Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 5621

Genetic Testing Summary

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

Last Updated: 07/27/18

Donor Reported Ancestry: Pakistani

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: Glycogen Storage Disease: Type 3 (AGL) Carrier: Lamellar Ichthyosis: Type 1 (TGM1) Carrier: Primary Carnitine Deficiency (SLC22A5) Negative for other genes sequenced	Carrier testing recommended for those using this donor

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

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

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

CarrierMap℠

Ordering Practice

Practice Code: [redacted]
Fairfax Cryobank - [redacted]
[redacted]
Physician: [redacted]
Report Generated: 2018-06-28

Donor 5621

DOB: [redacted]
Gender: Male
Ethnicity: South Asian
Procedure ID: 106,508
Kit Barcode: [redacted]
Specimen: Blood, #108,002
Specimen Collection: 2017-10-19
Specimen Received: 2017-10-20
Specimen Analyzed: 2018-06-27

TEST INFORMATION

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

Partner Not Tested

SUMMARY OF RESULTS: MUTATION(S) IDENTIFIED

Disease	Donor 5621	Partner Not Tested
Glycogen Storage Disease: Type III (AGL) ○ High Impact ○ Treatment Benefits	Carrier (1 abnormal copy) Mutation: c.1858_1859delCT (p.L620fs) Method: Sequencing **Reproductive Risk & Next Steps:** Reproductive risk detected. Consider partner testing.	
Lamellar Ichthyosis: Type 1 (TGM1) ○ High Impact ○ Treatment Benefits	Carrier (1 abnormal copy) Mutation: c.652G>A (p.G218S) Method: Sequencing **Reproductive Risk & Next Steps:** Reproductive risk detected. Consider partner testing.	
Primary Carnitine Deficiency (SLC22A5) ○ High Impact ○ Treatment Benefits	Carrier (1 abnormal copy) Mutation: c.248G>T (p.R83L) 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 .

ADDITIONAL RESULTS

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

	Donor 5621	Partner Not Tested
CFTR Results	No Mutations Detected Method: Sequencing & Genotyping Interpretation: **NORMAL**	
SMN1 Copy Number † *Spinal Muscular Atrophy*	SMN1 Copy Number: 2 or more copies Method: dPCR & Genotyping 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.

Glycogen Storage Disease: Type III

Glycogen storage disease III causes a buildup of a complex sugar called glycogen in the body's cells. In this disease, the AGL gene responsible for breaking down glycogen is defective. As a result, abnormal glycogen is stored to toxic levels throughout the body, particularly damaging the liver and muscles. Affected patients often have slow growth because of their liver problems, which can lead to short stature. In a small percentage of patients, noncancerous (benign) tumors called adenomas may form in the liver. By childhood, patients develop muscle weakness that can become severe by adulthood. Heart failure is not common in patients with this disease but some do exhibit heart muscle weakness.

High Impact

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

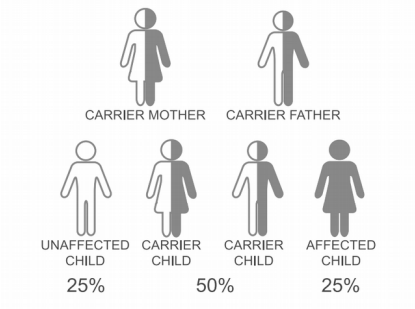
Treatment Benefits

Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.

Clinical Information

✓	Physical Impairment
	Cognitive Impairment
	Shortened Lifespan
	Effective Treatment

Inheritance: Autosomal Recessive



Prognosis

Prognosis is generally mild. Muscle weakness is progressive for GSD III but the progression is very slow. Most symptoms can be avoided with good metabolic control. With proper management, life expectancy is not impacted.

Treatment

Treatment primarily consists of a high protein diet with cornstarch supplementation. Intravenous infusion of dextrose is used to treat acute metabolic incidents. Overall, treatment is very effective in the management of GSD III.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Faroese	>99%	1/30	<1/3000
General	39.81%	1/159	1/264
North African Jewish	>99%	1/35	<1/3500

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

Lamellar Ichthyosis: Type 1

Lamellar Ichthyosis: Type 1 is a condition that mainly affects the skin. It is caused by mutations in the TGM1 gene, a gene responsible for the makeup of the outermost layer of skin (epidermis). Infants with this condition are typically born a collodion membrane (tight, clear membrane) covering their skin, which is shed during the first few weeks of life. After this layer is shed, they develop large, dark, plate-like scales covering their skin. They may also have hair loss (alopecia), decreased ability to sweat (hypohidrosis), eyelid abnormalities, and a thickening of the skin on the palms of the hands and soles of the feet (keratoderma). Infants may develop infections, dehydration, and respiratory problems as a result of the condition.

