Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 5623

Genetic Testing Summary

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

Last Updated: 07/05/22

Donor Reported Ancestry: Scottish, English, French, Irish

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 sequencing in the CFTR gene	1/1250
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/628
Expanded Genetic Disease Testing Panel attached- 289 diseases by gene sequencing	Carrier: Usher Syndrome Type 2 A (USH2A) Negative for other genes sequenced	Carrier testing recommended for those using this donor
Special Testing		
Genes: ALG6, LAMA2, GJB2, LDLR	Negative by gene sequencing. See attached for residual risks.	

*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.

CooperGenomics℠
a CooperSurgical company
Reprogenetics℠ Recombine℠ Genesis Genetics℠

CarrierMap℠

Ordering Practice

Practice Code: [redacted]
Fairfax Cryobank - [redacted]
[redacted]
Physician: [redacted]
Report Generated: 2018-06-19

Donor 5623

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 107,940
Kit Barcode: [redacted]
Specimen: Blood, #109,660
Specimen Collection: 2017-11-10
Specimen Received: 2017-11-11
Specimen Analyzed: 2018-06-19

TEST INFORMATION

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

Partner Not Tested

SUMMARY OF RESULTS: MUTATION(S) IDENTIFIED

Disease	Donor 5623	Partner Not Tested
Usher Syndrome: Type 2A (USH2A) ○ High Impact	Carrier (1 abnormal copy) Mutation: c.2276G>T (p.C759F) Method: Sequencing	

Reproductive Risk & Next Steps: Reproductive risk detected. Consider partner testing.

No other pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.
For additional disease information, please visit www.coopergenomics.com/diseases . To speak with a genetic counselor, call 855.687.4363 .

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

Recombine CLIA ID: 31D2100763
Reviewed by: Pere Colls, PhD, HCLD

ADDITIONAL RESULTS

The following results ARE NOT associated with an increased reproductive risk.

	Donor 5623	**Partner Not Tested**
SMN1 Copy Number [†] *Spinal Muscular Atrophy*	SMN1 Copy Number: 2 or more copies Method: Genotyping & dPCR Interpretation: **NORMAL** (See Tables Below)	

[†] 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.

Usher Syndrome: Type 2A

Usher Syndrome Type 2A is caused by mutations in the USH2A gene, which normally makes a protein that is important for normal hearing and vision. Affected individuals are usually born with hearing loss that ranges from mild to severe and typically affects the ability to hear high pitch and soft speech sounds. Affected individuals also develop an eye disease called retinitis pigmentosa in childhood or adolescence, which leads to night blindness and loss of peripheral vision (causing tunnel vision).

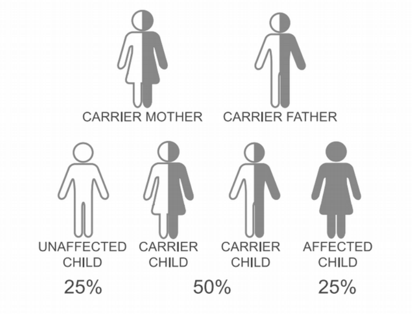
○ High Impact

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

Clinical Information

✓	Physical Impairment
	Cognitive Impairment
	Shortened Lifespan
	Effective Treatment

Inheritance: Autosomal Recessive



Prognosis

Prognosis for hearing loss and language development is fair, as it can be improved with early intervention. Prognosis for vision loss is unfavorable, as it is progressive. Individuals with Usher Syndrome Type 2A typically have normal cognition and life expectancy.

Treatment

There is no cure for the hearing and vision loss associated with Usher syndrome Type 2A. Hearing aids may be helpful for this type of Usher syndrome, and cochlear implants are sometimes considered. Treatments for vision loss may include use of low vision aids. Experimental therapies for retinitis pigmentosa, such as vitamin A palmitate supplements and lutein supplements may also be considered.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Chinese	83.33%	Unknown	Unknown
European	40.00%	1/136	1/227
French Canadian	66.67%	Unknown	Unknown
General	46.92%	1/136	1/256
Japanese	55.56%	Unknown	Unknown
Non-Ashkenazi Jewish	61.11%	Unknown	Unknown
Scandinavian	39.22%	1/125	1/206
Spaniard	39.02%	1/133	1/218

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

To learn more, visit www.coopergenomics.com/diseases

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. VUS reporting can be requested and will be assessed on a case-by-case basis. Variants may be re-curated over time due to emerging literature or other information. 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 existing mutations 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 does not currently regulate laboratory developed tests (LDTs).

Diseases & Mutations Assayed

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

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

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

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

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

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

3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related (MCCC1): Mutation(s) (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): Mutation(s) (8): ♂ Genotyping | c.1309A>G (p.I437V), c.295G>C (p.E99Q), c.464G>A (p.R155Q), c.499T>C (p.C167R), c.569A>G (p.H190R), c.803G>C (p.R268T), c.838G>T (p.D280Y), c.929C>G (p.P310R) | Sequencing | NM_022132:1-17

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Alport Syndrome: COL4A3 Related (COL4A3): Mutation(s) (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): Mutation(s) (5): ♂ Genotyping | c.3601G>A (p.G1201S), c.3713C>G (p.S1238X), c.4129C>T (p.R1377X), c.4715C>T (p.P1572L), c.4923C>A (p.C1641X) | Sequencing | NM_000092:2-48

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

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

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

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

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

Aromatase Deficiency (CYP19A1): Mutation(s) (10): ♂ Genotyping | c.1094G>A (p.R365Q), c.1123C>T (p.R375C), c.1224delC (p.K409fs), c.1303C>T (p.R435C), c.1310G>A (p.C437Y), c.296+1G>A (IVS3+1G>A), c.468delC, c.628G>A (p.E210K), c.629-3C>A (IVS4-3C>A), c.743+2T>C (IVS6+2T>C) | Sequencing | NM_000103:2-10

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

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

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

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

Ataxia-Telangiectasia (ATM): Mutation(s) (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.7449G>A (p.W2483X), c.7517_7520delGAGA (p.R2506fs), c.7630-2A>C, c.7638_7646delTAGAATTTC (p.R2547_S2549delRIS), c.7876G>C (p.A2626P), c.7967T>C (p.L2656P), c.8030A>G (p.Y2677C), c.8480T>G (p.F2827C) | Sequencing | NM_000051:2-63

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

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

Bardet-Biedl Syndrome: BBS10 Related (BBS10): Mutation(s) (3): ♂ Genotyping | c.101G>C (p.R34P), c.271_273ins1bp (p.C91fsX95), c.931T>G (p.S311A) | Sequencing | NM_024685:1-2

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

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

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

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

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

Beta Thalassemia (HBB): Mutation(s) (81): ♂ Genotyping | c.-136C>G, c.-137c>g, c.-137c>t, c.-138c>t, c.-140c>t, c.-142C>T, c.-151C>T, c.-29G>A, c.-50A>C, c.-78a>g, c.-79A>G, c.-80t>a, c.-81A>G, c.112delT, c.113G>A (p.W38X), c.114G>A (p.W38X), c.118C>T (p.Q40X), c.124_127delTTCT (p.F42Lfs), c.126delC, c.135delC (p.F46fs), c.154delC (p.P52fs), c.169G>C (p.G57R), c.17_18delCT, c.1A>G, c.203_204delTG (p.V68Afs), c.20delA (p.E7Gfs), c.217_218insA (p.S73Kfs), c.223+702_444+342del620insAAGTAGA, c.225delC, c.230delC, c.250delG, c.25_26delAA, c.271G>T (p.E91X), c.287_288insA (p.L97fs), c.295G>A (p.V99M), c.2T>C, c.2T>G, c.315+1G>A, c.315+2T>C, c.315+745C>G, c.316-146T>G, c.316-197C>T, c.316-1G>A, c.316-1G>C, c.316-1G>T, c.316-2A>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, c.321_322insG (p.N109fs), c.36delT (p.T13fs), c.383_385delAGG (p.Q128_A129delQAinsP), c.415G>C (p.A139P), c.444+111A>G, c.444+113A>G, c.45_46insG (p.W16fs), c.46delT (p.W16Gfs), c.47G>A (p.W16X), c.48G>A (p.W16X), c.4delG (p.V2Cfs), c.51delC (p.K18Rfs), c.52A>T (p.K18X), c.59A>G (p.N20S), c.68_74delAAGTTGG, c.75T>A (p.G25G), c.84_85insC (p.L29fs), c.90C>T (p.G30G), c.92+1G>A, c.92+1G>T, c.92+2T>A, c.92+2T>C, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.92G>C (p.R31T), c.93-15T>G, c.93-1G>A, c.93-1G>C, c.93-1G>T, c.93-21G>A | Sequencing | NM_000518:1-3

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

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

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

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

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

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

Carnitine Palmitoyltransferase II Deficiency (CPT2): Mutation(s) (20): ♂ Genotyping | c.109_110insGC, c.1148T>A (p.F383Y), c.1238_1239delAG, c.1342T>C (p.F448L), c.149C>A (p.P50H), c.1646G>A (p.G549D), c.1649A>G (p.Q550R), c.1737delC, c.1810C>T (p.P604S), c.1883A>C (p.Y628S), c.1891C>T (p.R631C), c.1923_1935delGAAGGCCTTAGAA, c.338C>T (p.S113L), c.359A>G (p.Y120C), c.370C>T (p.R124X), c.452G>A (p.R151Q), c.520G>A (p.E174K), c.534_558delGAACCCTGCAAAAAGTGACACTATCinsT, c.680C>T (p.P227L), c.983A>G (p.D328G) | Sequencing | NM_000098:1-5

