Fairfax Cryobank
The Trusted Choice for Donor Sperm

# Donor 5195

# Genetic Testing Summary

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

Last Updated: 10/31/18

Donor Reported Ancestry: Mexican, Finnish, Swedish  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 | 1/632 |
| Expanded Genetic Disease Testing Panel attached- 289 diseases by gene sequencing | Carrier: Multiple Sulfatase Deficiency (SUMF1)<br><br>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.

Recombine

CarrierMap™

**Ordering Practice:**

**Practice Code:** [redacted]
Fairfax Cryobank - [redacted]
[redacted]
**Physician:** [redacted]
**Report Generated:** 2018-04-04

**Donor 5195**

**DOB:** [redacted]
**Gender:** Male
**Ethnicity:** Latin American and European
**Procedure ID:** 114425
**Kit Barcode:** [redacted]
**Specimen:** Blood, #116931
**Specimen Collection:** 2018-03-13
**Specimen Received:** 2018-03-14
**Specimen Analyzed:** 2018-04-04

**Partner Not Tested**

## TEST INFORMATION

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

## SUMMARY OF RESULTS: MUTATION(S) IDENTIFIED

| Disease | Donor 5195 | Partner Not Tested |
|---|---|---|
| **Multiple Sulfatase Deficiency** (SUMF1)<br>○ High Impact | Carrier (1 abnormal copy)<br>Mutation: c.2T>G<br>Method: Sequencing | |

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

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

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

Assay performed by
Reprogenetics
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Lab Technician: Bo Chu

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

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## ADDITIONAL RESULTS: NO INCREASED REPRODUCTIVE RISK

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

| Disease (Gene) | Donor 5195 | Partner Not Tested |
|---|---|---|
| Spinal Muscular Atrophy: SMN1 Linked (SMN1)* | SMN1 Copy Number: 2 or more copies<br>Method: Genotyping & dPCR | |

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

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

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

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# Multiple Sulfatase Deficiency (SUMF1)

Multiple Sulfatase Deficiency (MSD) is a very rare metabolic disorder in which all of the known sulfatase enzymes are deficient or inoperative. The condition is caused by disease-causing mutations in the SUMF1 gene, which normally creates a protein that plays a role in the breakdown of certain sugars, fats, and hormones. There are three major types of MSD, categorized by age of onset neonatal, late-infantile, and juvenile. The neonatal form is most severe. Symptoms appear soon after birth and include seizures, developmental delay, slow growth, movement problems, ichthyosis (dry, scaly skin), hypertrichosis (excess hair growth), skeletal problems (including scoliosis and joint stiffness), and coarse facial features. Affected children can also have hearing loss, an enlarged liver and spleen, and heart abnormalities. The late-infantile type is the most common form of the condition and shares some of the features seen in the neonatal type, including ichthyosis, skeletal problems, and coarse facial features. Children with the late-infantile type of MSD have normal development in early childhood but then progressively lose their cognitive and movement skills due to brain abnormalities. The juvenile type of MSD is rare and onset of symptoms is in mid- to late-childhood. These children also are typically normal at birth and in early childhood, and then experience a loss of motor and cognitive skills; however, this loss of skills is typically slower. They also experience ichthyosis.

○ High Impact

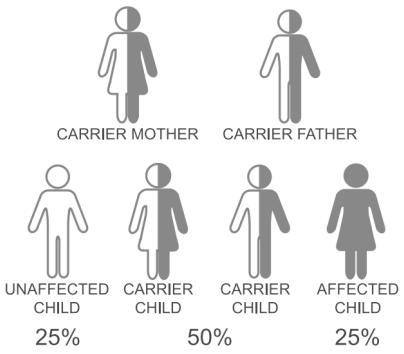
These diseases have a significant impact on life expectancy and quality of life.

## Clinical Information

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

Effective Treatment

## Inheritance:
## Autosomal Recessive




## Prognosis

The prognosis for Multiple Sulfatase Deficiency is very poor. Children rarely live beyond age 10 and some die at younger ages.

## Treatment

There is no curative treatment for MSD at this time. Physiotherapy and hydrotherapy can be useful to help individuals with MSD.