High Impact

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

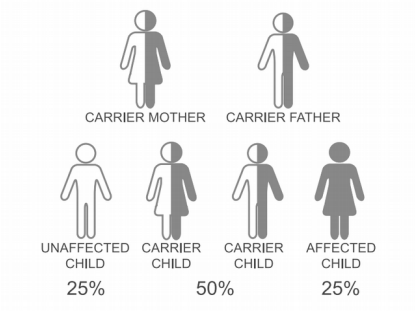
Treatment Benefits

Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.

Clinical Information

✓	Physical Impairment
	Cognitive Impairment
	Shortened Lifespan
	Effective Treatment

Inheritance: Autosomal Recessive



Prognosis

Prognosis for Lamellar Ichthyosis: Type 1 is variable. During the neonatal period, there is a high risk of sepsis but the disease often remains stable over time, with periods of exacerbation. Life expectancy is normal but the disease has a high impact on the quality of life.

Treatment

Treatment for Lamellar Ichthyosis: Type 1 includes daily applications of emollients or keratolytics (moisturizers and skin softeners). During the neonatal period, hygienic handling is very important to prevent infection. Oral retinoids are also useful in more severe forms; however, there are significant side effects, such as bone toxicity, that should be considered when used long-term.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Norwegian	81.40%	1/151	1/812

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

Primary Carnitine Deficiency

Primary Carnitine Deficiency is caused by mutations in the SLC22A5 gene. Carnitine is a natural substance that is acquired through the diet and diminished levels prevent the body from processing certain fats to produce energy. The disease has a variable spectrum of symptoms, from a severe infantile presentation to completely asymptomatic adults. In affected infants, periods of fasting or illness can trigger symptoms, which include low blood sugar, lethargy, and irritability. These symptoms must be treated immediately or there is risk of coma and death. Affected individuals who present during childhood can have dilated cardiomyopathy and skeletal muscle weakness. These affected children can die from cardiac failure if not treated promptly. Some affected individuals can be mostly asymptomatic throughout their lives, though there is still risk for cardiac symptoms.

High Impact

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

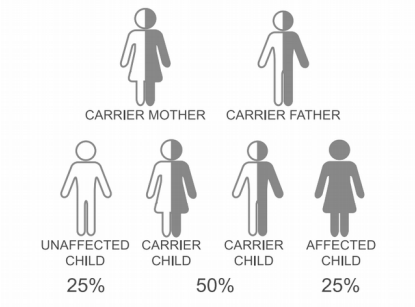
Treatment Benefits

Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.

Clinical Information

✓	Physical Impairment
	Cognitive Impairment
✓	Shortened Lifespan
✓	Effective Treatment

Inheritance: Autosomal Recessive



Prognosis

Prognosis is good with treatment but can be extremely poor if left untreated.

Treatment

L-carnitine supplementation is the preferred method of treatment for this disorder. Individuals respond well to treatment if started before irreversible organ damage occurs and there are relatively few side effects.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
European	58.33%	1/101	1/242
Faroese	53.95%	1/9	1/20
General	20.22%	Unknown	Unknown

