

Donor 5427

Genetic Testing Summary

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

Last Updated: 01/14/22

Donor Reported Ancestry: Irish, Italian, English, Scottish, Welsh, Polish, Ukrainian, German 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
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: Cystic Fibrosis (CFTR) Carrier: Congenital Myasthenic Syndrome: RAPSN Related (RAPSN) Carrier: Cystinuria: Type 1 (SLC3A1) Carrier: Nephrotic Syndrome (NPHS2- Realated)/Steroid Resistant Nephrotic Syndrome-Mild variant (NPHS2) Negative for other genes sequenced	Carrier testing recommended for those using this donor
Special Testing		
Genes: CYP21A2, SLC26A4, DHCR7, PPT1	Negative by gene sequencing-see attached for residual risks	

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

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



Ordering Practice:	5427	Partner Not Tested
Practice Code: Fairfax Cryobank - Physician: Report Generated: 2018-02-21	DOB: Gender: Male Ethnicity: European Procedure ID: 91100 Kit Barcode: Specimen: Blood, #92246 Specimen Collection: 2017-04-21 Specimen Received: 2017-04-24 Specimen Analyzed: 2018-02-20	
	TEST INFORMATION Test: CarrierMap ^{SEQ} (Genotyping & Sequencing) Panel: CarrierMap Expanded v3 - Sequencing Diseases Tested: 289 Genes Tested: 278 Genes Sequenced: 273	
SUMMARY OF RESULTS: MU	TATION(S) IDENTIFIED	
Disease	5427	Partner Not Tested
Congenital Myasthenic Syndrome: RAPSN Related (RAPSN) O High Impact	Carrier (1 abnormal copy) Mutation: c.264C>A (p.N88K) Method: Genotyping & Sequencing Reproductive Risk & Next Ste partner testing.	ps : Reproductive risk detected. Consider -
RAPSN Related (RAPSN)	Mutation: c.264C>A (p.N88K) Method: Genotyping & Sequencing Reproductive Risk & Next Ster partner testing. Carrier (1 abnormal copy) Mutation: c.1521_1523delCTT (p.508delF) Method: Genotyping & Sequencing	ps : Reproductive risk detected. Consider ps : Reproductive risk detected. Consider



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

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

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

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



ADDITIONAL RESULTS: NO INCREASED REPRODUCTIVE RISK

The following results <u>are not</u> associated with an increased reproductive risk.

Disease (Gene)	5427	Partner Not Tested
Spinal Muscular Atrophy: SMN1 Linked (SMN1)*	SMN1 Copy Number: 2 or more copies Method: dPCR & Genotyping	

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

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

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

Congenital Myasthenic Syndrome: RAPSN Related (RAPSN)

Congenital myasthenic syndromes (CMS) are a group of conditions affecting neuromuscular function. The disorder is mainly characterized by muscle weakness that worsens with physical exertion. Onset is typically shortly after birth or in early childhood and severity and progression vary. Generally, the earlier symptoms present, the more severe the progression of the disease. CMS caused by mutations in the RAPSN gene is considered postsynaptic; affected infants are typically hypotonic (have low muscle tone) and can experience respiratory failure at birth. Affected infants also typically have episodic apnea (brief stops in breathing) and may have arthrogryposis multiplex congenita, which is a condition associated with multiple joint contractures. Those with late-onset disease typically experience limb weakness in adolescence or adulthood.

O High Impact

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

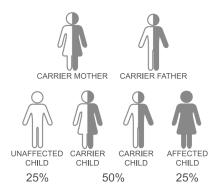
Clinical Information

Physical Impairment
Cognitive Impairment
Shortened Lifespan

Effective Treatment

Inheritance:

Autosomal Recessive



Prognosis

Prognosis for congenital myasthenic syndrome is variable and is dependent on the form and available treatment.

Treatment

Treatment for congenital myasthenic syndrome depends on the form of the disease. Individuals with CHAT-related CMS may respond well to treatment with cholinesterase inhibitors (AChE). Treatment is recommended prophylactically to prevent sudden respiratory insufficiency or apneic attacks provoked by illness.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
General	88.57%	1/437	1/3824
Non-Ashkenazi Jewish	>99%	Unknown	Unknown

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

To learn more, visit recombine.com/diseases/congenital-myasthenic-syndrome-rapsn-related

Carrier Map™

Cystic Fibrosis (CFTR)

Cystic fibrosis (CF) severely affects the respiratory and digestive systems. This disease is caused by mutations in the CFTR gene, which is responsible for controlling the water content of mucus. As a result, mucus glands produce mucus that is overly thick and sticky. In affected individuals, this abnormally thick mucus can obstruct the airways, leading to problems with breathing, as well as bacterial infections in the lungs that can cause permanent lung damage. Most affected individuals also have digestive problems because the thick, sticky mucus blocks the ducts of the pancreas and prevents it from excreting enzymes necessary for digestion. Other problems associated with CF include diarrhea, malnutrition, and poor growth. The majority of affected men experience fertility issues, as the vas deferens, the tubes that carry sperm, are absent.

O High Impact

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

O Treatment Benefits

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

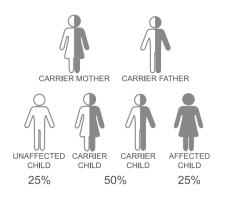
Clinical Information

Physical Impairment
Cognitive Impairment

Shortened Lifespan

Effective Treatment

Inheritance: Autosomal Recessive



Prognosis

Prognosis is fair. With current treatments, 80% of affected patients live to adulthood, and the overall median survival is 36.5 years. The median survival of males is longer than that of females.

Treatment

Treatment involves oral and inhaled antibiotics to prevent and control lung infections, inhaled bronchodilators to open the airway if breathing becomes compromised, mucolytic agents to break apart mucus, and chest physiotherapy to shake apart mucus buildup in the lungs. Pancreatic enzyme supplements may be required to help individuals with pancreatic insufficiency properly digest their food. A high-fat, high-calorie diet is recommended for individuals with cystic fibrosis to help maintain weight. Lung transplant is often an option for individuals with severe lung disease. Assisted reproductive technology allows most affected men to father children.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
African American	69.99%	1/62	1/207
Ashkenazi Jewish	96.81%	1/23	1/721
Asian	65.42%	1/94	1/272
European	94.96%	1/25	1/496
Hispanic American	77.32%	1/48	1/212
Native American	84.34%	1/53	1/338

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/cystic-fibrosis

Cystinuria: Type I (SLC3A1)

Cystinuria is characterized by the buildup of cystine in the kidneys and bladder. If it is caused by mutations in the SLC3A1 gene, it is referred to as type I cystinuria. Kidneys are responsible for filtering blood to create urine. Cystine is normally absorbed back into the bloodstream in this process. Individuals with Cystinuria cannot properly reabsorb cystine; the amino acid accumulates in their urine causing crystals to form and become stones. Stones can be lodged in the kidneys, ureter, and bladder. The stones formed as a result of Cystinuria are most common in young adults under age 40. As with non-Cystinuria-related urinary tract stones, blood in the urine and flank pain are seen in affected individuals.

O High Impact

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

O Treatment Benefits

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

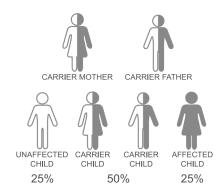
Clinical Information

Physical Impairment
Cognitive Impairment

Shortened Lifespan

Effective Treatment

Inheritance: Autosomal Recessive



To learn more, visit recombine.com/diseases/cystinuria-type-i

Prognosis

The prognosis is generally favorable. While Cystinuria is a chronic, lifelong condition, the condition rarely results in kidney failure and the number of kidney stones can be managed with diet changes and medication. Individuals with Cystinuria may also experience periods of pain due to kidney stones that may require management with medication.