## Risk Information

| Ethnicity | Detection Rate | Pre-Test Risk | Post-Test Risk |
|---|---|---|---|
| Ashkenazi Jewish | 95.00% | 1/320 | 1/6400 |
| General | 18.18% | 1/501 | 1/612 |

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

To learn more, visit recombine.com/diseases/multiple-sulfatase-deficiency

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# Methods and Limitations

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

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

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

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

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

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## Diseases & Mutations Assayed

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Alport Syndrome: COL4A3 Related (COL4A3):** Mutations (3): ♂ Genotyping | c.4420_4424delCTTTT, c.4441C>T (p.R1481X), c.4571C>G (p.S1524X) Sequencing | NM_000091:2-52

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**GM1-Gangliosidoses (GLB1):** Mutations (17): ♂ Genotyping | c.1480-2A>G, c.75+2_75+3insT, c.1772A>G (p.Y591C), c.947A>G (p.Y316C), c.1051C>T (p.R351X),

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c.1369C>T (p.R457X), c.145C>T (p.R49C), c.202C>T (p.R68W), c.245C>T (p.T82M), c.601C>T (p.R201C), c.622C>T (p.R208C), c.1370G>A (p.R457Q), c.176G>A (p.R59H), c.367G>A (p.G123R), c.152T>C (p.I51T), c.1771T>A (p.Y591N), c.1577_1578insG Sequencing | NM_000404:1-16

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Leber Congenital Amaurosis: GUCY2D Related (GUCY2D):** Mutations (3): ♂ Genotyping | c.1694T>C (p.F565S), c.2943delG (p.G982V), c.387delC (p.P130Lfx) Sequencing | NM_000180:2-19

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Medium-Chain Acyl-CoA Dehydrogenase Deficiency (ACADM):** Mutations (8): ♂ Genotyping | c.985A>G (p.K329E), c.362C>T (p.T121I), c.595G>A (p.G199R), c.799G>A (p.G267R), c.199T>C (p.Y67H), c.262C>T (p.L88F), c.616C>T (p.R206C), c.617G>A (p.C206H) Sequencing | NM_001127328:1-12

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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Sequencing | NM_004646:1-29

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Primary Carnitine Deficiency (SLC22A5):** Mutations (12): ♂ Genotyping | c.506G>A

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Tay-Sachs Disease (HEXA):** Mutations (78): ♂ Genotyping | c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.532C>T (p.R178C), c.533G>A (p.R178H), c.805G>A (p.G269S), c.1510C>T (p.R504C), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.1003A>T (p.I335F), c.910_912delTTC (p.305delF), c.749G>A (p.G250D), c.632T>C (p.F211S), c.629C>T (p.S210F), c.613delC, c.611A>G (p.H204R), c.598G>A (p.V200M), c.590A>C (p.K197T), c.571-1G>T, c.540C>G (p.Y180X), c.538T>C (p.Y180H), c.533G>T (p.R178L),

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

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

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

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

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

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

**Usher Syndrome: Type 1C (USH1C):** Mutations (6): ♂ Genotyping | c.496+1G>A, c.238_239insC, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.36+1G>T, c.496+1G>T Sequencing | NM_153676:1-27

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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## Residual Risk Information