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_V1024delIT), 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	80.00%	1/1370
	♂ Italian: 1/224	97.73%	1/9856
	♂ North African: 1/159	>99%	<1/15900
Ataxia-Telangiectasia	♂ Costa Rican: 1/100	68.52%	1/318
	♂ North African Jewish: 1/81	96.97%	1/2673
	♂ Norwegian: 1/197	50.00%	1/394
	♂ Sardinians: Unknown	85.71%	Unknown
	♂ US Amish: Unknown	>99%	Unknown
Autosomal Recessive Polycystic Kidney Disease	♂ Finnish: 1/45	84.21%	1/285
	♂ French: 1/71	62.50%	1/189
	♂ General: 1/71	37.11%	1/113
Bardet-Biedl Syndrome: BBS1 Related	♂ General: 1/376	70.27%	1/1265
	♂ Northern European: 1/376	85.90%	1/2666
	♂ Puerto Rican: Unknown	90.00%	Unknown
Bardet-Biedl Syndrome: BBS10 Related	♂ General: 1/404	47.79%	1/774
Bardet-Biedl Syndrome: BBS11 Related	♂ Bedouin: 1/59	>99%	<1/5900
Bardet-Biedl Syndrome: BBS12 Related	♂ General: Unknown	50.00%	Unknown
Bardet-Biedl Syndrome: BBS2 Related	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: 1/638	38.46%	1/1037
	♂ Middle Eastern: Unknown	>99%	Unknown
Bare Lymphocyte Syndrome: Type II	♂ General: Unknown	66.67%	Unknown
Bartter Syndrome: Type 4A	♂ General: 1/457	81.82%	1/2514
Beta Thalassemia	♂ African American: 1/75	84.21%	1/475
	♂ Indian: 1/24	74.12%	1/93
	♂ Sardinians: 1/23	97.14%	1/804
	♂ Spaniard: 1/51	93.10%	1/740
Beta-Hexosaminidase Pseudodeficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: Unknown	>99%	Unknown
Beta-Ketothiolase Deficiency	♂ Japanese: Unknown	58.33%	Unknown
	♂ Spaniard: Unknown	90.00%	Unknown
Biotinidase Deficiency	♂ General: 1/123	78.32%	1/567