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Congenital Lipoid Adrenal Hyperplasia (STAR): Mutation(s) (12): ♂ Genotyping | c.178+1_178+2insT (IVS2+3insT), c.201_202delCT, c.466-11T>A (IVS4-11T>A), c.545G>A (p.R182H), c.545G>T (p.R182L), c.559G>A (p.V187M), c.562C>T (p.R188C), c.64+1G>A, c.64+1G>T (IVS1+1G>T), c.650G>C (p.R217T), c.749G>A (p.W250X), c.772C>T (p.Q258X) | Sequencing | NM_000349:1-7

Congenital Myasthenic Syndrome: CHRNE Related (CHRNE): Mutation(s) (13): ♂ Genotyping | c.1327delG (p.E443fs), c.1353_1354insG (p.N452Efs), c.250C>G (p.R84G), c.344+1G>A, c.37G>A (p.G13R), c.422C>T (p.P141L), c.488C>T (p.S163L), c.500G>T (p.R167L), c.613_619delTGGGCCA (p.W205fs), c.850A>C (p.T284P), c.865C>T (p.L289F), c.911delT (p.L304fs), c.991C>T (p.R331W) | Sequencing | NM_000080:1-12

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

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

Congenital Neutropenia: Recessive (HAX1): Mutation(s) (6): ♂ Genotyping | c.121_125insG, c.130_131insA, c.256C>T (p.R86X), c.423_424insG, c.568C>T (p.Q190X), c.91delG | Sequencing | NM_006118:1-7

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

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

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

Cystic Fibrosis (CFTR): Mutation(s) (150): ♂ Genotyping | c.1000C>T (p.R334W), c.1013C>T (p.T338I), c.1029delC, c.1040G>A (p.R347H), c.1040G>C (p.R347P), c.1055G>A (p.R352Q), c.1075C>A (p.Q359K), c.1079C>A (p.T360K), c.1090T>C (p.S364P), c.1116+1G>A, c.1153_1154insAT, c.1175T>G (p.V392G), c.11C>A (p.S4X), c.1364C>A (p.A455E), c.1408_1417delGTGATTATGG (p.V470fs), c.1438G>T (p.G480C), c.1477C>T (p.Q493X), c.1477delCA, c.14C>T (p.P5L), c.1519_1521delATC (p.507delI), c.1521_1523delCTT (p.508delF), c.1526delG (p.G509fs), c.1545_1546delTA (p.Y515Xfs), c.1558G>T (p.V520F), c.1572C>A (p.C524X), c.1585-1G>A, c.1585-8G>A, c.1610_1611delAC (p.D537fs), c.1624G>T (p.G542X), c.164+12T>C, c.1645A>C (p.S549R), c.1646G>A (p.S549N), c.1646G>T (p.S549I), c.1647T>G (p.S549R), c.1652G>A (p.G551D), c.1654C>T (p.Q552X), c.1657C>T (p.R553X), c.1675G>A (p.A559T), c.1679G>C (p.R560T), c.1680-1G>A, c.1680-886A>G, c.171G>A (p.W57X), c.1721C>A (p.P574H), c.1766+1G>A, c.1766+1G>T, c.1766+5G>T, c.178G>T (p.E60X), c.1818del84, c.1865G>A (p.G622D), c.1911delG, c.1923delCTCAAAACTinsA, c.1973delGAAATTCAATCCTinsAGAAA, c.1976delA (p.N659fs), c.1986_1989delAACT (p.T663R), c.19G>T (p.E7X), c.200C>T (p.P67L), c.2051_2052delAAinsG (p.K684SfsX38), c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2089_2090insA (p.R697Kfs), c.2125C>T (p.R709X), c.2128A>T (p.K710X), c.2174insA, c.2215delG (p.V739Y), c.223C>T (p.R75X), c.2290C>T (p.R764X), c.2538G>A (p.W846X), c.254G>A (p.G85E), c.261delTT, c.263T>G (p.L196X), c.2657+5G>A, c.2668C>T (p.Q890X), c.271G>A (p.G91R), c.273+1G>A, c.273+3A>C, c.2737_2738insG (p.Y913X), c.274-1G>A, c.274G>T (p.E92X), c.2908+1085_3367+260del7201, c.2909G>A (p.G970D), c.293A>G (p.Q98R), c.2988+1G>A, c.3022delG (p.V1008S), c.3039delC, c.3067_3072delATAGTG (p.I1023_V1024delT), c.3139_3139+1delGG, c.313delA (p.I105fs), c.3140-26A>G, c.3196C>T (p.R1066C), c.3209G>A (p.R1070Q), c.3254A>G (p.H1085R), c.325delTATinsG, c.3266G>A (p.W1089X), c.3276C>G (p.Y1092X), c.328G>C (p.D110H), c.3302T>A (p.M1101K), c.3368-2A>G, c.3454G>C (p.D1152H), c.3472C>T (p.R1158X), c.3484C>T (p.R1162X), c.349C>T (p.R117C), c.350G>A (p.R117H), c.3527delC, c.3535delACCA, c.3536_3539delCCAA (p.T1179fs), c.3587C>G (p.S1196X), c.3611G>A (p.W1204X), c.3659delC (p.T1220fs), c.366T>A (p.Y122X), c.3691delT, c.3700A>G (p.I1234V), c.3712C>T (p.Q1238X), c.3717+12191C>T, c.3717+4A>G (IVS22+4A>G), c.3731G>A (p.G1244E), c.3744delA, c.3752G>A (p.S1251N), c.3764C>A (p.S1255X), c.3767_3768insC (p.A1256fs), c.3773_3774insT (p.L1258fs), c.3846G>A (p.W1282X), c.3848G>T (p.R1283M), c.3878_3881delTATT (p.V1293fs), c.3908dupA (p.N1303Kfs), c.3909C>G (p.N1303K), c.4003C>T (p.L1335F), c.416A>T (p.H139L), c.4364C>G (p.S1455X), c.4426C>T (p.Q1476X), c.442delA, c.455T>G (p.M152R), c.489+1G>T, c.496A>G (p.K166E), c.531delT, c.532G>A (p.G178R), c.535C>A (p.Q179K), c.54-5940_273+10250del21080bp (p.S18fs), c.579+1G>T, c.579+5G>A (IVS4+5G>A), c.580-1G>T, c.613C>T (p.P205S), c.617T>G (p.L206W), c.653T>A (p.L218X), c.658C>T (p.Q220X), c.803delA (p.N268fs), c.805_806delAT (p.I269fs), c.868C>T (p.Q290X), c.933_935delCTT (p.311delF), c.946delT, c.988G>T (p.G330X) | Sequencing | NM_000492:1-27

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

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

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

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

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

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

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

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

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

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

Ellis-van Creveld Syndrome: EVC2 Related (EVC2,EVC): Mutation(s) (3): ♂ Genotyping | c. 1858_1879delTTGGGCCGACTGGGCGGCCTC (p.L620_L626del), c.1868T>C (p.L623Q), c.3025C>T (p.Q1009X) | Sequencing | NM_147127:1-22

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

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

Familial Chloride Diarrhea (SLC26A3): Mutation(s) (6): ♂ Genotyping | c.1386G>A (p.W462X), c.2023_2025dupATC (p.I675L), c.344delT (p.I115I), c.371A>T (p.H124L), c.559G>T (p.G187X), c.951delGGT (p.V318del) | Sequencing | NM_000111:2-21

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

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

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

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

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

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

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

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

Fumarase Deficiency (FH): Mutation(s) (1): ♂ Genotyping | c.1433_1434insAAA | Sequencing | NM_000143:1-10

GM1-Gangliosidoses (GLB1): Mutation(s) (17): ♂ Genotyping | c.1051C>T (p.R351X), c.1369C>T (p.R457X), c.1370G>A (p.R457Q), c.145C>T (p.R49C), c.1480-2A>G, c.152T>C (p.I51T), c.1577_1578insG, c.176G>A (p.R59H), c.1771T>A (p.Y591N), c.1772A>G (p.Y591C), c.202C>T (p.R68W), c.245C>T (p.T82M), c.367G>A (p.G123R), c.601C>T (p.R201C), c.622C>T (p.R208C), c.75+2_75+3insT, c.947A>G (p.Y316C) | Sequencing | NM_000404:1-16

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

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

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

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

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

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

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

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

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

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

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

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

Glycogen Storage Disease: Type IB (SLC37A4): Mutation(s) (5): ♂ Genotyping | c.1016G>A (p.G339D), c.1042_1043delCT, c.1099G>A (p.A367T), c.133T>C (p.W45R), c.796G>T (p.G266C) | Sequencing | NM_001164277:3-11