Treatment

Treatment for Cystinuria involves drinking plenty of fluids to produce large amounts of water. In some cases, fluids may need to be administered intravenously. A diet low in salt may also reduce the amount cystine release and stone formation. Smaller stones will pass in the urine while larger stones may need to be surgically removed. There are also medication that can be used to prevent cystine from becoming stones.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
European	46.67%	1/42	1/79
Swedish	55.88%	1/159	1/360

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



Methods and Limitations

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

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

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

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

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



CarrierMap™

Diseases & Mutations Assayed

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

17-Alpha-Hydroxylase Deficiency (CYP17A1): Mutations (20): d^{*} Genotyping | c.157_159delTTC (p.53delF), c.316T>C (p.5106P), 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): o* 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): of Genotyping | c.293-13C>G

21-Hydroxylase-Deficient Nonclassical Congenital Adrenal Hyperplasia (CYP21A2): Mutations (1): 0^a Genotyping | c.1360C>T (p.P454S)

3-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD3B2): Mutations (6): o^a 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): d³ 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): d^{*} 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): o^{*} 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): of 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): d^{*} 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): of Genotyping | c.12973C>T (p.R4325X), c.7504C>T (p.R2502X), c.9742T>C (p.W3248R), c.8844delT (p.12949fs), c.5836T>C (p.W1946R), c.3161T>C (p.F1054S) Sequencing | NM_014363:2-10

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

Acrodermatitis Enteropathica (SLC39A4): Mutations (7): 3^o 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): 3^o 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): d^{*} 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): of 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): o^{*} 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): o^{*} Genotyping | c.2426T>C (p.I.809P), c.2248C>T (p.R750W), c.1830+1G>C (p.V549_E610del) Sequencing | NM_000528:1-24 Alport Syndrome: COL4A3 Related (COL4A3): Mutations (3): o^{*} Genotyping | c.4420_4423delCTTTT, c.4441C>T (p.R1481X), c.4571C>G (p.S1524X) Sequencing | NM_000091:2-52

Alport Syndrome: COL4A4 Related (COL4A4): Mutations (4): o^{*} 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): d^{*} 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.R136L), 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): 0^a 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): d^a 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): d^{*} 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.111T), 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): d^{*} 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): of 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): o^a Genotyping | c.1012A>G (p.S338G), c.514C>T (p.Q172X) Sequencing | NM_001271685:1-8 Asparagine Synthetase Deficiency (ASNS): Mutations (1): o^a Genotyping | c.1084T>G (p.F362V) Sequencing | NM_001673:3-13

Aspartylglycosaminuria (AGA): Mutations (7): o* 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): d^{*} 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): d^{*} 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): d* 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.13553T), c.10174C>T (p.Q3392X), c.9530T>C (p.13177T), c.9053C>T (p.S3018F), c.8870T>C (p.12957T), c.8011C>T (p.R2671X), c.6992T>A (p.12331K), c.5221G>A (p.V1741M), c.4991C>T (p.S1664F), c.3761_3762delCCinsG (p.A1254fs), c.2414C>T (p.P805L), c.664A>G (p.1222V), 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.13468V), c.1529delG (p.G510fs), c.657C>T (p.G219G), c.5513A>G (p.Y1838C), c.10856delA (p.K3619fs), c.5381-9T>G (IVS38-9T>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): 0^a 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): 0^a 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): o* Genotyping | c.388C>T (p.P130S) Sequencing | NM_001099679:2

Bardet-Biedl Syndrome: BBS12 Related (BBS12): Mutations (5): d^{*} 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): 0^a 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): of 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): 0⁸ 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): d^a 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.93-1G>A, c.92+1G>A, c.92+5G>A, c.92+5G>L, 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.-137*c*>g, c.-138*c*>t, c.-151C>T, c.118C>T (p.Q40X), c.169G>C (p.G57R), c.295G>A (p.V9PM), c.415G>C (p.A139P), c.47G>A (p.W16X), c.48G>A (p.W16X), c.80I>g, c.27C, 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.134delC (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.W166fs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.27LG, 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): 0^a 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.1312T), 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): of 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.Y414lfs), c.1240_1251 delTATCTCCACGTC (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): of Genotyping |

c.2207_2212delATCTGAinsTAGATTC (p.Y736Lfs), c.2407insT, c.557_559delCAA (p.S186X), c.1284GSA (p.W428X), c.1701GSA (p.W567X), c.1933CST (p.Q645X), c.2528CST (p.T843I), c.2695CST (p.R899X), c.3107GST (p.C1036F), c.2923delC (p.Q975K), c.3558+1GST, c.3875-2ASG, c.2074+2TSA, c.2343_2344dupGA (p.781EfsX), c.318_319insT (p.1107fs), c.380delC (p.127ffs), c.3564delC (p.1188Dfs), c.4008delG (p.1336Rfs), c.947CSG (p.S316X), c.2193+1_2193+9del9, c.1642CST (p.Q548X), c.3143delA (p.1048NfsX), c.356_357delTA (p.C120Hfs), c.4076+1delG, c.3281CSA (p.S1094X) Sequencing | NM_000057:2-22

Canavan Disease (ASPA): Mutations (8): d^{*} 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): d^{*} 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): d^{*} 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_00098:1-5

Carnitine-Acylcarnitine Translocase Deficiency (SLC25A20): Mutations (7): o^{*} 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): of Genotyping | c.434T>A (p.L145X), c.408_409insT (p.136fsX) Sequencing | NM_016277:2-7

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

Cerebrotendinous Xanthomatosis (CYP27A1): Mutations (14): o^a 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): d^{*} 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): 0^{*} 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): O^{*} Genotyping | c.6058delC (p.P2020fs) Sequencing | NM_033305:1-72

Chronic Granulomatous Disease: CYBA Related (CYBA): Mutations (12): of 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): d^{*} 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): d^{*} 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): d^{*} 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): d^a 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): d^{*} 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): d^{*} 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.12820T), 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): d⁷ 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): d^a 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): 5^a Genotyping | c.884G>A (p.R295H) Sequencing | NM_002435:1-8

Congenital Disorder of Glycosylation: Type 1C: ALG6 Related (ALG6): Mutations (4): of 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): of 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): of 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_208deITG (p.E70Afs), c.1550G>A (p.G517E), c.717+4A>T, c.429-1G>C, c.1660deIC (p.R554fs), c.2046+3A>C, c.2084C>T (p.P695L) Sequencing | NM_002529:2-17

Congenital Lipoid Adrenal Hyperplasia (STAR): Mutations (12): 5^a 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

CarrierMap™

(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): d^a 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): of Genotyping | c.601C>T (p.R201X), c.539G>C (p.G180A), c.548_551delTCCT (p.F183fs), c.1263_1264insC (p.5422fs), c.101-1G>T, c.331+1G>T Sequencing | NM_173660:3-7

Congenital Myasthenic Syndrome: RAPSN Related (RAPSN): Mutations (11): o^{*} 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): d^a 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): of 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): of 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): of 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): d^a Genotyping | c.1029delC, c.1153_1154insAT, c.1477delCA, c.1519_1521 delATC (p.507dell), 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.1911 delG,

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.1105fs), 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.Q990X), c.1526delG (p.G509fs), c.2908+1085_3367+260del7201, c.11C>A (p.S4X), c.3878_3881delTATT (p.V1293fs), c.3700A>G (p.11234V), 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): d^{*} 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.167_P73del) Sequencing | NM_001031681:1,3-13 Cystinuria: Non-Type I (SLC7A9): Mutations (15): d^{*} 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.144T), 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): d^{*} 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): d^{*} 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): d^{*} Genotyping | c.215A>G (p.N72S), c.1144G>A (p.E382K) Sequencing | NM_000352:1-39

Du Pan Syndrome (GDF5): Mutations (4): d^a 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): d^a 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): of Genotyping | c.2470_2471 insG, 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): d^{*} 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): of 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): of 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.M11), c.488G>A (p.R163Q) Sequencing | NM_014297:1-7

Familial Chloride Diarrhea (SLC26A3): Mutations (6): of Genotyping | c.344delT (p.11151), 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): d^{*} 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): d^{*} Genotyping | c.3989-9G>A, c.4159_4161delTTC (p.1387delF), c.4258C>T (p.R1420C), c.4477C>T (p.R1493W), c.2147G>T (p.G716V), c.4055G>C (p.R1352P), c.560T>A (p.V187D), c.4516G>A (p.E1506K), c.2506C>T (p.Q836X), c.579+2T>A, c.1333-1013A>G (IVS8-1013A>G) Sequencing | NM_000352:1-39

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

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

Fanconi Anemia: Type A (FANCA): Mutations (10): of 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): 0^a 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): d^{*} 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): o^a Genotyping | c.2392C>T (p.R798X) Sequencing | NM_032043:2-20

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

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

CarrierMap™

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

GRACILE Syndrome (BCS1L): Mutations (12): d^{*} 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): of 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): 0^a 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): d^{*} 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.1305V), c.683_694delATCTCTGGGAGTinsCTC (p.N228_5232del5insTP), c.246A>G (p.182M), c.1161+6555_*9573del31670bp Sequencing | NM_000153:2-17

Glutaric Acidemia: Type I (GCDH): Mutations (8): 0^a 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): d^{*} Genotyping | c.797C>T (p.T266M), c.470T>G (p.V157G), c.346G>A (p.G116R), c.809_811 delTAG (p.V270_A271 delinsA), c.963+1 delG Sequencing | NM_000126:1-12

Glutaric Acidemia: Type IIC (ETFDH): Mutations (8): o^{*} 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): of 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): 0^a 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): 0⁸ 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): 0^a 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): of 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 (14): Ø 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) Sequencing | NM_000642:2-34

Glycogen Storage Disease: Type IV (GBE1): Mutations (3): 0^a 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): of 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): d^a 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): d^{*} 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): of 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): of Genotyping | c.959G>T (p.G320V) Sequencing | NM_213653:2-4