Disease	Carrier Rate	Detection Rate	Residual Risk
Bloom Syndrome	♂ Ashkenazi Jewish: 1/134	96.67%	1/4020
	♂ European: Unknown	66.22%	Unknown
	♂ Japanese: Unknown	50.00%	Unknown
Canavan Disease	♂ Ashkenazi Jewish: 1/55	98.86%	1/4840
	♂ European: Unknown	53.23%	Unknown
Carnitine Palmitoyltransferase IA Deficiency	♂ General: Unknown	38.89%	Unknown
	♂ Hutterite: 1/16	>99%	<1/1600
	♂ Japanese: 1/101	66.67%	1/303
Carnitine Palmitoyltransferase II Deficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ General: Unknown	71.43%	Unknown
Carnitine-Acylcarnitine Translocase Deficiency	♂ Asian: Unknown	95.45%	Unknown
	♂ General: Unknown	18.75%	Unknown
Carpenter Syndrome	♂ Brazilian: Unknown	40.00%	Unknown
	♂ Northern European: Unknown	85.00%	Unknown
Cartilage-Hair Hypoplasia	♂ Finnish: 1/76	93.33%	1/1140
	♂ US Amish: 1/19	>99%	<1/1900
Cerebrotendinous Xanthomatosis	♂ Dutch: Unknown	78.57%	Unknown
	♂ Italian: Unknown	45.95%	Unknown
	♂ Japanese: Unknown	92.86%	Unknown
	♂ Moroccan Jewish: 1/6	87.50%	1/48
Chediak-Higashi Syndrome	♂ General: Unknown	19.64%	Unknown
Cholesteryl Ester Storage Disease	♂ General: 1/101	68.97%	1/325
Choreoacanthocytosis	♂ Ashkenazi Jewish: Unknown	66.67%	Unknown
Chronic Granulomatous Disease: CYBA Related	♂ Iranian: Unknown	71.43%	Unknown
	♂ Japanese: 1/274	>99%	<1/27400
	♂ Korean: 1/105	>99%	<1/10500
	♂ Moroccan Jewish: 1/234	>99%	<1/23400
Citrin Deficiency	♂ Japanese: 1/70	>99%	<1/7000
Citrullinemia: Type I	♂ European: 1/120	18.18%	1/147
	♂ General: 1/120	52.27%	1/251
	♂ Japanese: Unknown	64.71%	Unknown
	♂ Mediterranean: 1/120	50.00%	1/240
Classical Galactosemia	♂ African American: 1/78	73.13%	1/290
	♂ Ashkenazi Jewish: 1/127	>99%	<1/12700
	♂ Dutch: 1/91	75.47%	1/371
	♂ European: 1/112	88.33%	1/960
	♂ General: 1/125	80.00%	1/625
	♂ Irish: 1/76	91.30%	1/874
	♂ Irish Travellers: 1/14	>99%	<1/1400
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	19.05%	Unknown
	♂ Finnish: 1/140	67.24%	1/427
	♂ US Amish: 1/12	>99%	<1/1200
Combined Pituitary Hormone Deficiency: PROP1 Related	♂ European: 1/45	93.29%	1/671
	♂ General: 1/45	82.35%	1/255
Congenital Disorder of Glycosylation: Type 1A: PMM2 Related	♂ Danish: 1/71	90.00%	1/710
	♂ Dutch: 1/68	39.29%	1/112
	♂ European: 1/71	55.33%	1/159
Congenital Disorder of Glycosylation: Type 1B: MPI Related	♂ French: Unknown	54.17%	Unknown
Congenital Disorder of Glycosylation: Type 1C: ALG6 Related	♂ French: Unknown	59.09%	Unknown
	♂ General: Unknown	86.21%	Unknown
Congenital Ichthyosis: ABCA12 Related	♂ North African: Unknown	>99%	Unknown
	♂ South Asian: Unknown	66.67%	Unknown
Congenital Insensitivity to Pain with Anhidrosis	♂ Japanese: Unknown	56.52%	Unknown
	♂ Moroccan Jewish: Unknown	>99%	Unknown
Congenital Lipoid Adrenal Hyperplasia	♂ Japanese: 1/201	51.11%	1/411
	♂ Korean: 1/251	63.64%	1/690
Congenital Myasthenic Syndrome: CHRNE Related	♂ European Gypsy: 1/26	>99%	<1/2600
	♂ North African: Unknown	60.87%	Unknown
Congenital Myasthenic Syndrome: DOK7 Related	♂ European: 1/472	19.05%	1/583
	♂ General: 1/472	18.75%	1/581
Congenital Myasthenic Syndrome: RAPSN Related	♂ General: 1/437	88.57%	1/3824
	♂ Non-Ashkenazi Jewish: Unknown	>99%	Unknown
Congenital Neutropenia: Recessive	♂ English: Unknown	11.76%	Unknown
	♂ Japanese: Unknown	22.22%	Unknown
	♂ Turkish: Unknown	89.47%	Unknown