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

Glycogen Storage Disease: Type III (AGL): Mutation(s) (14): ♂ Genotyping | c.1222C>T (p.R408X), c.1384delG (p.V462X), c.16C>T (p.Q6X), c.17_18delAG, c.2039G>A (p.W680X), c.2590C>T (p.R864X), c.2681+1G>A, c.3439A>G (p.R1147G), c.3682C>T (p.R1228X), c.3965delT (p.V1322AfsX27), c.3980G>A (p.W1327X), c.4260-12A>G (IVS32-12A>G), c.4342G>C (p.G1448R), c.4455delT (p.S1486fs) | Sequencing | NM_000642:2-34

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Hermansky-Pudlak Syndrome: Type 1 (HPS1): Mutation(s) (1): ♂ Genotyping | c.1472_1487dup16 (p.H497Qfs) | Sequencing | NM_000195:3-20

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Leber Congenital Amaurosis: GUCY2D Related (GUCY2D): Mutation(s) (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): Mutation(s) (3): ♂ Genotyping | c.1151delC, c.1476_1477insA (p.P493TfsX1), c.835C>T (p.Q279X) | Sequencing | NM_001122769:2-8

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Maple Syrup Urine Disease: Type 3 (DLD): Mutation(s) (8): ♂ Genotyping | c.104_105insA (p.Y35fs), c.1081A>G (p.M361V), c.1123G>A (p.E375K), c.1178T>C (p.I393T), c.1463C>T (p.P488L), c.1483A>G (p.R495G), c.214A>G (p.K72E), c.685G>T (p.G229C) | Sequencing | NM_000108:1-14

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

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

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (ACADM): Mutation(s) (8): ♂ Genotyping | c.199T>C (p.Y67H), c.262C>T (p.L88F), c.362C>T (p.T121I), c.595G>A (p.G199R), c.616C>T (p.R206C), c.617G>A (p.C206H), c.811C>T (p.G267R), c.985A>G (p.K329E) | Sequencing | NM_001127328:1-12

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

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

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

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

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

Methylmalonic Aciduria and Homocystinuria: Type cblC (MMACHC): Mutation(s) (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): Mutation(s) (1): ♂ Genotyping | c.344G>A (p.C115Y) | Sequencing | NM_004553:1-4

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

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

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

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

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

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

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

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

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

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

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

Nemaline Myopathy: NEB Related (NEB): Mutation(s) (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): Mutation(s) (5): ♂ Genotyping | c.121_122delCT (p.L41Dfs), c.1481delC, c.2335-1G>A, c.3325C>T (p.R1109X), c.3478C>T (p.R1160X) | Sequencing | NM_004646:1-29

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

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

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

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

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

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

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

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

Niemann-Pick Disease: Type B (SMPD1): Mutation(s) (3): ♂ Genotyping | c.1280A>G (p.H427R), c.1829_1831delGCC (p.610delR), c.880C>A (p.Q294K) | Sequencing | NM_000543:1-6

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

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

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

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutation(s) (29): ♂ Genotyping | c.-23+1G>A, c.-259C>T, c.109G>A (p.V37I), c.134G>A (p.G45E), c.139G>T (p.E47X), c.167delT, c.229T>C (p.W77R), c.231G>A (p.W77X), c.235delC, c.250G>C (p.V84L), c.269T>C (p.L90P), c.283G>A (p.V95M), c.290_291insA (p.Y97fs), c.299_300delAT (p.H100Rfs), c.313_326delAAGTTCATCAAGGG, c.334_335delAA (p.K112fs), c.358delGAG (p.120delE), c.35G>T (p.G12V), c.35delG (p.G12fs), c.370C>T (p.Q124X), c.427C>T (p.R143W), c.439G>A (p.E147K), c.44A>C (p.K15T), c.487A>G (p.M163V), c.516G>A (p.W172X), c.550C>T (p.R184W), c.551G>C (p.R184P), c.617A>G (p.N206S), c.71G>A (p.W24X) | Sequencing | NM_004004:1-2

Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related (LOXHD1): Mutation(s) (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): Mutation(s) (10): ♂ Genotyping | c.3313G>T (p.E1105X), c.3334delG (p.G1112fs), c.3685C>T (p.Q1229X), c.3866+1G>A, c.3866+1G>T, c.453_455delCGAinsTGGACGCCTGGTCGGGCAGTGG (p.E152GfsX81), c.6331A>T (p.N2111Y), c.6337A>T (p.I2113F), c.7801A>T (p.K2601X), c.8148G>T (p.Q2716H) | Sequencing | NM_016239:2-65

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

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

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

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

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

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

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

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

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

Pendred Syndrome (SLC26A4): Mutation(s) (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): Mutation(s) (6): ♂ Genotyping | c.1144G>T (p.E382X), c.1518C>G (p.H506Q), c.1574G>A (p.C525Y), c.17_18delTC, c.283C>T (p.R95X), c.571C>T (p.R191X) | Sequencing | NM_000479:1-4

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

Phenylalanine Hydroxylase Deficiency (PAH): Mutation(s) (62): ♂ Genotyping | c.1042C>G (p.L348V), c.1045T>C (p.S349P), c.1066-11G>A (IVS10-11G>A), c.1068C>G (p.Y356X), c.1139C>T (p.T380M), c.1157A>G (p.Y386C), c.1169A>G (p.E390G), c.117C>G (p.F39L), c.1222C>T (p.R408W), c.1223G>A (p.R408Q), c.1238G>C (p.R413P), c.1241A>G (p.Y414C), c.1301C>A (p.A434D), c.1315+1G>A (IVS12+1G>A), c.136G>A (p.G46S), c.143T>C (p.L48S), c.194T>C (p.I65T), c.199T>C (p.S67P), c.1A>G (p.M1V), c.241_256delACCCATTTGGATAAAC (p.T81fs), c.331C>T (p.R111X), c.3G>A (p.M1I), c.442-1G>A (IVS4-1G>A), c.456_706+138del11653, c.463_464insTGTGTACC (p.R155fs), c.473G>A (p.R158Q), c.533A>G (p.E178G), c.569T>G (p.V190G), c.581T>C (p.L194P), c.611A>G (p.Y204C), c.682G>T (p.E228X), c.721C>T (p.R241C), c.722G>A (p.R241H), c.722G>T (p.R241L), c.727C>T (p.R243X), c.728G>A (p.R243Q), c.734T>C (p.V245A), c.745C>T (p.L249F), c.754C>T (p.R252W), c.755G>A (p.R252Q), c.764T>C (p.L255S), c.770G>T (p.G257V), c.781C>T (p.R261X), c.782G>A (p.R261Q), c.800A>G (p.Q267R), c.814G>T (p.G272X), c.818C>T (p.S273F), c.829T>G (p.Y277D), c.838G>A (p.E280K), c.842+2T>A (IVS7+2T>A), c.842+5G>A (IVS7+5G>A), c.842C>T (p.P281L), c.856G>A (p.E286K), c.896T>G (p.F299C), c.898G>T (p.A300S), c.899C>T (p.A300V), c.904delT (p.F302fs), c.913-7A>G (IVS8-7A>G), c.926C>A (p.A309D), c.926C>T (p.A309V), c.935G>T (p.G312V), c.997C>T (p.L333F) | Sequencing | NM_000277:1-13

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

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

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

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

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

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

Pontocerebellar Hypoplasia: VRK1 Related (VRK1): Mutation(s) (2): ♂ Genotyping | c.1072C>T (p.R358X), c.397C>T (p.R133C) | Sequencing | NM_003384:2-13

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Short-Chain Acyl-CoA Dehydrogenase Deficiency (ACADS): Mutation(s) (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): Mutation(s) (1): ♂ Genotyping | c.20A>T (p.E7V) | Sequencing | NM_000518:1-3

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

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

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

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

Stargardt Disease (ABCA4): Mutation(s) (17): ♂ Genotyping | c.1018T>G (p.Y340D), c.1622T>C (p.L541P), c.1715G>A (p.R572Q), c.1938-1G>A, c.2461T>A (p.W821R), c.2565G>A (p.W855X), c.2588G>C (p.G863A), c.3083C>T (p.A1028V), c.3106G>A (p.E1036K), c.3113C>T (p.A1038V), c.3210_3211insGT (p.S1071Vfs), c.3364G>A (p.E1122K), c.52C>T (p.R18W), c.5338C>G (p.P1780A), c.571-2A>G, c.6079C>T (p.L2027F), c.634C>T (p.R212C) | Sequencing | NM_000350:1-50