Hemochromatosis: Type 3: TFR2 Related (TFR2): Mutations (4): of 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): 0^a 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): 3^a Genotyping | c.79G>A (p.E27K) Sequencing | NM_000518:1-3

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

Hereditary Fructose Intolerance (ALDOB): Mutations (10): of 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): o^{*} Genotyping | c.3416delT (p.L1139fs) Sequencing | NM_014844:2-20

Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3): Mutations (1): 3^e Genotyping | c.1981C>T (p.R661X) Sequencing | NM_000227:1-38

Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3): Mutations (6): σ³ 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): σ^a Genotyping | c.283C>T (p.R95X) Sequencing | NM_005562:1-23

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

Hermansky-Pudlak Syndrome: Type 3 (HPS3): Mutations (4): d^{*} 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): 0^a 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): σ^3 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): d^a Genotyping | c.919G>A (p.G307S), c.833T>C (p.1278T), 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): 0^a 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): d^{*} 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): 0^a 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): 0^a Genotyping | c.218G>T (p.R73L), c.218G>A (p.R73H) Sequencing | NM_001173991:1-5

Lamellar Ichthyosis: Type 1 (TGM1): Mutations (1): 0^a Genotyping | c.877-2A>G (IVS5-2A>G) Sequencing | NM_000359:2-15

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

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

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

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

Leber Congenital Amaurosis: RDH12 Related (RDH12): Mutations (6): o^{*} 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): 0^a Genotyping | c.1061C>T (p.A354V) Sequencing | NM_133259:1-38

Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related (EIF2B5): Mutations (9): d^{*} 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): o^{*} 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.152T), c.537-3C>A Sequencing | NM_000233:1-11

Limb-Girdle Muscular Dystrophy: Type 2A (CAPN3): Mutations (6): 3^o 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): d³ 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): 3^o 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): o" Genotyping | c.229C>T (p.R77C) Sequencing | NM_000023:1-9

Limb-Girdle Muscular Dystrophy: Type 2E (SGCB): Mutations (6): of 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): of 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.12761) Sequencing | NM_001039885:1-4

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

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

Lysinuric Protein Intolerance (SLC7A7): Mutations (4): of 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): d^{*} 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): 0^a 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): d^{*} 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): d^a 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): o⁷ 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): d^{*} 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): d^a Genotyping | c.985A>G (p.K329E), c.362C>T (p.T1211), c.583G>A (p.G195R), c.799G>A (p.G267R), c.199T>C (p.Y67H), c.262C>T (p.L88F), c.616C>T (p.R206C), c.617G>A (p.C206H) Sequencing | NM_001127328:1-12

Megalencephalic Leukoencephalopathy (MLC1): Mutations (6): o⁷ Genotyping | c.176G>A (p.G59E), c.278C>T (p.S93L), c.135_136insC (p.C46fsX), c.908_918delTGCTGCTGCTGGTGGCA (p.V303GfsX96), c.880C>T (p.P294S), c.178-10T>A Sequencing | NM_139202:2-12

Metachromatic Leukodystrophy (ARSA): Mutations (18): of Genotyping | c.1210+1G>A, c.465+1G>A (IVS2+1G>A), c.862A>C (p.7288P), c.1136C>T (p.P379L), c.1283C>T (p.P428L), c.827C>T (p.T276M), c.542T>G (p.1181S), c.1232C>T (p.T4111), 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): of 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): σ^{*} 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): of 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): σ^a 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): 3^a Genotyping | c.344G>A (p.C115Y) Sequencing | NM_004553:1-4

Mitochondrial DNA Depletion Syndrome: MNGIE Type (TYMP): Mutations (6): of 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): o⁷ 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): o⁷ 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_7777insT (p.G259fs), c.1175C>T (p.A392V) Sequencing | NM_000183:2-16

Morquio Syndrome: Type A (GALNS): Mutations (6): d^{*} 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.1113F), c.178G>A (p.D60N) Sequencing | NM_000512:2-14

Morquio Syndrome: Type B (GLB1): Mutations (8): o^{*} 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): 0^a 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): d[®] 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): d^a 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): of Genotyping | c.463T>C (p.S155P) Sequencing | NM_182760:1-9

Muscle-Eye-Brain Disease (POMGNT1): Mutations (3): d^{*} 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): d^a Genotyping | c.149G>A (p.R50Q) Sequencing | NM_002437:2-8

Nemaline Myopathy: NEB Related (NEB): Mutations (2): 3^a 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): 0^a Genotyping | c.121_122delCT (p.L41Dfs), c.1481delC, c.3325C>T (p.R1109X), c.3478C>T (p.R1160X), c.2335-1G>A

CarrierMap™

Sequencing | NM_004646:1-29

Nephrotic Syndrome: Type 2 (NPHS2): Mutations (27): of 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.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): d^{*} 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): d^{*} 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): d^a Genotyping | c.881C>A (p.T294K), c.754+2T>A Sequencing | NM_152778:2-13

Neuronal Ceroid-Lipofuscinosis: PPT1 Related (PPT1): Mutations (8): o^{*} 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): d^{*} 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): 0^a 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): d^{*} Genotyping | c.1828_1830delCGC (p.610delR), c.880C>A (p.Q294K), c.1280A>G (p.H427R) Sequencing | NM_000543:1-6

Niemann-Pick Disease: Type C1 (NPC1): Mutations (14): of 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.11061T), 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): d^{*} Genotyping | c.58G>T (p.E20X), c.436C>T (p.Q146X), c.358C>T (p.P120S), c.352G>T (p.E118X), c.332delA (p.N1111fs), 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): d^a Genotyping | c.657_661delACAAA (p.K219fs) Sequencing | NM_002485:1-16

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutations (29): o^{*} Genotyping | c.167delT, c.235delC, c.313_326delAAGTTCATCAAGGG, c.358delGAG (p.120delE), c.35delG, c.370C>T (p.Q124X), c.427C>T (p.R143W), c.109G>A (p.V37I), c.231G>A (p.W77X), c.551G>C (p.R184P), c.71G>A (p.W24X), c.229T>C (p.W77R), c.269T>C (p.190P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T, c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147X), 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): of 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): d^a 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): d⁷ 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.0326fs), c.1138_1158delTCTGCCAACGATCCTATCTTC (p.S380_F386del) Sequencing | NM_000372:1-5 Oculocutaneous Albinism: Type 3 (TYRP1): Mutations (6): o^a 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): d^{*} Genotyping | c.469G>A (p.D157N), c.563G>T (p.G188V) Sequencing | NM_016180:1-7

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

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

Ornithine Translocase Deficiency (SLC25A15): Mutations (3): d^{*} 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): d^{*} 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): o^{*} 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): d^{*} 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): 0^a 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): of 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): of 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 (62): Or Genotyping | c.1066-11 G>A (IVS10-11 G>A), c. 1315+1 G>A (IVS12+1 G>A), c. 1241 A>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), c.456_706+138del11653 Sequencing | NM_000277:1-13

Polyglandular Autoimmune Syndrome: Type I (AIRE): Mutations (5): d^a Genotyping | c.769C>T (p.R257X), c.254A>G (p.Y85C), c.1163_1164insA (p.M388lfsX36), c.967_979delCTGTCCCCTCCGC (p.L323SfsX51), c.415C>T (p.R139X) Sequencing | NM_000383:1-14

Pontocerebellar Hypoplasia: EXOSC3 Related (EXOSC3): Mutations (4): d^{*} 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): σ^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): σ^3 Genotyping | c.1001A>G (p.Y334C) Sequencing | NM_016955:1-11

Pontocerebellar Hypoplasia: TSEN54 Related (TSEN54): Mutations (3): o^{*} 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): 0^a Genotyping | c.2084A>G (p.Q695R), c.1556+5G>A Sequencing | NM_001128159:1-22

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

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

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

Primary Ciliary Dyskinesia: DNA11 Related (DNA11): Mutations (5): d³ 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): d^a 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): d^a 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): d^{*} Genotyping | c.508G>A (p.G170R), c.454T>A (p.F152I), c.731T>C (p.1244T), c.121G>A (p.G41R), c.198C>G (p.Y66X), c.245G>A (p.G82E), c.466G>A (p.G156R), c.613T>C (p.5205P), 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): of Genotyping | c.103delG, c.404+3delAAGT, c.295C>T (p.R99X) Sequencing | NM_012203:1-9

Primary Hyperoxaluria: Type 3 (HOGA1): Mutations (2): 0^a 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): o^{*} 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): d^a 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): d[®] Genotyping | c.280G>T (p.G94X), c.335G>A (p.G112D), c.457G>C (p.A153P), c.502G>A (p.E168K), c.1218_1231 delGGGCATCATCCGGCinsTAGAGCACAGGA (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): o^{*} Genotyping | c.293A>G (p.D98G) Sequencing | NM_000055:2-4

Pycnodysostosis (CTSK): Mutations (2): d^a Genotyping | c.990A>G (p.X330W), c.926T>C (p.I309P) Sequencing | NM_000396:2-8