Corneal Dystrophy and Perceptive Deafness	♂ General: Unknown	71.43%	Unknown
Corticosterone Methyloxidase Deficiency	♂ Iranian Jewish: 1/32	>99%	<1/3200
Crigler-Najjar Syndrome	♂ Sardinians: Unknown	80.00%	Unknown
	♂ Tunisian: Unknown	>99%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Cystic Fibrosis	♂ African American: 1/62	69.99%	1/207
	♂ Ashkenazi Jewish: 1/23	96.81%	1/721
	♂ Asian: 1/94	65.81%	1/275
	♂ European: 1/25	94.96%	1/496
	♂ Hispanic American: 1/48	77.32%	1/212
	♂ Native American: 1/53	84.34%	1/338
Cystinosis	♂ Dutch: 1/194	73.08%	1/721
	♂ French Canadian: 1/40	75.00%	1/160
	♂ General: 1/194	54.51%	1/426
Cystinuria: Non-Type I	♂ European: 1/42	61.11%	1/108
	♂ General: 1/42	37.50%	1/67
	♂ Libyan Jewish: 1/26	93.48%	1/399
	♂ United States: 1/42	56.25%	1/96
Cystinuria: Type I	♂ European: 1/42	46.67%	1/79
	♂ Swedish: 1/159	55.88%	1/360
D-Bifunctional Protein Deficiency	♂ General: 1/159	38.64%	1/259
Diabetes: Recessive Permanent Neonatal	♂ General: Unknown	25.00%	Unknown
Du Pan Syndrome	♂ Pakistani: Unknown	>99%	Unknown
Dyskeratosis Congenita: RTEL1 Related	♂ Ashkenazi Jewish: 1/203	>99%	<1/20300
	♂ General: 1/501	50.00%	1/1002
Dystrophic Epidermolysis Bullosa: Recessive	♂ Italian: Unknown	45.00%	Unknown
	♂ Mexican American: 1/345	56.25%	1/789
Ehlers-Danlos Syndrome: Type VIIC	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
Ellis-van Creveld Syndrome: EVC Related	♂ General: 1/123	32.14%	1/181
Ellis-van Creveld Syndrome: EVC2 Related	♂ General: Unknown	<10%	Unknown
Enhanced S-Cone	♂ Ashkenazi Jewish: Unknown	90.48%	Unknown
	♂ General: Unknown	52.50%	Unknown
Ethylmalonic Aciduria	♂ Arab/Mediterranean: Unknown	29.17%	Unknown
	♂ General: Unknown	38.24%	Unknown
Familial Chloride Diarrhea	♂ Finnish: 1/51	>99%	<1/5100
	♂ Kuwaiti: 1/38	90.00%	1/380
	♂ Polish: 1/224	45.24%	1/409
	♂ Saudi Arabian: 1/38	>99%	<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
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	>99%	<1/6400
	♂ Indian: 1/16	>99%	<1/1600
	♂ Iranian: 1/11	>99%	<1/1100
Hemoglobinopathy: Hb E	♂ Cambodia: 1/4	>99%	<1/400
	♂ Chinese: 1/13	>99%	<1/1300
	♂ Indian: 1/10	>99%	<1/1000
	♂ Thai: 1/9	>99%	<1/900
Hemoglobinopathy: Hb O	♂ African American: 1/87	>99%	<1/8700
	♂ Middle Eastern: Unknown	>99%	Unknown
Hereditary Fructose Intolerance	♂ European: 1/81	72.73%	1/297
	♂ Italian: 1/81	90.91%	1/891
	♂ Slavic: 1/81	>99%	<1/8100
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	70.00%	Unknown
	♂ General: 1/781	52.27%	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	>99%	<1/23500
	♂ European: 1/434	12.50%	1/496
Hermansky-Pudlak Syndrome: Type 4	♂ European: Unknown	54.17%	Unknown
Holocarboxylase Synthetase Deficiency	♂ European: 1/148	83.33%	1/888
	♂ Japanese: 1/159	76.92%	1/689
Homocystinuria Caused by CBS Deficiency	♂ European: 1/224	64.29%	1/627
	♂ Irish: 1/128	70.59%	1/435
	♂ Italian: 1/224	35.71%	1/348
	♂ Norwegian: 1/41	84.38%	1/262
	♂ Qatari: 1/22	>99%	<1/2200
	♂ Saudi Arabian: Unknown	92.31%	Unknown
Hurler Syndrome	♂ Czech: 1/190	52.50%	1/400
	♂ European: 1/194	81.71%	1/1061
	♂ General: 1/194	62.50%	1/517
	♂ Italian: 1/194	61.11%	1/499
	♂ Japanese: 1/194	23.68%	1/254
	♂ Moroccan Jewish: 1/194	92.31%	1/2522
	♂ Scandinavian: 1/194	79.41%	1/942
	♂ Spaniard: 1/194	52.50%	1/408