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

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

Tay-Sachs Disease (HEXA): Mutation(s) (78): ♂ Genotyping | c.1003A>T (p.I335F), c.1008G>T (p.Q336H), c.1043_1046delTCAA (p.F348fs), c.1061_1063delTCT (p.F354_Y355delinsX), c.1073+1G>A, c.1121A>G (p.Q374R), c.1123delG (p.E375fs), c.1141delG (p.V381fs), c.1146+1G>A, c.116T>G (p.L39R), c.1177C>T (p.R393X), c.1178G>C (p.R393P), c.1211_1212delTG (p.L404fs), c.1277_1278insTATC, c.1292G>A (p.W431X), c.1302C>G (p.F434L), c.1307_1308delTA (p.I436fs), c.1351C>G (p.L451V), c.1385A>T (p.E462V), c.1421+1G>C, c.1422-2A>G, c.1426A>T (p.R476X), c.1432G>A (p.G478R), c.1451T>C (p.L484P), c.1495C>T (p.R499C), c.1496G>A (p.R499H), c.1510C>T (p.R504C), c.1510delC (p.R504fs), c.1511G>A (p.R504H), c.1511G>T (p.R504L), c.1537C>T (p.Q513X), c.155C>A (p.S52X), c.1A>G (p.M1V), c.2T>C (p.M1T), c.340G>A (p.E114K), c.346+1G>C, c.380T>G (p.L127R), c.409C>T (p.R137X), c.413-2A>G, c.426delT (p.F142fs), c.459+5G>A (IVS4+5G>A), c.508C>T (p.R170W), c.509G>A (p.R170Q), c.532C>T (p.R178C), c.533G>A (p.R178H), c.533G>T (p.R178L), c.535C>T (p.H179Y), c.536A>G (p.H179R), c.538T>C (p.Y180H), c.540C>G (p.Y180X), c.570+3A>G, c.571-1G>T, c.571-2A>G (IVS5-2A>G), c.571-8A>G, c.590A>C (p.K197T), c.598G>A (p.V200M), c.607T>G (p.W203G), c.611A>G (p.H204R), c.613delC, c.615delG (p.L205fs), c.621T>G (p.D207E), c.623A>T (p.D208V), c.624_627delTCCT (p.D208fs), c.629C>T (p.S210F), c.632T>C (p.F211S), c.736G>A (p.A246T), c.749G>A (p.G250D), c.778C>T (p.P260S), c.78G>A (p.W26X), c.796T>G (p.W266G), c.805+1G>A, c.805+1G>C, c.805+2T>C, c.805G>A (p.G269S), c.910_912delTTC (p.305delF), c.947_948insA (p.Y316fs), c.964G>A (p.D322N), c.964G>T (p.D322Y) | Sequencing | NM_000520:1-14

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

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

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

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

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

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

Usher Syndrome: Type 1D (CDH23): Mutation(s) (15): ♂ 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), c.9524G>A (p.R3175H) | Sequencing | NM_022124:2-68

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Zellweger Spectrum Disorders: PEX6 Related (PEX6): Mutation(s) (8): ♂ Genotyping | c.1130+1G>A (IVS3+1G>A), c.1301delC (p.S434Ffs), c.1601T>C (p.L534P), c.1688+1G>A (IVS7+1G>A), c.1715C>T (p.T572I), c.1962-1G>A (p.L655fsX3), c.511insT (p.G171Wfs), c.802_815delGACGGACTGGCGCT (p.D268Cfs) | 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 ♂ Japanese: Unknown	54.55% 45.45%	Unknown Unknown
17-Beta-Hydroxysteroid Dehydrogenase Deficiency	♂ Arab: 1/8 ♂ Dutch: 1/192	>99% 13.89%	<1/800 1/223
21-Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia	♂ European: 1/62 ♂ General: 1/62	27.65% 29.34%	1/86 1/88
21-Hydroxylase-Deficient Nonclassical Congenital Adrenal Hyperplasia	♂ Argentinian: 1/4 ♂ European: 1/16	<10% <10%	1/4 1/16
3-Beta-Hydroxysteroid Dehydrogenase Deficiency	♂ General: Unknown	16.13%	Unknown
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related	♂ European: 1/146 ♂ General: 1/112	26.32% 37.50%	1/198 1/179
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related	♂ General: 1/112 ♂ Japanese: 1/112 ♂ Korean: 1/141 ♂ Turkish: 1/112	35.29% 33.33% 66.67% 24.07%	1/173 1/168 1/423 1/148
3-Methylglutaconic Aciduria: Type 3	♂ Iraqi Jewish: 1/10	>99%	<1/1000
3-Phosphoglycerate Dehydrogenase Deficiency	♂ Ashkenazi Jewish: 1/400	>99%	<1/40000
5-Alpha Reductase Deficiency	♂ Dominican: Unknown ♂ Mexican: Unknown	>99% 68.75%	Unknown Unknown
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	♂ Chinese: 1/183 ♂ East Asian: 1/180	78.95% 64.20%	1/869 1/503
ARSACS	♂ French Canadian: 1/22	95.45%	1/484
Abetalipoproteinemia	♂ Ashkenazi Jewish: 1/131	>99%	<1/13100
Acrodermatitis Enteropathica	♂ Arab: Unknown ♂ Egyptian: Unknown ♂ French: Unknown ♂ Tunisian: Unknown	40.00% 33.33% 27.78% 77.78%	Unknown Unknown Unknown Unknown
Acute Infantile Liver Failure: TRMU Related	♂ Yemenite Jewish: 1/40	71.43%	1/140
Acyl-CoA Oxidase I Deficiency	♂ General: Unknown ♂ Japanese: Unknown	35.00% 42.86%	Unknown Unknown
Adenosine Deaminase Deficiency	♂ General: 1/388	36.96%	1/615
Alkaptonuria	♂ Dominican: Unknown ♂ Finnish: 1/251 ♂ Slovak: 1/69	>99% 60.00% 59.38%	Unknown 1/628 1/170
Alpha Thalassemia	♂ General: 1/48	50.67%	1/97
Alpha-1-Antitrypsin Deficiency	♂ European: 1/35 ♂ General: Unknown	95.00% 95.00%	1/700 Unknown
Alpha-Mannosidosis	♂ European: 1/354 ♂ General: 1/354	30.23% 35.19%	1/507 1/546
Alport Syndrome: COL4A3 Related	♂ Dutch: 1/409	22.73%	1/529
Alport Syndrome: COL4A4 Related	♂ General: 1/409	26.67%	1/558
Amegakaryocytic Thrombocytopenia	♂ Ashkenazi Jewish: 1/76 ♂ General: Unknown	>99% 64.81%	<1/7600 Unknown
Andermann Syndrome	♂ French Canadian: 1/24	99.38%	1/3888
Antley-Bixler Syndrome	♂ General: Unknown ♂ Japanese: Unknown	45.65% 60.47%	Unknown Unknown
Argininemia	♂ Chinese: Unknown ♂ French Canadian: Unknown ♂ Japanese: Unknown	40.00% 75.00% >99%	Unknown Unknown Unknown
Argininosuccinate Lyase Deficiency	♂ European: 1/133 ♂ Saudi Arabian: 1/80	57.41% 51.72%	1/312 1/166
Aromatase Deficiency	♂ General: Unknown	25.00%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Arthrogryposis, Mental Retardation, & Seizures	♂ Ashkenazi Jewish: 1/205	>99%	<1/20500
Asparagine Synthetase Deficiency	♂ Iranian Jewish: 1/80	>99%	<1/8000
Aspartylglycosaminuria	♂ Finnish: 1/69	96.12%	1/1780
Ataxia with Vitamin E Deficiency	♂ European: 1/274 ♂ Italian: 1/224 ♂ North African: 1/159	80.00% 97.73% >99%	1/1370 1/9856 <1/15900
Ataxia-Telangiectasia	♂ Costa Rican: 1/100 ♂ North African Jewish: 1/81 ♂ Norwegian: 1/197 ♂ Sardinians: Unknown ♂ US Amish: Unknown	68.52% 96.97% 50.00% 85.71% >99%	1/318 1/2673 1/394 Unknown Unknown
Autosomal Recessive Polycystic Kidney Disease	♂ Finnish: 1/45 ♂ French: 1/71 ♂ General: 1/71	84.21% 62.50% 37.11%	1/285 1/189 1/113
Bardet-Biedl Syndrome: BBS1 Related	♂ General: 1/376 ♂ Northern European: 1/376 ♂ Puerto Rican: Unknown	70.27% 85.90% 90.00%	1/1265 1/2666 Unknown
Bardet-Biedl Syndrome: BBS10 Related	♂ General: 1/404	47.79%	1/774
Bardet-Biedl Syndrome: BBS11 Related	♂ Bedouin: 1/59	>99%	<1/5900
Bardet-Biedl Syndrome: BBS12 Related	♂ General: Unknown	50.00%	Unknown
Bardet-Biedl Syndrome: BBS2 Related	♂ Ashkenazi Jewish: Unknown ♂ General: 1/638 ♂ Middle Eastern: Unknown	>99% 38.46% >99%	Unknown 1/1037 Unknown
Bare Lymphocyte Syndrome: Type II	♂ General: Unknown	66.67%	Unknown
Bartter Syndrome: Type 4A	♂ General: 1/457	81.82%	1/2514
Beta Thalassemia	♂ African American: 1/75 ♂ Indian: 1/24 ♂ Sardinians: 1/23 ♂ Spaniard: 1/51	84.21% 74.12% 97.14% 93.10%	1/475 1/93 1/804 1/740
Beta-Hexosaminidase Pseudodeficiency	♂ Ashkenazi Jewish: Unknown ♂ General: Unknown	>99% >99%	Unknown Unknown
Beta-Ketothiolase Deficiency	♂ Japanese: Unknown ♂ Spaniard: Unknown	58.33% 90.00%	Unknown Unknown
Biotinidase Deficiency	♂ General: 1/123	78.32%	1/567
Bloom Syndrome	♂ Ashkenazi Jewish: 1/134 ♂ European: Unknown ♂ Japanese: Unknown	96.67% 66.22% 50.00%	1/4020 Unknown Unknown
Canavan Disease	♂ Ashkenazi Jewish: 1/55 ♂ European: Unknown	98.86% 53.23%	1/4840 Unknown
Carnitine Palmitoyltransferase IA Deficiency	♂ General: Unknown ♂ Hutterite: 1/16 ♂ Japanese: 1/101	38.89% >99% 66.67%	Unknown <1/1600 1/303
Carnitine Palmitoyltransferase II Deficiency	♂ Ashkenazi Jewish: Unknown ♂ General: Unknown	>99% 71.43%	Unknown Unknown
Carnitine-Acylcarnitine Translocase Deficiency	♂ Asian: Unknown ♂ General: Unknown	95.45% 18.75%	Unknown Unknown
Carpenter Syndrome	♂ Brazilian: Unknown ♂ Northern European: Unknown	40.00% 85.00%	Unknown Unknown
Cartilage-Hair Hypoplasia	♂ Finnish: 1/76 ♂ US Amish: 1/19	93.33% >99%	1/1140 <1/1900
Cerebrotendinous Xanthomatosis	♂ Dutch: Unknown ♂ Italian: Unknown ♂ Japanese: Unknown ♂ Moroccan Jewish: 1/6	78.57% 45.95% 92.86% 87.50%	Unknown Unknown Unknown 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 ♂ Japanese: 1/274 ♂ Korean: 1/105 ♂ Moroccan Jewish: 1/234	71.43% >99% >99% >99%	Unknown <1/27400 <1/10500 <1/23400
Citrin Deficiency	♂ Japanese: 1/70	>99%	<1/7000
Citrullinemia: Type I	♂ European: 1/120 ♂ General: 1/120 ♂ Japanese: Unknown ♂ Mediterranean: 1/120	18.18% 52.27% 64.71% 50.00%	1/147 1/251 Unknown 1/240
Classical Galactosemia	♂ African American: 1/78 ♂ Ashkenazi Jewish: 1/127 ♂ Dutch: 1/91 ♂ European: 1/112 ♂ General: 1/125 ♂ Irish: 1/76 ♂ Irish Travellers: 1/14	73.13% >99% 75.47% 88.33% 80.00% 91.30% >99%	1/290 <1/12700 1/371 1/960 1/625 1/874 <1/1400
Cockayne Syndrome: Type A	♂ Christian Arab: Unknown	50.00%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Cockayne Syndrome: Type B	♂ General: 1/378	19.30%	1/468
Cohen Syndrome	♂ European: Unknown ♂ Finnish: 1/140 ♂ US Amish: 1/12	19.05% 67.24% >99%	Unknown 1/427 <1/1200
Combined Pituitary Hormone Deficiency: PROP1 Related	♂ European: 1/45 ♂ General: 1/45	93.29% 82.35%	1/671 1/255
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related	♂ Danish: 1/71 ♂ Dutch: 1/68 ♂ European: 1/71	90.00% 39.29% 55.33%	1/710 1/112 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 ♂ General: Unknown	59.09% 86.21%	Unknown Unknown
Congenital Ichthyosis: ABCA12 Related	♂ North African: Unknown ♂ South Asian: Unknown	>99% 66.67%	Unknown Unknown
Congenital Insensitivity to Pain with Anhidrosis	♂ Japanese: Unknown ♂ Moroccan Jewish: Unknown	56.52% >99%	Unknown Unknown
Congenital Lipoid Adrenal Hyperplasia	♂ Japanese: 1/201 ♂ Korean: 1/251	51.11% 63.64%	1/411 1/690
Congenital Myasthenic Syndrome: CHRNE Related	♂ European Gypsy: 1/26 ♂ North African: Unknown	>99% 60.87%	<1/2600 Unknown
Congenital Myasthenic Syndrome: DOK7 Related	♂ European: 1/472 ♂ General: 1/472	19.05% 18.75%	1/583 1/581
Congenital Myasthenic Syndrome: RAPSN Related	♂ General: 1/437 ♂ Non-Ashkenazi Jewish: Unknown	88.57% >99%	1/3824 Unknown
Congenital Neutropenia: Recessive	♂ English: Unknown ♂ Japanese: Unknown ♂ Turkish: Unknown	11.76% 22.22% 89.47%	Unknown Unknown Unknown
Corneal Dystrophy and Perceptive Deafness	♂ General: Unknown	71.43%	Unknown
Corticosterone Methyloxidase Deficiency	♂ Iranian Jewish: 1/32	>99%	<1/3200
Crigler-Najjar Syndrome	♂ Sardinians: Unknown ♂ Tunisian: Unknown	80.00% >99%	Unknown Unknown
Cystic Fibrosis	♂ African American: 1/62 ♂ Ashkenazi Jewish: 1/23 ♂ Asian: 1/94 ♂ European: 1/25 ♂ Hispanic American: 1/48 ♂ Native American: 1/53	69.99% 96.81% 65.81% 94.96% 77.32% 84.34%	1/207 1/721 1/275 1/496 1/212 1/338
Cystinosis	♂ Dutch: 1/194 ♂ French Canadian: 1/40 ♂ General: 1/194	73.08% 75.00% 54.51%	1/721 1/160 1/426
Cystinuria: Non-Type I	♂ European: 1/42 ♂ General: 1/42 ♂ Libyan Jewish: 1/26 ♂ United States: 1/42	61.11% 37.50% 93.48% 56.25%	1/108 1/67 1/399 1/96
Cystinuria: Type I	♂ European: 1/42 ♂ Swedish: 1/159	46.67% 55.88%	1/79 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 ♂ General: 1/501	>99% 50.00%	<1/20300 1/1002
Dystrophic Epidermolysis Bullosa: Recessive	♂ Italian: Unknown ♂ Mexican American: 1/345	45.00% 56.25%	Unknown 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 ♂ General: Unknown	90.48% 52.50%	Unknown Unknown
Ethylmalonic Aciduria	♂ Arab/Mediterranean: Unknown ♂ General: Unknown	29.17% 38.24%	Unknown Unknown
Familial Chloride Diarrhea	♂ Finnish: 1/51 ♂ Kuwaiti: 1/38 ♂ Polish: 1/224 ♂ Saudi Arabian: 1/38	>99% 90.00% 45.24% >99%	<1/5100 1/380 1/409 <1/3800
Familial Dysautonomia	♂ Ashkenazi Jewish: 1/31	>99%	<1/3100