Pyruvate Carboxylase Deficiency (PC): Mutations (15): of 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): d^{*} Genotyping | c.395A>G (p.Y132C), c.1030C>T (p.P344S) Sequencing | NM_000925:1-10

Renal Tubular Acidosis and Deafness (ATP6V1B1): Mutations (7): of 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): of 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): 0^a 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): d^a Genotyping | c.420delT (p.1141Lfs), 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): 0^a Genotyping | c.124A>G (p.K42E) Sequencing | NM_024887:2-9

Retinitis Pigmentosa: FAM161A Related (FAM161A): Mutations (5): of 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): d^{*} 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): o⁷ Genotyping | c.802_816delTCATCATCAGAAAT (p.I.336fsX13), 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

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Sandhoff Disease (HEXB): Mutations (14): of 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): o^{*} 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): d^{*} 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.R51L), 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): 0^a 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): d^{*} 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): d^{*} 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): 0^a Genotyping | c.20A>T (p.E7V) Sequencing | NM_000518:1-3

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

Sly Syndrome (GUSB): Mutations (5): 0^a 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): σ⁸ 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.1178F), 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_855deITTC (p.285deIF), c.1327C>T (p.R443C), c.513C>T (p.P51S), c.296T>C (p.L9PP), 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.W37X) Sequencing | NM_001360:3-9

Spinal Muscular Atrophy: SMN1 Linked (SMN1): Mutations (19): d³ 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): 0^a 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): of 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): d^a 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.1335F), c.910_912deITTC (p.305deIF), c.749G>A (p.G250D), c.632T>C (p.F211S), c.629C>T (p.S210F), c.613deIC, 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.776T>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.108G>T (p.G336H), 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.D208fs), c.1211_1212delTG (p.L404fs), c.621T>G (p.D207E), c.151G>A (p.R504H), c.1177C>T (p.R393X), c.21>C (p.M1T), c.1292G>A (p.W31X), c.947_948insA (p.Y316fs), c.607T>G (p.W203G), c.1061_1063delTCT (p.F354_Y355delinsX), c.1043_1046delTCAA (p.F348fs), c.1513C4G (p.E375fs), c.1121A>G (p.Q374R), c.1043_1046delTCAA (p.F348fs), c.1510elC (p.F375s), c.1511C> (p.L484F), c.964G>T (p.D322Y), c.351C>G (p.L451V), c.571-2A>G (IVS5-2A>G) Sequencing | NM_000520:1-14

Trichohepatoenteric Syndrome: Type 1 (TTC37): Mutations (9): 0^a 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.2578-7delTTTT Sequencing | NM_014639:4-43

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

Tyrosinemia: Type I (FAH): Mutations (10): d^{*} 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): of 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): 0^a Genotyping | c.93C>A (p.C31X), c.448C>T (p.R150X), c.634C>T (p.R212C), c.635G>A (p.R212H), c.700C>T (p.Q234X), c.1797G>A (p.M599I), c.1996C>T (p.R666X), c.2476G>A (p.A826T), c.3719G>A (p.R1240Q), c.5581C>T (p.R1861X), c.6025delG (p.A2009fs), c.640G>A (p.G214R), c.1190C>A (p.A397D) Sequencing | NM_000260:2-49

Usher Syndrome: Type 1C (USH1C): Mutations (5): 0^{*} Genotyping | c.496+1G>A, c.238_239insC, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.36+1G>T Sequencing | NM_153676:1-27

Usher Syndrome: Type 1D (CDH23): Mutations (14): d³ 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): d^a 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): d° Genotyping | c.14020A>G (p.R4674G), c.12067-2A>G, c.4338_4339delCT (p.C1447fs), c.2299delG (p.E7675fsX21), c.2209C>T (p.R737X), c.1256G>T (p.C149F), c.1000C>T (p.R34W), 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.R777W), c.9165_9168delCTAT (p.13055MfsX2), 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): d^{*} 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): of 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.G411), 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.383_931delGAGA (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.R229W), 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): d^{*} 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): d^{*} 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): of 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): d^o 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): d* 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): d^{*} 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): d^{*} 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): d^a 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): O^a Genotyping | c.764_765insA, c.874_875delCT Sequencing | NM_153818:2-6

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

Zellweger Spectrum Disorders: PEX6 Related (PEX6): Mutations (8): 0^a 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.T572l) Sequencing | NM_000287:1-17

Residual Risk Information

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

Disease	Carrier Rate	Detection	Residual
136036		Rate	Risk
11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	♂ Moroccan Jewish: 1/39	91.67%	1/468
17-Alpha-Hydroxylase Deficiency	o [®] Brazilian: Unknown	54.55%	Unknown
	o ⁷ Japanese: Unknown	45.45%	Unknown
17-Beta-Hydroxysteroid Dehydrogenase Deficiency	o" Arab: 1/8	>99%	<1/800
	o [*] Dutch: 1/192	13.89%	1/223
21 -Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia	♂ European: 1/62	27.65%	1/86
	o [*] General: 1/62	29.34%	1/88
21 -Hydroxylase-Deficient Nonclassical Congenital Adrenal Hyperplasia	o" Argentinian: 1/4	<10%	1/4
	o" European: 1/16	<10%	1/16
3-Beta-Hydroxysteroid Dehydrogenase Deficiency	o' General: Unknown	16.13%	Unknown
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related	o ^a European: 1/146	26.32%	1/198
	o ^a General: 1/112	37.50%	1/179
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related	ð General: 1/112	35.29%	1/173
	o" Japanese: 1/112	33.33%	1/168
	o" Korean: 1/141	66.67%	1/423
	o [*] Turkish: 1/112	24.07%	1/148
3-Methylglutaconic Aciduria: Type 3	0" Iraqi Jewish: 1/10	>99%	<1/1,000
3-Phosphoglycerate Dehydrogenase Deficiency	o" Ashkenazi Jewish: 1/400	>99%	<1/40,00 0
5-Alpha Reductase Deficiency	o [®] Dominican: Unknown	>99%	Unknown
	o [®] Mexican: Unknown	68.75%	Unknown
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	o ^a Chinese: 1/183	78.95%	1/869
	o" East Asian: 1/180	64.20%	1/503
ARSACS	o" French Canadian: 1/22	95.45%	1/484
Abetalipoproteinemia	o ^a Ashkenazi Jewish: 1/131	>99%	<1/13,10 0
Acrodermatitis Enteropathica	o [*] Arab: Unknown	40.00%	Unknown
	o [*] Egyptian: Unknown	33.33%	Unknown
	o ^a French: Unknown	27.78%	Unknown
	o [®] Tunisian: Unknown	77.78%	Unknown
Acute Infantile Liver Failure: TRMU Related	o ^a Yemenite Jewish: 1/40	71.43%	1/140
Acyl-CoA Oxidase I Deficiency	o ^r General: Unknown	35.00%	Unknown
	o ⁷ Japanese: Unknown	42.86%	Unknown
Adenosine Deaminase Deficiency	o [∎] General: 1/388	36.96%	1/615

CarrierMap[™]

Disease	Carrier Rate	Detection Rate	Residual Risk
Alkaptonuria	o" Dominican: Unknown	>99%	Unknown
	o " Finnish: 1/251	60.00%	1/628
	ð" Slovak: 1/69	59.38%	1/170
Alpha Thalassemia	o' General: 1/48	50.67%	1/97
Alpha-1-Antitrypsin Deficiency	o" European: 1/35	95.00%	1/700
	o' General: Unknown	95.00%	Unknown
Alpha-Mannosidosis	o" European: 1/354	30.23%	1/507
	o' General: 1/354	35.19%	1/546
Alport Syndrome: COL4A3 Related	o" Dutch: 1/409	22.73%	1/529
Alport Syndrome: COL4A4 Related	o' General: 1/409	23.33%	1/533
Amegakaryocytic Thrombocytopenia	o [*] Ashkenazi Jewish: 1/76	>99%	<1/7,600
	o' General: Unknown	64.81%	Unknown
Andermann Syndrome	o [*] French Canadian: 1/24	99.38%	1/3,888
Antley-Bixler Syndrome	o" General: Unknown	45.65%	Unknown
	o ^r Japanese: Unknown	60.47%	Unknown
Argininemia	o ^r Chinese: Unknown	40.00%	Unknown
	o ^a French Canadian: Unknown	75.00%	Unknown
	o ^r Japanese: Unknown	>99%	Unknown
Argininosuccinate Lyase Deficiency	o" European: 1/133	57.41%	1/312
	o" Saudi Arabian: 1/80	51.72%	1/166
Aromatase Deficiency	o' General: Unknown	25.00%	Unknown
Arthrogryposis, Mental Retardation, & Seizures	♂ Ashkenazi Jewish: 1/205	>99%	<1/20,50 0
Asparagine Synthetase Deficiency	o" Iranian Jewish: 1/80	>99%	<1/8,000
Aspartylglycosaminuria	ơ" Finnish: 1/69	96.12%	1/1,780
Ataxia with Vitamin E Deficiency	o" European: 1/274	80.00%	1/1,370
	o" Italian: 1/224	97.73%	1/9,856
	♂ North African: 1/159	>99%	<1/15,90 0
Ataxia-Telangiectasia	o" Costa Rican: 1/100	68.52%	1/318
	ð" North African Jewish: 1/81	96.97%	1/2,673
	o" Norwegian: 1/197	50.00%	1/394
	o ^a Sardinians: Unknown	85.71%	Unknown
	o ^r US Amish: Unknown	>99%	Unknown
Autosomal Recessive Polycystic Kidney Disease	o" Finnish: 1/45	84.21%	1/285
	o' French: 1/71	62.50%	1/189
	o" General: 1/71	37.11%	1/113
Bardet-Biedl Syndrome: BBS1 Related	o' General: 1/376	70.27%	1/1,265
	♂ Northern European: 1/376	85.90%	1/2,666
	o" Puerto Rican: Unknown	90.00%	Unknown
Bardet-Biedl Syndrome: BBS10 Related	o ^r General: 1/404	47.79%	1/774
Bardet-Biedl Syndrome: BBS11 Related	o" Bedouin: 1/59	>99%	<1/5,900
Bardet-Biedl Syndrome: BBS12 Related	o'' General: Unknown	50.00%	Unknown

