21,123,519-21,123,538 (partial exon 14); *NPHP1* (NM_000272.3) chr2:110,937,251-110,937,263 (partial exon 3); *OCRL* (NM_000276.3) chrX:128,674,450-128,674,460 (partial exon 1); *PHKB* (NM_000293.2) exon 1 and chr16:47,732,498-47,732,504 (partial exon 30); *PIGN* (NM_176787.4) chr18:59,815,547-59,815,576 (partial exon 8); *PIP5K1C* (NM_012398.2) exon 1 and chr19:3637602-3637616 (partial exon 17); *POU1F1* (NM_000306.3) exon 5; *PTPRC* (NM_002838.4) exons 11 and 23; *PUS1* (NM_025215.5 chr12:132,414,446-132,414,532 (partial exon 2); *RPGRIP1L* (NM_015272.2) exon 23; *SGSH* (NM_000199.3) chr17:78,194,022-78,194,072 (partial exon 1); *SLC6A8* (NM_005629.3) exons 3 and 4; *ST3GAL5* (NM_003896.3) exon 1; *SURF1* (NM_003172.3) chr9:136,223,269-136,223,307 (partial exon 1); *TRPM6* (NM_017662.4) chr9:77,362,800-77,362,811 (partial exon 31); *TSEN54* (NM_207346.2) exon 1; *TYR* (NM_000372.4) exon 5; *VWF* (NM_000552.3) exons 24-26, chr12:6,125,675-6,125,684 (partial exon 30), chr12:6,121,244-6,121,265 (partial exon 33), and exon 34.

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, or regions that fall into the Exceptions mentioned above. 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.

Next Generation Sequencing for SMN1

Exonic regions and intron/exon splice junctions of *SMN1* and *SMN2* were captured, sequenced, and analyzed as described above. Any variants located within exons 2a-7 and classified as pathogenic or likely pathogenic were confirmed to be in either *SMN1* or *SMN2* using gene-specific long-range PCR analysis followed by Sanger sequencing. Variants located in exon 1 cannot be accurately assigned to either *SMN1* or *SMN2* using our current methodology, and so these variants are considered to be of uncertain significance and are not reported.

Copy Number Variant Analysis (Analytical Detection Rate >95%)

Large duplications and deletions were called from the relative read depths on an exon-by-exon basis using a custom exome hidden Markov model (XHMM) algorithm. Deletions or duplications determined to be pathogenic or likely pathogenic were confirmed by either a custom arrayCGH platform, quantitative PCR, or MLPA (depending on CNV size and gene content). While this algorithm is designed to pick up deletions and duplications of 2 or more exons in length, potentially pathogenic single-exon CNVs will be confirmed and reported, if detected.

Exon Array (Confirmation method) (Accuracy >99%)

The customized oligonucleotide microarray (Oxford Gene Technology) is a highly-targeted exon-focused array capable of detecting medically relevant microdeletions and microduplications at a much higher resolution than traditional aCGH methods. Each array matrix has approximately 180,000 60-mer oligonucleotide probes that cover the entire genome. This platform is designed based on human genome NCBI Build 37 (hg19) and the CGH probes are enriched to target the exonic regions of the genes in this panel.

Quantitative PCR (Confirmation method) (Accuracy >99%)

Th relative quantification PCR is utilized on a Roche Universal Library Probe (UPL) system, which relates the PCR signal of the target region in one group to another. To test for genomic imbalances, both sample DNA and reference DNA is amplified with primer/probe sets that specific to the target region and a control region with known genomic copy number. Relative genomic copy numbers are calculated based on the standard ΔΔCt formula.

Long-Range PCR (Analytical Detection Rate >99%)

Long-range PCR was performed to generate locus-specific amplicons for *CYP21A2*, *HBA1* and *HBA2* and *GBA*. The PCR products were then prepared for short-read NGS sequencing and sequenced. Sequenced reads were mapped back to the original genomic locus and run through the bioinformatics pipeline. If indicated, copy number from MLPA was correlated with the sequencing output to analyze the results. For *CYP21A2*, a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where two copies of a gene are located on the same chromosome in tandem, only the second copy will be amplified and assessed for potentially pathogenic variants, due to size limitations of the PCR reaction. However, because these alleles contain at least two copies of the *CYP21A2* gene in tandem, it is expected that this patient has at least one functional gene in the tandem allele and this patient is therefore less likely to be a carrier. When an individual carries both a duplication allele and a pathogenic variant, or multiple pathogenic variants, the current analysis may not be able to determine the phase (cis/trans configuration) of the *CYP21A2* alleles identified. Family studies may be required in certain scenarios where phasing is required to determine the carrier status.

Residual Risk Calculations

Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >30,000 variants and genomic frequency data from >138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining the *a priori* risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate. They are provided only as a guide for assessing approximate risk given a negative result, and values will vary based on the exact ethnic background of an individual. This report does not represent medical advice but should be interpreted by a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

Personalized Residual Risk Calculations

Agilent SureSelect^{TM}XT Low-Input technology was utilized in order to create whole-genome libraries for each patient sample. Libraries were then pooled and sequenced on the Illumina NovaSeq platform. Each sequencing lane was multiplexed to achieve 0.4-2x genome coverage, using paired-end 100 bp reads. The sequencing data underwent ancestral analysis using a customized, licensed bioinformatics algorithm that was validated in house. Identified sub-ethnic groupings were binned into one of 7 continental-level groups (African, East Asian, South Asian, Non-Finnish European, Finnish, Native American, and Ashkenazi Jewish) or, for those ethnicities that matched poorly to the continental-level groups, an 8th "unassigned" group, which were then used to select residual risk values for each gene. For individuals belonging to multiple high-

level ethnic groupings, a weighting strategy was used to select the most appropriate residual risk. For genes that had insufficient data to calculate ethnic-specific residual risk values, or for sub-ethnic groupings that fell into the "unassigned" group, a "worldwide" residual risk was used. This "worldwide" residual risk was calculated using data from all available continental-level groups.

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.

Tay-Sachs Disease (TSD) Enzyme Analysis (Analytical Detection Rate ≥98%)

Hexosaminidase activity and Hex A% activity were measured by a standard heat-inactivation, fluorometric method using artificial 4-MU-β-N-acetyl glucosaminide (4-MUG) substrate. This assay is highly sensitive and accurate in detecting Tay-Sachs carriers and individuals affected with TSD. Normal ranges of Hex A% activity are 55.0-72.0 for white blood cells and 58.0-72.0 for plasma. It is estimated that less than 0.5% of Tay-Sachs carriers have non-carrier levels of percent Hex A activity, and therefore may not be identified by this assay. In addition, this assay may detect individuals that are carriers of or are affected with Sandhoff disease. False positive results may occur if benign variants, such as pseudodeficiency alleles, interfere with the enzymatic assay. False negative results may occur if both *HEXA* and *HEXB* pathogenic or pseudodeficiency variants are present in the same individual.

Please note these tests were developed and their performance characteristics were determined by Sema4 Opco, 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.

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Additional disease-specific references available upon request.