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

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

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

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| Disease | Carrier Rate | Detection Rate | Residual Risk |
|---|---|---|---|
| Congenital Neutropenia: Recessive | ♂ English: Unknown | 11.76% | Unknown |
| | ♂ Japanese: Unknown | 22.22% | Unknown |
| | ♂ Turkish: Unknown | 89.47% | Unknown |
| Corneal Dystrophy and Perceptive Deafness | ♂ General: Unknown | 71.43% | Unknown |
| Corticosterone Methyloxidase Deficiency | ♂ Iranian Jewish: 1/32 | >99% | <1/3,200 |
| Crigler-Najjar Syndrome | ♂ Sardinians: Unknown | 80.00% | Unknown |
| | ♂ Tunisian: Unknown | >99% | Unknown |
| Cystic Fibrosis | ♂ African American: 1/62 | 69.99% | 1/207 |
| | ♂ Ashkenazi Jewish: 1/23 | 96.81% | 1/721 |
| | ♂ Asian: 1/94 | 65.42% | 1/272 |
| | ♂ European: 1/25 | 94.96% | 1/496 |
| | ♂ Hispanic American: 1/48 | 77.32% | 1/212 |
| | ♂ Native American: 1/53 | 84.34% | 1/338 |
| Cystinosis | ♂ Dutch: 1/194 | 73.08% | 1/721 |
| | ♂ French Canadian: 1/40 | 75.00% | 1/160 |
| | ♂ General: 1/194 | 54.51% | 1/426 |
| Cystinuria: Non-Type I | ♂ European: 1/42 | 61.11% | 1/108 |
| | ♂ General: 1/42 | 37.50% | 1/67 |
| | ♂ Libyan Jewish: 1/26 | 93.48% | 1/399 |
| | ♂ United States: 1/42 | 56.25% | 1/96 |
| Cystinuria: Type I | ♂ European: 1/42 | 46.67% | 1/79 |
| | ♂ Swedish: 1/159 | 55.88% | 1/360 |
| D-Bifunctional Protein Deficiency | ♂ General: 1/159 | 38.64% | 1/259 |
| Diabetes: Recessive Permanent Neonatal | ♂ General: Unknown | 25.00% | Unknown |
| Du Pan Syndrome | ♂ Pakistani: Unknown | >99% | Unknown |
| Dyskeratosis Congenita: RTEL1 Related | ♂ Ashkenazi Jewish: 1/203 | >99% | <1/20,300 |
| | ♂ General: 1/501 | 50.00% | 1/1,002 |
| Dystrophic Epidermolysis Bullosa: Recessive | ♂ Italian: Unknown | 45.00% | Unknown |
| | ♂ Mexican American: 1/345 | 56.25% | 1/789 |
| Ehlers-Danlos Syndrome: Type VIIC | ♂ Ashkenazi Jewish: Unknown | >99% | Unknown |
| Ellis-van Creveld Syndrome: EVC Related | ♂ General: 1/123 | 32.14% | 1/181 |
| Ellis-van Creveld Syndrome: EVC2 Related | ♂ General: Unknown | <10% | Unknown |
| Enhanced S-Cone | ♂ Ashkenazi Jewish: Unknown | 90.48% | Unknown |
| | ♂ General: Unknown | 52.50% | Unknown |
| Ethylmalonic Aciduria | ♂ Arab/Mediterranean: Unknown | 29.17% | Unknown |
| | ♂ General: Unknown | 38.24% | Unknown |
| Familial Chloride Diarrhea | ♂ Finnish: 1/51 | >99% | <1/5,100 |
| | ♂ Kuwaiti: 1/38 | 90.00% | 1/380 |
| | ♂ Polish: 1/224 | 45.24% | 1/409 |
| | ♂ Saudi Arabian: 1/38 | >99% | <1/3,800 |
| Familial Dysautonomia | ♂ Ashkenazi Jewish: 1/31 | >99% | <1/3,100 |
| Familial Hyperinsulinism: Type 1: ABCC8 Related | ♂ Ashkenazi Jewish: 1/52 | 98.75% | 1/4,160 |
| | ♂ Finnish: 1/101 | 45.16% | 1/184 |
| Familial Hyperinsulinism: Type 2: KCNJ11 Related | ♂ Arab: Unknown | 40.00% | Unknown |
| Familial Mediterranean Fever | ♂ Arab: 1/4 | 51.18% | 1/8 |
| | ♂ Armenian: 1/5 | 94.51% | 1/91 |
| | ♂ Ashkenazi Jewish: 1/81 | 39.52% | 1/134 |
| | ♂ Iraqi Jewish: 1/4 | 76.92% | 1/17 |
| | ♂ Israeli Jewish: 1/5 | 62.26% | 1/13 |
| | ♂ Lebanese: 1/6 | 91.67% | 1/72 |
| | ♂ North African Jewish: 1/5 | 95.69% | 1/116 |
| | ♂ Syrian: 1/6 | 85.14% | 1/40 |
| | ♂ Turkish: 1/5 | 74.25% | 1/19 |
| Fanconi Anemia: Type A | ♂ Moroccan Jewish: 1/100 | >99% | <1/10,000 |
| | ♂ Spanish Gypsy: 1/67 | >99% | <1/6,700 |
| Fanconi Anemia: Type C | ♂ Ashkenazi Jewish: 1/101 | >99% | <1/10,100 |
| | ♂ General: Unknown | 30.00% | Unknown |
| Fanconi Anemia: Type G | ♂ Black South African: 1/101 | 81.82% | 1/556 |
| | ♂ French Canadian: Unknown | 87.50% | Unknown |
| | ♂ Japanese: Unknown | 75.00% | Unknown |
| | ♂ Korean: Unknown | 66.67% | Unknown |
| Fanconi Anemia: Type J | ♂ General: Unknown | 86.36% | Unknown |
| Fumarase Deficiency | ♂ General: Unknown | 30.00% | Unknown |
| GM1-Gangliosidoses | ♂ Eurodescent Brazilian: 1/66 | 62.15% | 1/174 |
| | ♂ European: 1/194 | 50.00% | 1/388 |
| | ♂ General: 1/194 | 20.00% | 1/243 |
| | ♂ Hispanic American: 1/194 | 58.33% | 1/466 |
| | ♂ Japanese: Unknown | 62.82% | Unknown |
| GRACILE Syndrome | ♂ Finnish: 1/109 | 97.22% | 1/3,924 |
| Galactokinase Deficiency | ♂ Japanese: 1/501 | 50.00% | 1/1,002 |
| | ♂ Roma: 1/51 | >99% | <1/5,100 |
| Gaucher Disease | ♂ Ashkenazi Jewish: 1/15 | 87.16% | 1/117 |
| | ♂ General: 1/112 | 31.60% | 1/164 |
| | ♂ Spaniard: Unknown | 44.29% | Unknown |
| | ♂ Turkish: 1/236 | 59.38% | 1/581 |
| Gitelman Syndrome | ♂ European: 1/100 | 35.00% | 1/154 |
| | ♂ European Gypsy: Unknown | >99% | Unknown |
| | ♂ General: 1/101 | 30.00% | 1/144 |
| | ♂ Taiwanese: Unknown | 64.29% | Unknown |