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Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Bardet-Biedl Syndrome: BBS2 Related	o" Ashkenazi Jewish: Unknown	>99%	Unknown		o [*] Moroccan Jewish: 1/234	>99%	<1/23,40 0
	o" General: 1/638	38.46%	1/1,037	Citrin Deficiency	o" Japanese: 1/70	>99%	<1/7,000
	o" Middle Eastern: Unknown	>99%	Unknown	Citrullinemia: Type I	o" European: 1/120	18.18%	1/147
Bare Lymphocyte Syndrome: Type II	o" General: Unknown	66.67%	Unknown		o" General: 1/120	52.27%	1/251
Bartter Syndrome: Type 4A	o" General: 1/457	81.82%	1/2,514		o ^r Japanese: Unknown	64.71%	Unknown
Beta Thalassemia	o" African American: 1/75	84.21%	1/475		o [*] Mediterranean: 1/120	50.00%	1/240
	0" Indian: 1/24	74.12%	1/93	Classical Galactosemia	o [*] African American: 1/78	73.13%	1/290
	o" Sardinians: 1/23	97.14%	1/804		o [*] Ashkenazi Jewish: 1/127	>99%	<1/12,70
	o" Spaniard: 1/51	93.10%	1/739		2 0 1 1 (01	75 479/	0
Beta-Hexosaminidase Pseudodeficiency	♂" Ashkenazi Jewish: Unknown	>99%	Unknown		o" Dutch: 1/91 o" European: 1/112	75.47% 88.33%	1/371 1/960
	o" General: Unknown	>99%	Unknown		o' General: 1/125	80.00%	1/625
Beta-Ketothiolase Deficiency	o ^r Japanese: Unknown	58.33%	Unknown		o " Irish: 1/76	91.30%	1/874
	o" Spaniard: Unknown	90.00%	Unknown		o" Irish Travellers: 1/14	>99%	<1/1,400
Biotinidase Deficiency	o" General: 1/123	78.32%	1/567	Cockayne Syndrome: Type A	o ^r Christian Arab: Unknown	50.00%	Unknown
Bloom Syndrome	o" Ashkenazi Jewish: 1/134	96.67%	1/4,020	Cockayne Syndrome: Type B	o' General: 1/378	19.30%	1/468
	o ^r European: Unknown	66.22%	Unknown	Cohen Syndrome	o' European: Unknown	19.05%	Unknown
	o ^r Japanese: Unknown	50.00%	Unknown		o " Finnish: 1/140	67.24%	1/427
Canavan Disease	o ^a Ashkenazi Jewish: 1/55	98.86%	1/4,840		o" US Amish: 1/12	>99%	<1/1,200
	o ^a European: Unknown	53.23%	Unknown	Combined Pituitary Hormone	o" European: 1/45	93.29%	1/671
Carnitine Palmitoyltransferase IA Deficiency	o' General: Unknown	38.89%	Unknown	Deficiency: PROP1 Related	o' General: 1/45	82.35%	1/255
	o ^a Hutterite: 1/16	>99%	<1/1,600	Congenital Disorder of Glycosylation:	o" Danish: 1/71	90.00%	1/710
	o ^a Japanese: 1/101	66.67%	1/303	Type 1A: PMM2 Related			
Carnitine Palmitoyltransferase II	o" Ashkenazi Jewish:	>99%	Unknown		o [*] Dutch: 1/68	39.29%	1/112
Deficiency	Unknown				o" European: 1/71	55.33%	1/159
	o'' General: Unknown	71.43%	Unknown	Congenital Disorder of Glycosylation: Type 1B: MPI Related	o' French: Unknown	54.17%	Unknown
Carnitine-Acylcarnitine Translocase Deficiency	o ^r Asian: Unknown	95.45%	Unknown	Congenital Disorder of Glycosylation: Type 1C: ALG6 Related	o" French: Unknown	59.09%	Unknown
	o ^a General: Unknown	18.75%	Unknown		o ^r General: Unknown	86.21%	Unknown
Carpenter Syndrome	o ^a Brazilian: Unknown	40.00%	Unknown	Congenital Ichthyosis: ABCA12 Related		>99%	Unknown
	o ^a Northern European: Unknown	85.00%	Unknown		o [*] South Asian: Unknown	66.67%	Unknown
Cartilage-Hair Hypoplasia	o ^a Finnish: 1/76	93.33%	1/1,140	Congenital Insensitivity to Pain with	o ^r Japanese: Unknown	56.52%	Unknown
	o" US Amish: 1/19	>99%	<1/1,900	Anhidrosis		00.0270	
Cerebrotendinous Xanthomatosis	o ^r Dutch: Unknown	78.57%	Unknown		o ^a Moroccan Jewish: Unknown	>99%	Unknown
	o ^a Italian: Unknown	45.95%	Unknown	Congenital Lipoid Adrenal Hyperplasia	o " Japanese: 1/201	51.11%	1/411
	o ^a Japanese: Unknown	92.86%	Unknown		o [*] Korean: 1/251	63.64%	1/690
	o ^a Moroccan Jewish: 1/6	87.50%	1/48	Congenital Myasthenic Syndrome:	o [*] European Gypsy: 1/26	>99%	<1/2,600
Chediak-Higashi Syndrome	o ^a General: Unknown	19.64%	Unknown	CHRNE Related			
Cholesteryl Ester Storage Disease	o' General: 1/101	68.97%	1/325		o" North African: Unknown	60.87%	Unknown
Choreoacanthocytosis	o" Ashkenazi Jewish: Unknown	66.67%	Unknown	Congenital Myasthenic Syndrome: DOK7 Related	o" European: 1/472	19.05%	1/583
Chronic Granulomatous Disease:	o" Iranian: Unknown	71.43%	Unknown		o" General: 1/472	18.75%	1/581
CYBA Related	0 ⁷ Japanese: 1/274	>99%	<1/27,40 0	Congenital Myasthenic Syndrome: RAPSN Related	o' General: 1/437	88.57%	1/3,824
	o" Korean: 1/105	>99%	0 <1/10,50 0		ơ" Non-Ashkenazi Jewish: Unknown	>99%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Congenital Neutropenia: Recessive	o" English: Unknown	11.76%	Unknown		o" Saudi Arabian: 1/38	>99%	<1/3,800
	o ^r Japanese: Unknown	22.22%	Unknown	Familial Dysautonomia	o" Ashkenazi Jewish: 1/31	>99%	<1/3,100
	o ^r Turkish: Unknown	89.47%	Unknown	Familial Hyperinsulinism: Type 1:	o" Ashkenazi Jewish: 1/52	98.75%	1/4,160
Corneal Dystrophy and Perceptive Deafness	o' General: Unknown	71.43%	Unknown	ABCC8 Related	ð Finnish: 1/101	45.16%	1/184
Corticosterone Methyloxidase Deficiency	♂ ^a Iranian Jewish: 1/32	>99%	<1/3,200	Familial Hyperinsulinism: Type 2: KCNJ11 Related	o" Arab: Unknown	40.00%	Unknown
Crigler-Najjar Syndrome	o ^a Sardinians: Unknown	80.00%	Unknown	Familial Mediterranean Fever	0" Arab: 1/4	51.27%	1/8
	o" Tunisian: Unknown	>99%	Unknown		o" Armenian: 1/5	94.51%	1/91
Cystic Fibrosis	o" African American: 1/62	69.99%	1/207		o" Ashkenazi Jewish: 1/81	40.95%	1/137
	o" Ashkenazi Jewish: 1/23	96.81%	1/721		0" Iraqi Jewish: 1/4	76.92%	1/17
	0" Asian: 1/94	65.42%	1/272		o" Israeli Jewish: 1/5	62.67%	1/13
	o" European: 1/25	94.96%	1/496		o" Lebanese: 1/6	91.67%	1/72
	o" Hispanic American: 1/48	77.32%	1/212		o" North African Jewish: 1/5	95.69%	1/116
	o" Native American: 1/53	84.34%	1/338		o" Syrian: 1/6	85.14%	1/40
Cystinosis	o" Dutch: 1/194	73.08%	1/721		o" Turkish: 1/5	74.43%	1/20
	o' French Canadian: 1/40	75.00%	1/160	Fanconi Anemia: Type A	o [®] Moroccan Jewish: 1/100	>99%	<1/10,00
	o'' General: 1/194	54.51%	1/426				0
Cystinuria: Non-Type I	o" European: 1/42	61.11%	1/108		o" Spanish Gypsy: 1/67	>99%	<1/6,700
	o' General: 1/42	37.50%	1/67	Fanconi Anemia: Type C	♂ Ashkenazi Jewish: 1/101	>99%	<1/10,10 0
	ơ" Libyan Jewish: 1/26	93.48%	1/399		o'' General: Unknown	30.00%	Unknown
	o [*] United States: 1/42	56.25%	1/96	Fanconi Anemia: Type G	o [®] Black South African:	81.82%	1/556
Cystinuria: Type I	o" European: 1/42	46.67%	1/79		1/101		
	o" Swedish: 1/159	55.88%	1/360		♂ ¹ French Canadian: Unknown	87.50%	Unknown
D-Bifunctional Protein Deficiency	o'' General: 1/159	38.64%	1/259		o" Japanese: Unknown	75.00%	Unknown
Diabetes: Recessive Permanent	o'' General: Unknown	25.00%	Unknown		o [®] Korean: Unknown	66.67%	Unknown
Neonatal	م الم	> 00%		Fanconi Anemia: Type J	o [®] General: Unknown	86.36%	Unknown
Du Pan Syndrome	o ^a Pakistani: Unknown	>99%	Unknown <1/20,30	Fumarase Deficiency	o [®] General: Unknown	30.00%	Unknown
Dyskeratosis Congenita: RTEL1 Related	o" Ashkenazi Jewish: 1/203 o" General: 1/501	>99% 50.00%	0	GM1-Gangliosidoses	o" Eurodescent Brazilian: 1/66	62.15%	1/174
Dystrophic Epidermolysis Bullosa:	o" Italian: Unknown	45.00%	1/1,002 Unknown		o" European: 1/194	50.00%	1/388
Recessive	O halian. Onknown	43.00%	Unknown		0 [°] General: 1/194	20.00%	1/243
	o" Mexican American: 1/345	56.25%	1/789		♂" Hispanic American: 1/194	58.33%	1/466
Ehlers-Danlos Syndrome: Type VIIC	ơ" Ashkenazi Jewish: Unknown	>99%	Unknown		o" Japanese: Unknown	62.82%	Unknown
Ellis-van Creveld Syndrome: EVC	o" General: 1/123	32.14%	1/181	GRACILE Syndrome	ơ" Finnish: 1/109	97.22%	1/3,924
Related		52.1470	17 101	Galactokinase Deficiency	o" Japanese: 1/501	50.00%	1/1,002
Ellis-van Creveld Syndrome: EVC2 Related	o'' General: Unknown	<10%	Unknown		0 ⁷ Roma: 1/51	>99%	<1/5,100
Enhanced S-Cone	o" Ashkenazi Jewish:	90.48%	Unknown	Gaucher Disease	0° Ashkenazi Jewish: 1/15	87.16%	1/117
	Unknown				o" General: 1/112	31.60%	1/164
	o'' General: Unknown	52.50%	Unknown		o" Spaniard: Unknown	44.29%	Unknown
Ethylmalonic Aciduria	o ^r Arab/Mediterranean:	29.17%	Unknown		o [*] Turkish: 1/236	59.38%	1/581
	Unknown	20.040/	Link	Gitelman Syndrome	o" European: 1/100	35.00%	1/154
Familial Chlarida Diant	o ^a General: Unknown	38.24%	Unknown		o" European Gypsy: Unknown	>99%	Unknown
Familial Chloride Diarrhea	o" Finnish: 1/51	>99%	<1/5,100		o" General: 1/101	30.00%	1/144
	o" Kuwaiti: 1/38	90.00%	1/380		o" Taiwanese: Unknown	64.29%	, Unknown
	o" Polish: 1/224	45.24%	1/409				