Disease	Carrier Rate	Detection Rate	Residual Risk
Familial Hyperinsulinism: Type 1: ABCC8 Related	♂ Ashkenazi Jewish: 1/52 ♂ Finnish: 1/101	98.75% 45.16%	1/4160 1/184
Familial Hyperinsulinism: Type 2: KCNJ11 Related	♂ Arab: Unknown	40.00%	Unknown
Familial Mediterranean Fever	♂ Arab: 1/4 ♂ Armenian: 1/5 ♂ Ashkenazi Jewish: 1/81 ♂ Iraqi Jewish: 1/4 ♂ Israeli Jewish: 1/5 ♂ Lebanese: 1/6 ♂ North African Jewish: 1/5 ♂ Syrian: 1/6 ♂ Turkish: 1/5	51.27% 94.51% 40.95% 76.92% 62.67% 91.67% 95.69% 85.14% 74.43%	1/8 1/91 1/137 1/17 1/13 1/72 1/116 1/40 1/20
Fanconi Anemia: Type A	♂ Moroccan Jewish: 1/100 ♂ Spanish Gypsy: 1/67	>99% >99%	<1/10000 <1/6700
Fanconi Anemia: Type C	♂ Ashkenazi Jewish: 1/101 ♂ General: Unknown	>99% 30.00%	<1/10100 Unknown
Fanconi Anemia: Type G	♂ Black South African: 1/101 ♂ French Canadian: Unknown ♂ Japanese: Unknown ♂ Korean: Unknown	81.82% 87.50% 75.00% 66.67%	1/556 Unknown Unknown Unknown
Fanconi Anemia: Type J	♂ General: Unknown	86.36%	Unknown
Fumarase Deficiency	♂ General: Unknown	30.00%	Unknown
GM1-Gangliosidoses	♂ Eurodescent Brazilian: 1/66 ♂ European: 1/194 ♂ General: 1/194 ♂ Hispanic American: 1/194 ♂ Japanese: Unknown	62.15% 50.00% 20.00% 58.33% 62.82%	1/174 1/388 1/243 1/466 Unknown
GRACILE Syndrome	♂ Finnish: 1/109	97.22%	1/3924
Galactokinase Deficiency	♂ Japanese: 1/501 ♂ Roma: 1/51	50.00% >99%	1/1002 <1/5100
Gaucher Disease	♂ Ashkenazi Jewish: 1/15 ♂ General: 1/112 ♂ Spaniard: Unknown ♂ Turkish: 1/236	87.16% 31.60% 44.29% 59.38%	1/117 1/164 Unknown 1/581
Gitelman Syndrome	♂ European: 1/100 ♂ European Gypsy: Unknown ♂ General: 1/101 ♂ Taiwanese: Unknown	35.00% >99% 30.00% 64.29%	1/154 Unknown 1/144 Unknown
Globoid Cell Leukodystrophy	♂ Dutch: 1/137 ♂ European: 1/150 ♂ Japanese: 1/150	60.98% 26.47% 36.00%	1/351 1/204 1/234
Glutaric Acidemia: Type I	♂ European: 1/164 ♂ General: 1/164 ♂ US Amish: 1/12	57.78% 25.51% >99%	1/388 1/220 <1/1200