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

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

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

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

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

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

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| Disease | Carrier Rate | Detection Rate | Residual Risk |
|---|---|---|---|
| | ♂ Puerto Rican: Unknown | 91.67% | Unknown |
| Oculocutaneous Albinism: Type 3 | ♂ Black South African: 1/47 | 94.74% | 1/893 |
| Oculocutaneous Albinism: Type 4 | ♂ Japanese: 1/146 | 58.33% | 1/350 |
| Omenn Syndrome: DCLRE1C Related | ♂ Apache: 1/29 | >99% | <1/2,900 |
| | ♂ Navajo: 1/29 | 97.22% | 1/1,044 |
| Omenn Syndrome: RAG2 Related | ♂ Arab: Unknown | 40.00% | Unknown |
| | ♂ Non-Ashkenazi Jewish: Unknown | 70.00% | Unknown |
| Ornithine Translocase Deficiency | ♂ French Canadian: 1/20 | 95.00% | 1/400 |
| | ♂ Italian: Unknown | 18.75% | Unknown |
| | ♂ Japanese: Unknown | 60.00% | Unknown |
| Osteopetrosis: TCIRG1 Related | ♂ Ashkenazi Jewish: 1/350 | >99% | <1/35,000 |
| | ♂ Costa Rican: Unknown | >99% | Unknown |
| | ♂ General: 1/251 | 25.00% | 1/335 |
| POLG Related Disorders: Autosomal Recessive | ♂ Belgian: Unknown | 85.00% | Unknown |
| | ♂ Finnish: 1/140 | >99% | <1/14,000 |
| | ♂ General: Unknown | 93.10% | Unknown |
| | ♂ Norwegian: Unknown | >99% | Unknown |
| Papillon-Lefevre Syndrome | ♂ General: Unknown | 35.29% | Unknown |
| | ♂ Indian Jewish: Unknown | >99% | Unknown |
| | ♂ Turkish: Unknown | 50.00% | Unknown |
| Pendred Syndrome | ♂ European: 1/58 | 42.11% | 1/100 |
| | ♂ Japanese: Unknown | 45.83% | Unknown |
| | ♂ Pakistani: Unknown | 29.82% | Unknown |
| Persistent Mullerian Duct Syndrome: Type I | ♂ General: Unknown | 28.12% | Unknown |
| Persistent Mullerian Duct Syndrome: Type II | ♂ General: Unknown | 78.12% | Unknown |
| Phenylalanine Hydroxylase Deficiency | ♂ Arab: Unknown | 46.08% | Unknown |
| | ♂ Ashkenazi Jewish: 1/224 | 44.44% | 1/403 |
| | ♂ Brazilian: 1/71 | 56.41% | 1/163 |
| | ♂ Chinese: 1/51 | 76.57% | 1/218 |
| | ♂ Cuban: 1/71 | 69.64% | 1/234 |
| | ♂ European: 1/51 | 73.00% | 1/189 |
| | ♂ French Canadian: 1/80 | 76.27% | 1/337 |
| | ♂ Iranian: 1/31 | 66.94% | 1/94 |
| | ♂ Korean: 1/51 | 51.52% | 1/105 |
| | ♂ Non-Ashkenazi Jewish: Unknown | 63.64% | Unknown |
| | ♂ Slovakian Gypsy: 1/39 | >99% | <1/3,900 |
| | ♂ Spanish Gypsy: 1/4 | 93.75% | 1/64 |
| | ♂ Taiwanese: Unknown | 83.10% | Unknown |