CarrierMap™

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Globoid Cell Leukodystrophy	o [*] Dutch: 1/137	60.98%	1/351	Hemochromatosis: Type 2A: HFE2	o [*] European: Unknown	69.23%	Unknown
	o' European: 1/150	26.47%	1/204	Related	o [*] Mediterranean: Unknown	70 70%	Unknown
	o' Japanese: 1/150	36.00%	1/234			72.73%	
Glutaric Acidemia: Type I	o' European: 1/164	57.78%	1/388	Hemochromatosis: Type 3: TFR2 Related	o [*] Italian: Unknown	73.21%	Unknown
	o [*] General: 1/164	25.51%	1/220	Hemoglobinopathy: Hb C	o" African American: 1/51	>99%	<1/5,100
	o" US Amish: 1/12	>99%	<1/1,200	Hemoglobinopathy: Hb D	o" Canadian: 1/64	>99%	<1/6,400
Glutaric Acidemia: Type IIA	o'' General: Unknown	71.43%	Unknown		o " Indian: 1/16	>99%	<1/1,600
Glutaric Acidemia: Type IIB	o [*] General: Unknown	33.33%	Unknown		o" Iranian: 1/11	>99%	<1/1,100
Glutaric Acidemia: Type IIC	o" Taiwanese: Unknown	>99%	Unknown	Hemoglobinopathy: Hb E	o" Cambodia: 1/4	>99%	<1/400
	o" Turkish: Unknown	80.00%	Unknown		o" Chinese: 1/13	>99%	<1/1,300
Glycine Encephalopathy: AMT Related	o" General: Unknown	40.91%	Unknown		o " Indian: 1/10	>99%	<1/1,000
Glycine Encephalopathy: GLDC Related	o ^a Finnish: 1/118	78.00%	1/536		ơ" Thai: 1/9	>99%	<1/900
	o" General: 1/280	12.50%	1/320	Hemoglobinopathy: Hb O	o" African American: 1/87	>99%	<1/8,700
Glycogen Storage Disease: Type IA	♂ Ashkenazi Jewish: 1/71	>99%	<1/7,100		o" Middle Eastern: Unknown	>99%	Unknown
	o" Chinese: 1/159	80.00%	1/795	Hereditary Fructose Intolerance	o" European: 1/81	72.73%	1/297
	o" European: 1/177	76.88%	1/765		ơ" Italian: 1∕81	90.91%	1/891
	o" Hispanic American:	27.78%	1/245		o" Slavic: 1/81	>99%	<1/8,100
	1/177			Hereditary Spastic Paraplegia: TECPR2 Related	ð" Bukharan Jewish: 1/75	>99%	<1/7,500
	0 ⁴ Japanese: 1/177	89.22%	1/1,641	Herlitz Junctional Epidermolysis	o" Pakistani: Unknown	>99%	Unknown
Glycogen Storage Disease: Type IB	of Australian: 1/354	50.00%	1/708	Bullosa: LAMA3 Related			
	o ^a European: 1/354 o ^a Japanese: 1/354	45.74% 39.13%	1/652 1/582	Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	o ⁷ European: Unknown	70.00%	Unknown
Glycogen Storage Disease: Type II	o [®] African American: 1/60	45.83%	1/111		o ^r General: 1/781	52.27%	1/1,636
Ciycogen Slorage Disease. Type in	o [®] Chinese: 1/112	72.00%	1/400	Herlitz Junctional Epidermolysis	o [*] Italian: Unknown	28.57%	Unknown
	o [®] European: 1/97	51.76%	1/201	Bullosa: LAMC2 Related			
	o [*] North African: Unknown	60.00%	Unknown	Hermansky-Pudlak Syndrome: Type 1	o" Puerto Rican: 1/22	94.95%	1/436
Glycogen Storage Disease: Type III	o [®] Faroese: 1/30	>99%	<1/3,000	Hermansky-Pudlak Syndrome: Type 3	o" Ashkenazi Jewish: 1/235	>99%	<1/23,50 0
olycogen olorage biscuse. type m	o [*] General: 1/159	39.81%	1/264		o ^a European: 1/434	12.50%	1/496
	o [*] North African Jewish:	>99%	<1/3,500	Hermansky-Pudlak Syndrome: Type 4	o ^r European: Unknown	54.17%	Unknown
	1/35		.,	Holocarboxylase Synthetase	o [*] European: 1/148	83.33%	1/888
Glycogen Storage Disease: Type IV	♂ Ashkenazi Jewish: 1/35	>99%	<1/3,500	Deficiency	O Luiopedii. 17 146	03.33%	1/000
	o' General: 1/461	18.60%	1/566		o ^r Japanese: 1/159	76.92%	1/689
Glycogen Storage Disease: Type V	o ^a Caucasus Jewish: Unknown	>99%	Unknown	Homocystinuria Caused by CBS Deficiency	o" European: 1/224	64.29%	1/627
	o [™] European: 1/159	60.71%	1/405		o" Irish: 1/128	70.59%	1/435
	o' General: Unknown	74.10%	Unknown		o" Italian: 1/224	35.71%	1/348
	o" Spaniard: 1/159	67.11%	1/483		o" Norwegian: 1/41	84.38%	1/262
	o [*] Yemenite Jewish: Unknown	75.00%	Unknown		o" Qatari: 1/22	>99%	<1/2,200
Glycogen Storage Disease: Type VII	o" Ashkenazi Jewish: 1/250	>99%	<1/25,00		o" Saudi Arabian: Unknown	92.31%	Unknown
			0	Hurler Syndrome	o" Czech: 1/190	52.50%	1/400
Guanidinoacetate Methyltransferase Deficiency	o [*] General: Unknown	29.41%	Unknown		o [*] European: 1/194	81.71%	1/1,061
HMG-CoA Lyase Deficiency	o' General: 1/159	40.00%	1/265		o' General: 1/194	62.50%	1/517
-	o [*] Japanese: Unknown	30.00%	Unknown		0" Italian: 1/194	61.11%	1/499
	o [®] Portuguese: Unknown	86.36%	Unknown		o ⁷ Japanese: 1/194	23.68%	1/254
	o" Saudi Arabian: Unknown	93.33%	Unknown		o" Moroccan Jewish: 1/194	92.31%	1/2,522
					o" Scandinavian: 1/194	79.41%	1/942