Disease	Carrier Rate	Detection Rate	Residual Risk
Glutaric Acidemia: Type IIA	♂ General: Unknown	71.43%	Unknown
Glutaric Acidemia: Type IIB	♂ General: Unknown	33.33%	Unknown
Glutaric Acidemia: Type IIC	♂ Taiwanese: Unknown ♂ Turkish: Unknown	>99% 80.00%	Unknown Unknown
Glycine Encephalopathy: AMT Related	♂ General: Unknown	40.91%	Unknown
Glycine Encephalopathy: GLDC Related	♂ Finnish: 1/118 ♂ General: 1/280	78.00% 12.50%	1/536 1/320
Glycogen Storage Disease: Type IA	♂ Ashkenazi Jewish: 1/71 ♂ Chinese: 1/159 ♂ European: 1/177 ♂ Hispanic American: 1/177 ♂ Japanese: 1/177	>99% 80.00% 76.88% 27.78% 89.22%	<1/7100 1/795 1/765 1/245 1/1641
Glycogen Storage Disease: Type IB	♂ Australian: 1/354 ♂ European: 1/354 ♂ Japanese: 1/354	50.00% 45.74% 39.13%	1/708 1/652 1/582
Glycogen Storage Disease: Type II	♂ African American: 1/60 ♂ Chinese: 1/112 ♂ European: 1/97 ♂ North African: Unknown	45.83% 72.00% 51.76% 60.00%	1/111 1/400 1/201 Unknown
Glycogen Storage Disease: Type III	♂ Faroese: 1/30 ♂ General: 1/159 ♂ North African Jewish: 1/35	>99% 39.81% >99%	<1/3000 1/264 <1/3500
Glycogen Storage Disease: Type IV	♂ Ashkenazi Jewish: 1/35 ♂ General: 1/461	>99% 18.60%	<1/3500 1/566
Glycogen Storage Disease: Type V	♂ Caucasus Jewish: Unknown ♂ European: 1/159 ♂ General: Unknown ♂ Spaniard: 1/159 ♂ Yemenite Jewish: Unknown	>99% 60.71% 74.10% 67.11% 75.00%	Unknown 1/405 Unknown 1/483 Unknown
Glycogen Storage Disease: Type VII	♂ Ashkenazi Jewish: 1/250	>99%	<1/25000
Guanidinoacetate Methyltransferase Deficiency	♂ General: Unknown	29.41%	Unknown
HMG-CoA Lyase Deficiency	♂ General: 1/159 ♂ Japanese: Unknown ♂ Portuguese: Unknown ♂ Saudi Arabian: Unknown	40.00% 30.00% 86.36% 93.33%	1/265 Unknown Unknown Unknown
Hemochromatosis: Type 2A: HFE2 Related	♂ European: Unknown ♂ Mediterranean: Unknown	69.23% 72.73%	Unknown Unknown
Hemochromatosis: Type 3: TFR2 Related	♂ Italian: Unknown	73.21%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Hemoglobinopathy: Hb C	♂ African American: 1/51	>99%	<1/5100
Hemoglobinopathy: Hb D	♂ Canadian: 1/64 ♂ Indian: 1/16 ♂ Iranian: 1/11	>99% >99% >99%	<1/6400 <1/1600 <1/1100
Hemoglobinopathy: Hb E	♂ Cambodia: 1/4 ♂ Chinese: 1/13 ♂ Indian: 1/10 ♂ Thai: 1/9	>99% >99% >99% >99%	<1/400 <1/1300 <1/1000 <1/900
Hemoglobinopathy: Hb O	♂ African American: 1/87 ♂ Middle Eastern: Unknown	>99% >99%	<1/8700 Unknown
Hereditary Fructose Intolerance	♂ European: 1/81 ♂ Italian: 1/81 ♂ Slavic: 1/81	72.73% 90.91% >99%	1/297 1/891 <1/8100
Hereditary Spastic Paraplegia: TECPR2 Related	♂ Bukharan Jewish: 1/75	>99%	<1/7500
Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related	♂ Pakistani: Unknown	>99%	Unknown
Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	♂ European: Unknown ♂ General: 1/781	70.00% 52.27%	Unknown 1/1636
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 ♂ European: 1/434	>99% 12.50%	<1/23500 1/496
Hermansky-Pudlak Syndrome: Type 4	♂ European: Unknown	54.17%	Unknown
Holocarboxylase Synthetase Deficiency	♂ European: 1/148 ♂ Japanese: 1/159	83.33% 76.92%	1/888 1/689
Homocystinuria Caused by CBS Deficiency	♂ European: 1/224 ♂ Irish: 1/128 ♂ Italian: 1/224 ♂ Norwegian: 1/41 ♂ Qatari: 1/22 ♂ Saudi Arabian: Unknown	64.29% 70.59% 35.71% 84.38% >99% 92.31%	1/627 1/435 1/348 1/262 <1/2200 Unknown
Hurler Syndrome	♂ Czech: 1/190 ♂ European: 1/194 ♂ General: 1/194 ♂ Italian: 1/194 ♂ Japanese: 1/194 ♂ Moroccan Jewish: 1/194 ♂ Scandinavian: 1/194 ♂ Spaniard: 1/194	52.50% 81.71% 62.50% 61.11% 23.68% 92.31% 79.41% 52.50%	1/400 1/1061 1/517 1/499 1/254 1/2522 1/942 1/408

Disease	Carrier Rate	Detection Rate	Residual Risk
Hypophosphatasia	♂ Canadian Amish: 1/26 ♂ European: 1/159 ♂ Japanese: Unknown	>99% 19.23% 54.55%	<1/2600 1/197 Unknown
Inclusion Body Myopathy: Type 2	♂ General: Unknown ♂ Iranian Jewish: 1/16 ♂ Japanese: Unknown ♂ Korean: Unknown	85.83% >99% 71.88% 72.50%	Unknown <1/1600 Unknown Unknown
Infantile Cerebral and Cerebellar Atrophy	♂ Caucasus Jewish: 1/20	>99%	<1/2000
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%	<1/9200
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 ♂ European: Unknown	>99% 65.22%	Unknown Unknown
Leydig Cell Hypoplasia (Luteinizing Hormone Resistance)	♂ Brazilian: Unknown	>99%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2A	♂ Basque: 1/61 ♂ Croatian: 1/133 ♂ European: 1/103 ♂ General: 1/103 ♂ Italian: 1/162 ♂ Russian: 1/103 ♂ US Amish: Unknown	61.46% 76.00% 17.23% 26.47% 35.71% 53.33% >99%	1/158 1/554 1/124 1/140 1/252 1/221 Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Limb-Girdle Muscular Dystrophy: Type 2B	♂ Caucasus Jewish: 1/25 ♂ Libyan Jewish: 1/19	>99% >99%	<1/2500 <1/1900
Limb-Girdle Muscular Dystrophy: Type 2C	♂ European Gypsy: 1/50 ♂ General: Unknown ♂ Tunisian: Unknown	>99% 60.00% >99%	<1/5000 Unknown Unknown
Limb-Girdle Muscular Dystrophy: Type 2D	♂ Brazilian: Unknown ♂ European: 1/288 ♂ Finnish: 1/150 ♂ General: Unknown	64.29% 22.22% 95.45% 26.09%	Unknown 1/370 1/3300 Unknown
Limb-Girdle Muscular Dystrophy: Type 2E	♂ Brazilian: Unknown ♂ European: 1/539 ♂ General: Unknown ♂ US Amish: Unknown	57.14% 25.00% 12.50% >99%	Unknown 1/719 Unknown Unknown
Limb-Girdle Muscular Dystrophy: Type 2F	♂ Brazilian: Unknown ♂ General: Unknown	>99% 83.33%	Unknown Unknown
Limb-Girdle Muscular Dystrophy: Type 2I	♂ Brazilian: Unknown ♂ Danish: 1/100 ♂ General: Unknown ♂ German: 1/300	34.62% 85.53% 43.18% 82.50%	Unknown 1/691 Unknown 1/1714
Lipoprotein Lipase Deficiency	♂ French Canadian: 1/44 ♂ General: Unknown	28.95% 20.00%	1/62 Unknown
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	♂ European: 1/126 ♂ General: 1/126	88.98% 56.25%	1/1144 1/288
Lysinuric Protein Intolerance	♂ Finnish: 1/123 ♂ Italian: 1/120 ♂ Japanese: 1/115 ♂ North African: Unknown	>99% 45.45% 37.93% >99%	<1/12300 1/220 1/185 Unknown
MTHFR Deficiency: Severe	♂ Bukharan Jewish: 1/39	>99%	<1/3900
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%	<1/9700
Maple Syrup Urine Disease: Type 2	♂ General: 1/481 ♂ Norwegian: 1/481 ♂ Turkish: 1/112	42.31% 50.00% 58.33%	1/834 1/962 1/269
Maple Syrup Urine Disease: Type 3	♂ Ashkenazi Jewish: 1/94 ♂ General: Unknown	>99% 68.75%	<1/9400 Unknown
Maroteaux-Lamy Syndrome	♂ Argentinian: 1/274 ♂ General: 1/388 ♂ Spaniard: 1/274	75.00% 61.54% 29.17%	1/1096 1/1009 1/387
Meckel Syndrome: Type 1	♂ European: 1/212 ♂ Finnish: 1/48	72.22% >99%	1/763 <1/4800
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	♂ European: 1/50 ♂ Saudi Arabian: 1/68 ♂ United Kingdom: 1/51	90.91% 95.00% 90.00%	1/550 1/1360 1/510
Megalencephalic Leukoencephalopathy	♂ Japanese: Unknown ♂ Libyan Jewish: 1/40 ♂ Turkish: Unknown	50.00% >99% 20.00%	Unknown <1/4000 Unknown
Metachromatic Leukodystrophy	♂ European: 1/150 ♂ Habbanite Jewish: 1/5	43.88% 50.00%	1/267 1/10
Methylmalonic Acidemia: MMAA Related	♂ General: 1/274	63.51%	1/751
Methylmalonic Acidemia: MMAB Related	♂ General: 1/396	71.25%	1/1377
Methylmalonic Acidemia: MUT Related	♂ General: 1/177	43.62%	1/314
Methylmalonic Aciduria and Homocystinuria: Type cblC	♂ Chinese: Unknown ♂ General: 1/159 ♂ Italian: Unknown ♂ Portuguese: Unknown	61.39% 65.74% 75.00% 91.18%	Unknown 1/464 Unknown Unknown
Mitochondrial Complex I Deficiency: NDUFS6 Related	♂ Caucasus Jewish: 1/24	>99%	<1/2400
Mitochondrial DNA Depletion Syndrome: MNGIE Type	♂ Ashkenazi Jewish: Unknown ♂ General: Unknown ♂ Iranian Jewish: Unknown	>99% 47.37% >99%	Unknown Unknown 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 ♂ European: 1/257 ♂ Finnish: 1/257 ♂ Latin American: 1/257	85.00% 20.97% 50.00% 36.11%	1/1713 1/325 1/514 1/402
Morquio Syndrome: Type B	♂ European: Unknown	83.33%	Unknown
Mucolipidosis: Type II/III	♂ General: 1/158 ♂ Japanese: 1/252 ♂ Korean: Unknown ♂ Portuguese: 1/176	24.60% 51.25% 30.00% 50.00%	1/210 1/517 Unknown 1/352
Mucolipidosis: Type IV	♂ Ashkenazi Jewish: 1/97	96.15%	1/2522
Multiple Pterygium Syndrome	♂ European: Unknown ♂ Middle Eastern: Unknown ♂ Pakistani: Unknown	41.67% 60.00% 50.00%	Unknown Unknown Unknown
Multiple Sulfatase Deficiency	♂ Ashkenazi Jewish: 1/320 ♂ General: 1/501	95.00% 18.18%	1/6400 1/612