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

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

Disease	Carrier Rate	Detection Rate	Residual Risk
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

Disease	Carrier Rate	Detection Rate	Residual Risk
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	46.08%	Unknown
	♂ Ashkenazi Jewish: 1/224	44.44%	1/403
	♂ Brazilian: 1/71	56.41%	1/163
	♂ Chinese: 1/51	76.57%	1/218
	♂ Cuban: 1/71	69.64%	1/234
	♂ European: 1/51	73.00%	1/189
	♂ French Canadian: 1/80	76.27%	1/337
	♂ Iranian: 1/31	66.94%	1/94
	♂ Korean: 1/51	57.58%	1/120
	♂ Non-Ashkenazi Jewish: Unknown	63.64%	Unknown
		>99%	<1/3900
	♂ Slovakian Gypsy: 1/39	93.75%	1/64
	♂ Spanish Gypsy: 1/4	83.10%	Unknown
	♂ Taiwanese: Unknown	86.84%	1/122
	♂ US Amish: 1/16		
Polyglandular Autoimmune Syndrome: Type I	♂ Finnish: 1/80	90.48%	1/840
	♂ Iranian Jewish: 1/48	>99%	<1/4800
	♂ Italian: Unknown	27.78%	Unknown
	♂ Norwegian: 1/142	47.92%	1/273
	♂ Sardinians: 1/61	81.82%	1/336
	♂ United Kingdom: Unknown	70.00%	Unknown
	♂ United States: Unknown	65.62%	Unknown
Pontocerebellar Hypoplasia: EXOSC3 Related	♂ General: Unknown	83.33%	Unknown
Pontocerebellar Hypoplasia: RARS2 Related	♂ Sephardic Jewish: Unknown	>99%	Unknown
Pontocerebellar Hypoplasia: SEPSECS Related	♂ Iraqi Jewish: 1/42	>99%	<1/4200
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	58.33%	1/242
	♂ Faroese: 1/9	53.95%	1/20
	♂ General: Unknown	20.22%	Unknown
Primary Ciliary Dyskinesia: DNAI1 Related	♂ European: 1/211	52.38%	1/443
Primary Ciliary Dyskinesia: DNAI2 Related	♂ Ashkenazi Jewish: 1/200	>99%	<1/20000
Primary Congenital Glaucoma	♂ Moroccan: Unknown	>99%	Unknown
	♂ Saudi Arabian: 1/23	91.67%	1/276
	♂ Turkish: 1/51	70.59%	1/173
Primary Hyperoxaluria: Type 1	♂ Dutch: 1/174	62.12%	1/459
	♂ General: 1/189	52.68%	1/399
Primary Hyperoxaluria: Type 2	♂ General: Unknown	70.31%	Unknown
Primary Hyperoxaluria: Type 3	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ European: Unknown	25.00%	Unknown
Progressive Familial Intrahepatic Cholestasis: Type 2	♂ European: Unknown	33.33%	Unknown
Propionic Acidemia: PCCA Related	♂ Japanese: 1/102	86.67%	1/765
Propionic Acidemia: PCCB Related	♂ General: 1/182	42.86%	1/319
	♂ Greenlandic Inuit: 1/16	58.33%	1/38
	♂ Japanese: 1/102	78.00%	1/464
	♂ Korean: Unknown	56.25%	Unknown
	♂ Latin American: 1/182	75.00%	1/728
	♂ Spaniard: 1/182	52.38%	1/382
Pseudocholinesterase Deficiency	♂ General: 1/33	65.00%	1/94
	♂ Iranian Jewish: 1/9	>99%	<1/900
Pycnodysostosis	♂ Danish: Unknown	87.50%	Unknown
Pyruvate Carboxylase Deficiency	♂ General: 1/251	62.50%	1/669
	♂ Native American: 1/10	>99%	<1/1000
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	>99%	<1/10600
	♂ Swedish: 1/84	>99%	<1/8400
Retinal Dystrophies: RPE65 Related	♂ Dutch: 1/32	>99%	<1/3200
	♂ North African Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: CERKL Related	♂ Yemenite Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: DHDDS Related	♂ Ashkenazi Jewish: 1/91	>99%	<1/9100
Retinitis Pigmentosa: FAM161A Related	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ Non-Ashkenazi Jewish: 1/32	>99%	<1/3200
Rhizomelic Chondrodysplasia Punctata: Type I	♂ General: 1/159	72.68%	1/582
Salla Disease	♂ European: Unknown	33.33%	Unknown
	♂ Scandinavian: 1/200	94.27%	1/3491
Sandhoff Disease	♂ Argentinian: Unknown	95.45%	Unknown
	♂ Cypriot: 1/7	80.00%	1/35