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
	o" Spaniard: 1/194	52.50%	1/408	Limb-Girdle Muscular Dystrophy: Type	o ^a Brazilian: Unknown	57.14%	Unknown
Hypophosphatasia	o ^a Canadian Amish: 1/26	>99%	<1/2,600	2E			. (=
	o [™] European: 1/159	19.23%	1/197		o [*] European: 1/539	25.00%	1/719
	o ^a Japanese: Unknown	54.55%	Unknown		o' General: Unknown	12.50%	Unknown
Inclusion Body Myopathy: Type 2	o'' General: Unknown	85.83%	Unknown		o" US Amish: Unknown	>99%	Unknown
	ơ" Iranian Jewish: 1/16	>99%	<1/1,600	Limb-Girdle Muscular Dystrophy: Type 2F	o ^r Brazilian: Unknown	>99%	Unknown
	o ^a Japanese: Unknown	71.88%	Unknown		o' General: Unknown	83.33%	Unknown
	o" Korean: Unknown	72.50%	Unknown	Limb-Girdle Muscular Dystrophy: Type	o" Brazilian: Unknown	34.62%	Unknown
Infantile Cerebral and Cerebellar Atrophy	o ^a Caucasus Jewish: 1/20	>99%	<1/2,000	21	o ^a Danish: 1/100	85.53%	1/691
Isolated Microphthalmia: VSX2 Related	o ^r Middle Eastern: Unknown	71.43%	Unknown		o" General: Unknown	43.18%	Unknown
Isovaleric Acidemia	o" General: 1/251	47.37%	1/477		o [®] German: 1/300	82.50%	1/1,714
Joubert Syndrome	♂ Ashkenazi Jewish: 1/92	>99%	<1/9,200	Lipoprotein Lipase Deficiency	o [®] French Canadian: 1/44	28.95%	1/62
Lamellar Ichthyosis: Type 1	o" Norwegian: 1/151	81.40%	1/812		o" General: Unknown	20.00%	Unknown
Laryngoonychocutaneous Syndrome	o" Pakistani: Unknown	>99%	Unknown	Long-Chain 3-Hydroxyacyl-CoA	o [®] European: 1/126	88.98%	1/1,144
Leber Congenital Amaurosis: CEP290 Related	o" European: 1/251	47.32%	1/476	Dehydrogenase Deficiency	, <i>,</i>		
Leber Congenital Amaurosis: GUCY2D	o" Finnish: Unknown	>99%	Unknown		of General: 1/126	56.25%	1/288
Related	o ^a Pakistani: Unknown		Unknown	Lysinuric Protein Intolerance	ơ ^a Finnish: 1/123	>99%	<1/12,30 0
Leber Congenital Amaurosis: LCA5 Related	O Pakistani: Unknown	83.33%	Unknown		o" Italian: 1/120	45.45%	1/220
Leber Congenital Amaurosis: RDH12	o' General: 1/560	38.37%	1/909		o [*] Japanese: 1/115	37.93%	1/185
Related					o [®] North African: Unknown	>99%	Unknown
Leigh Syndrome: French-Canadian	o [*] French Canadian: 1/23	95.45%	1/506	MTHFR Deficiency: Severe	o" Bukharan Jewish: 1/39	>99%	<1/3,900
Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related	o'' Cree: Unknown	>99%	Unknown	Malonyl-CoA Decarboxylase Deficiency	o ^a General: Unknown	33.33%	Unknown
	o [®] European: Unknown	65.22%	Unknown	Maple Syrup Urine Disease: Type 1A	o" US Amish: 1/10	97.73%	1/440
Leydig Cell Hypoplasia (Luteinizing	o ^r Brazilian: Unknown	>99%	Unknown	Maple Syrup Urine Disease: Type 1B	o" Ashkenazi Jewish: 1/97	>99%	<1/9,700
Hormone Resistance)	-70 1//1	(1.4/0)	1 (150	Maple Syrup Urine Disease: Type 2	o" General: 1/481	42.31%	1/834
Limb-Girdle Muscular Dystrophy: Type 2A	O' Basque: 1/61	61.46%	1/158		o'' Norwegian: 1/481	50.00%	1/962
	o" Croatian: 1/133	76.00%	1/554		o" Turkish: 1/112	58.33%	1/269
	o" European: 1/103	17.23%	1/124	Maple Syrup Urine Disease: Type 3	o" Ashkenazi Jewish: 1/94	>99%	<1/9,400
	o" General: 1/103	26.47%	1/140		o" General: Unknown	68.75%	Unknown
	o ^a Italian: 1/162	35.71%	1/252	Maroteaux-Lamy Syndrome	o" Argentinian: 1/274	75.00%	1/1,096
	o ^r Russian: 1/103	53.33%	1/221		o" General: 1/388	61.54%	1/1,009
	o ^r US Amish: Unknown	>99%	Unknown		o" Spaniard: 1/274	29.17%	1/387
Limb-Girdle Muscular Dystrophy: Type	o ^r Caucasus Jewish: 1/25	>99%	<1/2,500	Meckel Syndrome: Type 1	o" European: 1/212	72.22%	1/763
2B			, ,		ơ" Finnish: 1/48	>99%	<1/4,800
	o ^a Libyan Jewish: 1/19	>99%	<1/1,900	Medium-Chain Acyl-CoA	o" European: 1/50	90.91%	1/550
Limb-Girdle Muscular Dystrophy: Type 2C	d ^a European Gypsy: 1/50	>99%	<1/5,000	Dehydrogenase Deficiency	♂ Saudi Arabian: 1/68	95.00%	1/1,360
	o'' General: Unknown	60.00%	Unknown		o ^r United Kingdom: 1/51	90.00%	1/510
	o ^a Tunisian: Unknown	>99%	Unknown	Megalencephalic	o ^r Japanese: Unknown	50.00%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2D	o ^a Brazilian: Unknown	64.29%	Unknown	Leukoencephalopathy			
	♂ European: 1/288	22.22%	1/370		o ^a Libyan Jewish: 1/40	>99%	<1/4,000
	of European: 1/200 of Finnish: 1/150	95.45%	1/3,300		o ^a Turkish: Unknown	20.00%	Unknown
	·			Metachromatic Leukodystrophy	o [*] European: 1/150	43.88%	1/267
	o" General: Unknown	26.09%	Unknown		ð" Habbanite Jewish: 1/5	50.00%	1/10