Disease	Carrier Rate	Detection Rate	Residual Risk
Muscle-Eye-Brain Disease	♂ European: Unknown	54.17%	Unknown
	♂ Finnish: 1/112	97.37%	1/4256
	♂ General: Unknown	23.53%	Unknown
	♂ United States: Unknown	25.00%	Unknown
Navajo Neurohepatopathy	♂ Navajo: 1/39	>99%	<1/3900
Nemaline Myopathy: NEB Related	♂ Ashkenazi Jewish: 1/108	>99%	<1/10800
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
Neuronal Ceroid-Lipofuscinosis: CLN5 Related	♂ Finnish: 1/101	>99%	<1/10100
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%	<1/13500
	♂ 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%	1/2436
	♂ 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%	1/2020
Niemann-Pick Disease: Type B	♂ Czech: 1/276	83.33%	1/1656
	♂ 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%	<1/15500
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%	<1/18000
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
	♂ 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%	<1/2900
	♂ Navajo: 1/29	97.22%	1/1044
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%	<1/35000
	♂ 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%	<1/14000
	♂ 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

Disease	Carrier Rate	Detection Rate	Residual Risk
Phenylalanine Hydroxylase Deficiency	♂ Arab: Unknown ♂ Ashkenazi Jewish: 1/224 ♂ Brazilian: 1/71 ♂ Chinese: 1/51 ♂ Cuban: 1/71 ♂ European: 1/51 ♂ French Canadian: 1/80 ♂ Iranian: 1/31 ♂ Korean: 1/51 ♂ Non-Ashkenazi Jewish: Unknown ♂ Slovakian Gypsy: 1/39 ♂ Spanish Gypsy: 1/4 ♂ Taiwanese: Unknown ♂ US Amish: 1/16	46.08% 44.44% 56.41% 76.57% 69.64% 73.00% 76.27% 66.94% 57.58% 63.64% >99% 93.75% 83.10% 86.84%	Unknown 1/403 1/163 1/218 1/234 1/189 1/337 1/94 1/120 Unknown <1/3900 1/64 Unknown 1/122
Polyglandular Autoimmune Syndrome: Type I	♂ Finnish: 1/80 ♂ Iranian Jewish: 1/48 ♂ Italian: Unknown ♂ Norwegian: 1/142 ♂ Sardinians: 1/61 ♂ United Kingdom: Unknown ♂ United States: Unknown	90.48% >99% 27.78% 47.92% 81.82% 70.00% 65.62%	1/840 <1/4800 Unknown 1/273 1/336 Unknown 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%	<1/4200
Pontocerebellar Hypoplasia: TSEN54 Related	♂ European: 1/250	95.65%	1/5750
Pontocerebellar Hypoplasia: VPS53 Related	♂ Moroccan Jewish: 1/37	>99%	<1/3700
Pontocerebellar Hypoplasia: VRK1 Related	♂ Ashkenazi Jewish: 1/225	>99%	<1/22500
Primary Carnitine Deficiency	♂ European: 1/101 ♂ Faroese: 1/9 ♂ General: Unknown	58.33% 53.95% 20.22%	1/242 1/20 Unknown
Primary Ciliary Dyskinesia: DNAI1 Related	♂ European: 1/211	52.38%	1/443
Primary Ciliary Dyskinesia: DNAI2 Related	♂ Ashkenazi Jewish: 1/200	>99%	<1/20000
Primary Congenital Glaucoma	♂ Moroccan: Unknown ♂ Saudi Arabian: 1/23 ♂ Turkish: 1/51	>99% 91.67% 70.59%	Unknown 1/276 1/173
Primary Hyperoxaluria: Type 1	♂ Dutch: 1/174 ♂ General: 1/189	62.12% 52.68%	1/459 1/399
Primary Hyperoxaluria: Type 2	♂ General: Unknown	70.31%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Primary Hyperoxaluria: Type 3	♂ Ashkenazi Jewish: Unknown ♂ European: Unknown	>99% 25.00%	Unknown 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 ♂ Greenlandic Inuit: 1/16 ♂ Japanese: 1/102 ♂ Korean: Unknown ♂ Latin American: 1/182 ♂ Spaniard: 1/182	42.86% 58.33% 78.00% 56.25% 75.00% 52.38%	1/319 1/38 1/464 Unknown 1/728 1/382
Pseudocholinesterase Deficiency	♂ General: 1/33 ♂ Iranian Jewish: 1/9	65.00% >99%	1/94 <1/900
Pycnodysostosis	♂ Danish: Unknown	87.50%	Unknown
Pyruvate Carboxylase Deficiency	♂ General: 1/251 ♂ Native American: 1/10	62.50% >99%	1/669 <1/1000
Pyruvate Dehydrogenase Deficiency	♂ General: Unknown	50.00%	Unknown
Renal Tubular Acidosis and Deafness	♂ Colombian (Antioquia): Unknown	92.86%	Unknown
Retinal Dystrophies: RLBP1 Related	♂ Newfoundlander: 1/106 ♂ Swedish: 1/84	>99% >99%	<1/10600 <1/8400
Retinal Dystrophies: RPE65 Related	♂ Dutch: 1/32 ♂ North African Jewish: Unknown	>99% >99%	<1/3200 Unknown
Retinitis Pigmentosa: CERKL Related	♂ Yemenite Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: DHDDS Related	♂ Ashkenazi Jewish: 1/91	>99%	<1/9100
Retinitis Pigmentosa: FAM161A Related	♂ Ashkenazi Jewish: Unknown ♂ Non-Ashkenazi Jewish: 1/32	>99% >99%	Unknown <1/3200
Rhizomelic Chondrodysplasia Punctata: Type I	♂ General: 1/159	72.68%	1/582
Salla Disease	♂ European: Unknown ♂ Scandinavian: 1/200	33.33% 94.27%	Unknown 1/3491
Sandhoff Disease	♂ Argentinian: Unknown ♂ Cypriot: 1/7 ♂ Italian: Unknown ♂ Spaniard: Unknown	95.45% 80.00% 29.17% 64.29%	Unknown 1/35 Unknown Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Sanfilippo Syndrome: Type A	♂ Australasian: 1/119 ♂ Dutch: 1/78 ♂ European: 1/159 ♂ United States: 1/159	44.12% 63.10% 35.16% 32.14%	1/213 1/211 1/245 1/234
Sanfilippo Syndrome: Type B	♂ Australasian: 1/230 ♂ Dutch: Unknown ♂ European: Unknown ♂ Japanese: 1/200	28.00% 42.31% 52.38% 81.82%	1/319 Unknown Unknown 1/1100
Sanfilippo Syndrome: Type C	♂ Dutch: 1/346 ♂ Greek: 1/415 ♂ Moroccan: Unknown ♂ Spaniard: Unknown	75.00% 25.00% 80.00% 64.29%	1/1384 1/553 Unknown Unknown
Sanfilippo Syndrome: Type D	♂ General: 1/501	83.33%	1/3006
Short-Chain Acyl-CoA Dehydrogenase Deficiency	♂ Ashkenazi Jewish: 1/15	65.00%	1/43
Sickle-Cell Anemia	♂ African American: 1/10 ♂ Hispanic American: 1/95	>99% >99%	<1/1000 <1/9500
Sjogren-Larsson Syndrome	♂ Dutch: Unknown ♂ Swedish: 1/205	25.86% >99%	Unknown <1/20500
Sly Syndrome	♂ General: 1/251	35.71%	1/390
Smith-Lemli-Opitz Syndrome	♂ Brazilian: 1/94 ♂ European: 1/71 ♂ Japanese: Unknown ♂ United States: 1/70	79.17% 84.72% 71.43% 95.00%	1/451 1/465 Unknown 1/1400
Stargardt Disease	♂ General: 1/51	18.05%	1/62
Stuve-Wiedemann Syndrome	♂ Emirati: 1/70 ♂ General: Unknown	>99% 75.00%	<1/7000 Unknown
Sulfate Transporter-Related Osteochondrodysplasia	♂ Finnish: 1/51 ♂ General: 1/100	95.83% 70.00%	1/1224 1/333
Tay-Sachs Disease	♂ Argentinian: 1/280 ♂ Ashkenazi Jewish: 1/29 ♂ Cajun: 1/30 ♂ European: 1/280 ♂ General: 1/280 ♂ Indian: Unknown ♂ Iraqi Jewish: 1/140 ♂ Japanese: 1/127 ♂ Moroccan Jewish: 1/110 ♂ Portuguese: 1/280 ♂ Spaniard: 1/280 ♂ United Kingdom: 1/161	82.35% 99.53% >99% 25.35% 32.09% 85.71% 56.25% 82.81% 22.22% 92.31% 67.65% 71.43%	1/1587 1/6177 <1/3000 1/375 1/412 Unknown 1/320 1/739 1/141 1/3640 1/865 1/564
Trichohepatoenteric Syndrome: Type 1	♂ European: 1/434 ♂ South Asian: 1/434	42.86% 66.67%	1/760 1/1302