	♂ Italian: Unknown	29.17%	Unknown
	♂ Spaniard: Unknown	64.29%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk
Sanfilippo Syndrome: Type A	♂ Australasian: 1/119	44.12%	1/213
	♂ Dutch: 1/78	63.10%	1/211
	♂ European: 1/159	35.16%	1/245
	♂ United States: 1/159	32.14%	1/234
Sanfilippo Syndrome: Type B	♂ Australasian: 1/230	28.00%	1/319
	♂ Dutch: Unknown	42.31%	Unknown
	♂ European: Unknown	52.38%	Unknown
	♂ Japanese: 1/200	81.82%	1/1100
Sanfilippo Syndrome: Type C	♂ Dutch: 1/346	75.00%	1/1384
	♂ Greek: 1/415	25.00%	1/553
	♂ Moroccan: Unknown	80.00%	Unknown
	♂ Spaniard: Unknown	64.29%	Unknown
Sanfilippo Syndrome: Type D	♂ General: 1/501	83.33%	1/3006
Short-Chain Acyl-CoA Dehydrogenase Deficiency	♂ Ashkenazi Jewish: 1/15	65.00%	1/43
Sickle-Cell Anemia	♂ African American: 1/10	>99%	<1/1000
	♂ Hispanic American: 1/95	>99%	<1/9500
Sjogren-Larsson Syndrome	♂ Dutch: Unknown	25.86%	Unknown
	♂ Swedish: 1/205	>99%	<1/20500
Sly Syndrome	♂ General: 1/251	35.71%	1/390
Smith-Lemli-Opitz Syndrome	♂ Brazilian: 1/94	79.17%	1/451
	♂ European: 1/71	84.72%	1/465
	♂ Japanese: Unknown	71.43%	Unknown
	♂ United States: 1/70	95.00%	1/1400
Stargardt Disease	♂ General: 1/51	18.05%	1/62
Stuve-Wiedemann Syndrome	♂ Emirati: 1/70	>99%	<1/7000
	♂ General: Unknown	75.00%	Unknown
Sulfate Transporter-Related Osteochondrodysplasia	♂ Finnish: 1/51	95.83%	1/1224
	♂ General: 1/100	70.00%	1/333
Tay-Sachs Disease	♂ Argentinian: 1/280	82.35%	1/1587
	♂ Ashkenazi Jewish: 1/29	99.53%	1/6177
	♂ Cajun: 1/30	>99%	<1/3000
	♂ European: 1/280	25.35%	1/375
	♂ General: 1/280	32.09%	1/412
	♂ Indian: Unknown	85.71%	Unknown
	♂ Iraqi Jewish: 1/140	56.25%	1/320
	♂ Japanese: 1/127	82.81%	1/739
	♂ Moroccan Jewish: 1/110	22.22%	1/141
	♂ Portuguese: 1/280	92.31%	1/3640
	♂ Spaniard: 1/280	67.65%	1/865
	♂ United Kingdom: 1/161	71.43%	1/564
Trichohepatoenteric Syndrome: Type 1	♂ European: 1/434	42.86%	1/760
	♂ South Asian: 1/434	66.67%	1/1302
Tyrosine Hydroxylase Deficiency	♂ General: Unknown	36.11%	Unknown
Tyrosinemia: Type I	♂ Ashkenazi Jewish: 1/158	>99%	<1/15800
	♂ European: 1/166	57.14%	1/387
	♂ Finnish: 1/123	97.22%	1/4428
	♂ French Canadian: 1/64	96.30%	1/1728
	♂ Pakistani: Unknown	92.86%	Unknown
Tyrosinemia: Type II	♂ General: 1/251	40.00%	1/418
Usher Syndrome: Type 1B	♂ European: 1/166	39.29%	1/273
	♂ General: 1/143	12.89%	1/164
	♂ North African: Unknown	66.67%	Unknown
	♂ Spaniard: 1/152	12.16%	1/173
Usher Syndrome: Type 1C	♂ Acadian: 1/82	98.86%	1/7216
	♂ French Canadian: 1/227	83.33%	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	83.33%	Unknown
	♂ European: 1/136	40.00%	1/227
	♂ French Canadian: Unknown	66.67%	Unknown
	♂ General: 1/136	46.92%	1/256
	♂ Japanese: Unknown	55.56%	Unknown
	♂ Non-Ashkenazi Jewish: Unknown	61.11%	Unknown
		39.22%	1/206
	♂ Scandinavian: 1/125	39.02%	1/218
	♂ Spaniard: 1/133		
Usher Syndrome: Type 3	♂ Ashkenazi Jewish: 1/120	>99%	<1/12000
	♂ Finnish: 1/134	>99%	<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	31.25%	1/326
	♂ Japanese: 1/87	65.62%	1/253
Wilson Disease	♂ Ashkenazi Jewish: 1/100	>99%	<1/10000
	♂ Canarian: 1/26	68.75%	1/83
	♂ Chinese: 1/51	55.97%	1/116
	♂ Cuban: Unknown	22.22%	Unknown
	♂ European: 1/93	41.64%	1/159
	♂ Greek: 1/90	44.94%	1/163
	♂ Korean: 1/88	51.53%	1/182
	♂ Spaniard: 1/93	38.18%	1/150
Wolcott-Rallison Syndrome	♂ Saudi Arabian: Unknown	66.67%	Unknown

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