| | ♂ US Amish: 1/16 | 86.84% | 1/122 |
| Polyglandular Autoimmune Syndrome: Type I | ♂ Finnish: 1/80 | 90.48% | 1/840 |
| | ♂ Iranian Jewish: 1/48 | >99% | <1/4,800 |
| | ♂ Italian: Unknown | 27.78% | Unknown |
| | ♂ Norwegian: 1/142 | 47.92% | 1/273 |
| | ♂ Sardinians: 1/61 | 81.82% | 1/336 |
| | ♂ United Kingdom: Unknown | 70.00% | Unknown |
| | ♂ United States: Unknown | 65.62% | Unknown |
| Pontocerebellar Hypoplasia: EXOSC3 Related | ♂ General: Unknown | 83.33% | Unknown |
| Pontocerebellar Hypoplasia: RARS2 Related | ♂ Sephardic Jewish: Unknown | >99% | Unknown |
| Pontocerebellar Hypoplasia: SEPSECS Related | ♂ Iraqi Jewish: 1/42 | >99% | <1/4,200 |
| Pontocerebellar Hypoplasia: TSEN54 Related | ♂ European: 1/250 | 95.65% | 1/5,750 |
| Pontocerebellar Hypoplasia: VPS53 Related | ♂ Moroccan Jewish: 1/37 | >99% | <1/3,700 |
| Pontocerebellar Hypoplasia: VRK1 Related | ♂ Ashkenazi Jewish: 1/225 | >99% | <1/22,500 |
| Primary Carnitine Deficiency | ♂ European: 1/101 | 58.33% | 1/242 |
| | ♂ Faroese: 1/9 | 53.95% | 1/20 |
| | ♂ General: Unknown | 20.22% | Unknown |
| Primary Ciliary Dyskinesia: DNAI1 Related | ♂ European: 1/211 | 52.38% | 1/443 |
| Primary Ciliary Dyskinesia: DNAI2 Related | ♂ Ashkenazi Jewish: 1/200 | >99% | <1/20,000 |
| Primary Congenital Glaucoma | ♂ Moroccan: Unknown | >99% | Unknown |
| | ♂ Saudi Arabian: 1/23 | 91.67% | 1/276 |
| | ♂ Turkish: 1/51 | 70.59% | 1/173 |
| Primary Hyperoxaluria: Type 1 | ♂ Dutch: 1/174 | 62.12% | 1/459 |
| | ♂ General: 1/189 | 52.68% | 1/399 |
| Primary Hyperoxaluria: Type 2 | ♂ General: Unknown | 70.31% | Unknown |
| Primary Hyperoxaluria: Type 3 | ♂ Ashkenazi Jewish: Unknown | >99% | Unknown |
| | ♂ European: Unknown | 25.00% | Unknown |
| Progressive Familial Intrahepatic Cholestasis: Type 2 | ♂ European: Unknown | 33.33% | Unknown |
| Propionic Acidemia: PCCA Related | ♂ Japanese: 1/102 | 86.67% | 1/765 |
| Propionic Acidemia: PCCB Related | ♂ General: 1/182 | 42.86% | 1/319 |
| | ♂ Greenlandic Inuit: 1/16 | 58.33% | 1/38 |
| | ♂ Japanese: 1/102 | 78.00% | 1/464 |
| | ♂ Korean: Unknown | 56.25% | Unknown |
| | ♂ Latin American: 1/182 | 75.00% | 1/728 |
| | ♂ Spaniard: 1/182 | 52.38% | 1/382 |
| Pseudocholinesterase Deficiency | ♂ General: 1/33 | 65.00% | 1/94 |
| | ♂ Iranian Jewish: 1/9 | >99% | <1/900 |
| Pycnodysostosis | ♂ Danish: Unknown | 87.50% | Unknown |
| Pyruvate Carboxylase Deficiency | ♂ General: 1/251 | 62.50% | 1/669 |
| | ♂ Native American: 1/10 | >99% | <1/1,000 |
| Pyruvate Dehydrogenase Deficiency | ♂ General: Unknown | 50.00% | Unknown |