Disease	Carrier Rate	Detection Rate	Residual Risk
Tyrosine Hydroxylase Deficiency	♂ General: Unknown	36.11%	Unknown
Tyrosinemia: Type I	♂ Ashkenazi Jewish: 1/158 ♂ European: 1/166 ♂ Finnish: 1/123 ♂ French Canadian: 1/64 ♂ Pakistani: Unknown	>99% 57.14% 97.22% 96.30% 92.86%	<1/15800 1/387 1/4428 1/1728 Unknown
Tyrosinemia: Type II	♂ General: 1/251	40.00%	1/418
Usher Syndrome: Type 1B	♂ European: 1/166 ♂ General: 1/143 ♂ North African: Unknown ♂ Spaniard: 1/152	39.29% 12.89% 66.67% 12.16%	1/273 1/164 Unknown 1/173
Usher Syndrome: Type 1C	♂ Acadian: 1/82 ♂ French Canadian: 1/227	98.86% 83.33%	1/7216 1/1362
Usher Syndrome: Type 1D	♂ General: 1/296	24.39%	1/391
Usher Syndrome: Type 1F	♂ Ashkenazi Jewish: 1/126	93.75%	1/2016
Usher Syndrome: Type 2A	♂ Chinese: Unknown ♂ European: 1/136 ♂ French Canadian: Unknown ♂ General: 1/136 ♂ Japanese: Unknown ♂ Non-Ashkenazi Jewish: Unknown ♂ Scandinavian: 1/125 ♂ Spaniard: 1/133	83.33% 40.00% 66.67% 46.92% 55.56% 61.11% 39.22% 39.02%	Unknown 1/227 Unknown 1/256 Unknown Unknown 1/206 1/218
Usher Syndrome: Type 3	♂ Ashkenazi Jewish: 1/120 ♂ Finnish: 1/134	>99% >99%	<1/12000 <1/13400
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	♂ General: 1/87	65.28%	1/251
Walker-Warburg Syndrome	♂ Ashkenazi Jewish: 1/150	>99%	<1/15000
Werner Syndrome	♂ General: 1/224 ♂ Japanese: 1/87	31.25% 65.62%	1/326 1/253
Wilson Disease	♂ Ashkenazi Jewish: 1/100 ♂ Canarian: 1/26 ♂ Chinese: 1/51 ♂ Cuban: Unknown ♂ European: 1/93 ♂ Greek: 1/90 ♂ Korean: 1/88 ♂ Spaniard: 1/93	>99% 68.75% 55.97% 22.22% 41.64% 44.94% 51.53% 38.18%	<1/10000 1/83 1/116 Unknown 1/159 1/163 1/182 1/150
Wolcott-Rallison Syndrome	♂ Saudi Arabian: Unknown	66.67%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Wolman Disease	♂ Iranian Jewish: 1/33	>99%	<1/3300
Xeroderma Pigmentosum: Group A	♂ Japanese: 1/75 ♂ North African: Unknown ♂ Tunisian: 1/112	97.62% 87.50% 90.91%	1/3150 Unknown 1/1232
Xeroderma Pigmentosum: Group C	♂ Moroccan: 1/71 ♂ Tunisian: 1/51	76.19% >99%	1/298 <1/5100
Zellweger Spectrum Disorders: PEX1 Related	♂ European: 1/139 ♂ General: 1/139	70.27% 67.84%	1/468 1/432
Zellweger Spectrum Disorders: PEX10 Related	♂ Japanese: Unknown	40.74%	Unknown
Zellweger Spectrum Disorders: PEX2 Related	♂ Ashkenazi Jewish: 1/123	>99%	<1/12300
Zellweger Spectrum Disorders: PEX6 Related	♂ General: 1/288	30.00%	1/411

Patient Information
Name: Donor 5623
Date of Birth: [redacted]
Sema4 ID: [redacted]
Client ID: [redacted]
Indication: Carrier Screening

Specimen Information
Specimen Type: Purified DNA
Date Collected: 01/20/2022
Date Received: 01/28/2022
Final Report: 02/09/2022

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

Custom Carrier Screen (3 genes)

with Personalized Residual Risk

SUMMARY OF RESULTS AND RECOMMENDATIONS

⊖ **Negative**

Negative for all genes tested: ***ALG6, GJB2, and LAMA2***
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.

Yan Bai

Yan Bai, Ph.D., FACMG, 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 Disorder of Glycosylation, Type Ic	*ALG6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
	Congenital Muscular Dystrophy (*LAMA2*-Related)	*LAMA2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 640
	Non-Syndromic Hearing Loss (*GJB2*-Related)	*GJB2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 600

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$^{\mathrm{TM}}$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.

SELECTED REFERENCES

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Fragile X syndrome:

Chen L et al. An information-rich CGG repeat primed PCR that detects the full range of Fragile X expanded alleles and minimizes the need for Southern blot analysis. *J Mol Diag* 2010 12:589-600.

Spinal Muscular Atrophy:

Luo M et al. An Ashkenazi Jewish SMN1 haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy. *Genet Med.* 2014 16:149-56.

Ashkenazi Jewish Disorders:

Scott SA et al. Experience with carrier screening and prenatal diagnosis for sixteen Ashkenazi Jewish Genetic Diseases. *Hum. Mutat.* 2010 31:1-11.

Duchenne Muscular Dystrophy:

Flanigan KM et al. Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. *Hum Mutat.* 2009 30:1657-66.

Variant Classification:

Richards S et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 May;17(5):405-24

Additional disease-specific references available upon request.

Patient Information
Name: Donor 5623
Date of Birth: [redacted]
Sema4 ID: [redacted]
Client ID: [redacted]
Indication: Carrier Screening

Specimen Information
Specimen Type: Purified DNA
Date Collected: 01/20/2022
Date Received: 01/28/2022
Final Report: 06/28/2022

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

Unmask Additional Gene(s) (1 gene)

with Personalized Residual Risk

SUMMARY OF RESULTS AND RECOMMENDATIONS

⊖ **Negative**

Negative for all genes tested: ***LDLR***
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. Please note that residual risks for X-linked diseases (including full repeat expansions for Fragile X syndrome) may not be accurate for males and the actual residual risk is likely to be lower.

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**				
	Familial Hypercholesterolemia	*LDLR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 280

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.

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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 $\Delta\Delta$Ct 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.