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

| Disease | Carrier Rate | Detection Rate | Residual Risk |
|---|---|---|---|
| | ♂ General: Unknown | 75.00% | Unknown |
| Sulfate Transporter-Related Osteochondrodysplasia | ♂ Finnish: 1/51 | 95.83% | 1/1,224 |
| | ♂ General: 1/100 | 70.00% | 1/333 |
| Tay-Sachs Disease | ♂ Argentinian: 1/280 | 82.35% | 1/1,587 |
| | ♂ Ashkenazi Jewish: 1/29 | 99.53% | 1/6,177 |
| | ♂ Cajun: 1/30 | >99% | <1/3,000 |
| | ♂ European: 1/280 | 25.35% | 1/375 |
| | ♂ General: 1/280 | 32.09% | 1/412 |
| | ♂ Indian: Unknown | 85.71% | Unknown |
| | ♂ Iraqi Jewish: 1/140 | 56.25% | 1/320 |
| | ♂ Japanese: 1/127 | 82.81% | 1/739 |
| | ♂ Moroccan Jewish: 1/110 | 22.22% | 1/141 |
| | ♂ Portuguese: 1/280 | 92.31% | 1/3,640 |
| | ♂ Spaniard: 1/280 | 67.65% | 1/865 |
| | ♂ United Kingdom: 1/161 | 71.43% | 1/564 |
| Trichohepatoenteric Syndrome: Type 1 | ♂ European: 1/434 | 42.86% | 1/760 |
| | ♂ South Asian: 1/434 | 66.67% | 1/1,302 |
| Tyrosine Hydroxylase Deficiency | ♂ General: Unknown | 36.11% | Unknown |
| Tyrosinemia: Type I | ♂ Ashkenazi Jewish: 1/158 | >99% | <1/15,800 |
| | ♂ European: 1/166 | 57.14% | 1/387 |
| | ♂ Finnish: 1/123 | 97.22% | 1/4,428 |
| | ♂ French Canadian: 1/64 | 96.30% | 1/1,728 |
| | ♂ Pakistani: Unknown | 92.86% | Unknown |
| Tyrosinemia: Type II | ♂ General: 1/251 | 40.00% | 1/418 |
| Usher Syndrome: Type 1B | ♂ European: 1/166 | 39.29% | 1/273 |
| | ♂ General: 1/143 | 12.89% | 1/164 |
| | ♂ North African: Unknown | 66.67% | Unknown |
| | ♂ Spaniard: 1/152 | 12.16% | 1/173 |
| Usher Syndrome: Type 1C | ♂ Acadian: 1/82 | 98.86% | 1/7,216 |
| | ♂ French Canadian: 1/227 | 83.33% | 1/1,362 |
| Usher Syndrome: Type 1D | ♂ General: 1/296 | 23.17% | 1/385 |
| Usher Syndrome: Type 1F | ♂ Ashkenazi Jewish: 1/126 | 93.75% | 1/2,016 |
| Usher Syndrome: Type 2A | ♂ Chinese: Unknown | 83.33% | Unknown |
| | ♂ European: 1/136 | 40.00% | 1/227 |
| | ♂ French Canadian: Unknown | 66.67% | Unknown |
| | ♂ General: 1/136 | 47.69% | 1/260 |
| | ♂ Japanese: Unknown | 55.56% | Unknown |
| | ♂ Non-Ashkenazi Jewish: Unknown | 61.11% | Unknown |
| | ♂ Scandinavian: 1/125 | 40.52% | 1/210 |
| | ♂ Spaniard: 1/133 | 53.66% | 1/287 |
| Usher Syndrome: Type 3 | ♂ Ashkenazi Jewish: 1/120 | >99% | <1/12,000 |
| | ♂ Finnish: 1/134 | >99% | <1/13,400 |

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

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