Disease	Carrier Rate	Detection	Residual
		Rate	Risk
Methylmalonic Acidemia: MMAA Related	o'' General: 1/274	63.51%	1/751
Methylmalonic Acidemia: MMAB Related	o" General: 1/396	71.25%	1/1,377
Methylmalonic Acidemia: MUT Related	ð" General: 1/177	43.62%	1/314
Methylmalonic Aciduria and Homocystinuria: Type cblC	o ^a Chinese: Unknown	61.39%	Unknown
	ð" General: 1/159	65.74%	1/464
	o ^a Italian: Unknown	75.00%	Unknown
	o ^r Portuguese: Unknown	91.18%	Unknown
Mitochondrial Complex I Deficiency: NDUFS6 Related	o ^a Caucasus Jewish: 1/24	>99%	<1/2,400
Mitochondrial DNA Depletion Syndrome: MNGIE Type	o" Ashkenazi Jewish: Unknown	>99%	Unknown
	o ^a General: Unknown	47.37%	Unknown
	o ^a Iranian Jewish: Unknown	>99%	Unknown
Mitochondrial Myopathy and Sideroblastic Anemia	o ^r Iranian Jewish: Unknown	>99%	Unknown
Mitochondrial Trifunctional Protein Deficiency: HADHB Related	o ^r Japanese: Unknown	60.00%	Unknown
Morquio Syndrome: Type A	o ^a Colombian: 1/257	85.00%	1/1,713
	o" European: 1/257	20.97%	1/325
	o ^a Finnish: 1/257	50.00%	1/514
	o" Latin American: 1/257	36.11%	1/402
Morquio Syndrome: Type B	o ⁷ European: Unknown	83.33%	Unknown
Mucolipidosis: Type II/III	o" General: 1/158	24.60%	1/210
	o" Japanese: 1/252	51.25%	1/517
	o ^r Korean: Unknown	30.00%	Unknown
	o [*] Portuguese: 1/176	50.00%	1/352
Mucolipidosis: Type IV	♂ Ashkenazi Jewish: 1/97	96.15%	1/2,522
Multiple Pterygium Syndrome	o ^r European: Unknown	41.67%	Unknown
	o ^a Middle Eastern: Unknown	60.00%	Unknown
	o ^r Pakistani: Unknown	50.00%	Unknown
Multiple Sulfatase Deficiency	ð [•] Ashkenazi Jewish: 1/320	95.00%	1/6,400
	o" General: 1/501	18.18%	1/612
Muscle-Eye-Brain Disease	o ⁷ European: Unknown	54.17%	Unknown
	o" Finnish: 1/112	97.37%	1/4,256
	o'' General: Unknown	23.53%	Unknown
	o ⁷ United States: Unknown	25.00%	Unknown
Navajo Neurohepatopathy	0 [°] Navajo: 1/39	>99%	<1/3,900
Nemaline Myopathy: NEB Related	o ^a Ashkenazi Jewish: 1/108	>99%	<1/10,80 0
Nephrotic Syndrome: Type 1	o [®] Finnish: 1/45	76.84%	1/194
	♂ US Amish: 1/12	50.00%	1/24
Nephrotic Syndrome: Type 2	o ⁷ Israeli-Arab: Unknown	55.56%	Unknown
	o ^a Pakistani: Unknown	20.00%	Unknown
	o ^a Polish: Unknown	16.18%	Unknown
	o" Saudi Arabian: Unknown	72.73%	Unknown

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Disease	Carrier Rate	Detection Rate	Residual Risk
Neuronal Ceroid-Lipofuscinosis: CLN5 Related	o" Finnish: 1/101	>99%	<1/10,10 0
Neuronal Ceroid-Lipofuscinosis: CLN6 Related	o" European: 1/159	36.36%	1/250
	o" General: 1/159	59.52%	1/393
	o ^a Portuguese: 1/128	81.00%	1/674
Neuronal Ceroid-Lipofuscinosis: CLN8 Related	o" Finnish: 1/135	>99%	<1/13,50 0
	o" Italian: 1/212	33.33%	1/318
	o" Turkish: Unknown	77.78%	Unknown
Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	ð" General: 1/159	56.25%	1/363
Neuronal Ceroid-Lipofuscinosis: PPT 1 Related	o" Finnish: 1/58	97.62%	1/2,436
	o" General: 1/159	72.50%	1/578
Neuronal Ceroid-Lipofuscinosis: TPP1 Related	ơ' Canadian: 1/159	67.50%	1/489
	o" European: 1/159	75.00%	1/636
	o" General: 1/159	50.00%	1/318
	o" Newfoundlander: 1/43	85.29%	1/292
Niemann-Pick Disease: Type A	o" Ashkenazi Jewish: 1/101	95.00%	1/2,020
Niemann-Pick Disease: Type B	o [*] Czech: 1/276	83.33%	1/1,656
	o'' General: Unknown	19.82%	Unknown
	o [*] North African: Unknown	86.67%	Unknown
	o" Spaniard: Unknown	38.10%	Unknown
Niemann-Pick Disease: Type C1	o' Acadian: Unknown	>99%	Unknown
	o' General: 1/194	15.60%	1/230
	o ^a Japanese: Unknown	18.18%	Unknown
	♂ Portuguese: 1/194	25.00%	1/259
Niemann-Pick Disease: Type C2	o' General: 1/194	75.00%	1/776
Nijmegen Breakage Syndrome	0 ^ª Eastern European: 1/155	>99%	<1/15,50 0
Nonsyndromic Hearing Loss and Deafness: GJB2 Related	o" Ashkenazi Jewish: 1/20	95.83%	1/480
	♂ Chinese: 1/100	82.26%	1/564
	o" European: 1/53	82.47%	1/302
	o" Ghanaian: Unknown	90.91%	Unknown
	o" Indian: Unknown	66.98%	Unknown
	o" Israeli: 1/16	93.10%	1/232
	O [*] Japanese: 1/75	75.00%	1/300
	o" Roma: Unknown	>99%	Unknown
	o [*] United States: 1/34	45.22%	1/62
Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related	0 [°] Ashkenazi Jewish: 1/180	>99%	<1/18,00 0
Nonsyndromic Hearing Loss and Deafness: MYO 15A Related	o" Balinese: 1/6	>99%	<1/600
	o" Pakistani: 1/77	24.00%	1/101
Oculocutaneous Albinism: Type 1	o" European: 1/101	26.32%	1/137
	o [*] Hutterite: 1/7	>99%	<1/700
	o" Moroccan Jewish: 1/30	71.88%	1/107

Disease	Carrier Rate	Detection Rate	Residual Risk
	o" Puerto Rican: Unknown	91.67%	Unknown
Oculocutaneous Albinism: Type 3	o" Black South African: 1/47	94.74%	1/893
Oculocutaneous Albinism: Type 4	o ^r Japanese: 1/146	58.33%	1/350
Omenn Syndrome: DCLRE1C Related	ơ' Apache: 1/29	>99%	<1/2,900
	o" Navajo: 1/29	97.22%	1/1,044
Omenn Syndrome: RAG2 Related	o" Arab: Unknown	40.00%	Unknown
	o ^a Non-Ashkenazi Jewish: Unknown	70.00%	Unknown
Ornithine Translocase Deficiency	o' French Canadian: 1/20	95.00%	1/400
	o" Italian: Unknown	18.75%	Unknown
	o ^r Japanese: Unknown	60.00%	Unknown
Osteopetrosis: TCIRG1 Related	♂ Ashkenazi Jewish: 1/350	>99%	<1/35,00 0
	o ^r Costa Rican: Unknown	>99%	Unknown
	o' General: 1/251	25.00%	1/335
POLG Related Disorders: Autosomal Recessive	o" Belgian: Unknown	85.00%	Unknown
	o ^a Finnish: 1/140	>99%	<1/14,00 0
	o ^r General: Unknown	93.10%	Unknown
	o ^r Norwegian: Unknown	>99%	Unknown
Papillon-Lefevre Syndrome	o ^r General: Unknown	35.29%	Unknown
	o ^r Indian Jewish: Unknown	>99%	Unknown
	o ^r Turkish: Unknown	50.00%	Unknown
Pendred Syndrome	o [*] European: 1/58	42.11%	1/100
	o ^r Japanese: Unknown	45.83%	Unknown
	o ^r Pakistani: Unknown	29.82%	Unknown
Persistent Mullerian Duct Syndrome: Type I	o ^a General: Unknown	28.12%	Unknown
Persistent Mullerian Duct Syndrome: Type II	o ^a General: Unknown	78.12%	Unknown
Phenylalanine Hydroxylase Deficiency	o" Arab: Unknown	46.08%	Unknown
	o" Ashkenazi Jewish: 1/224	44.44%	1/403
	o" Brazilian: 1/71	56.41%	1/163
	o ^r Chinese: 1/51	76.57%	1/218
	o" Cuban: 1/71	69.64%	1/234
	o" European: 1/51	73.00%	1/189
	o" French Canadian: 1/80	76.27%	1/337
	o" Iranian: 1/31	66.94%	1/94
	o" Korean: 1/51	51.52%	1/105
	ơ" Non-Ashkenazi Jewish: Unknown	63.64%	Unknown
	o" Slovakian Gypsy: 1/39	>99%	<1/3,900
	o" Spanish Gypsy: 1/4	93.75%	1/64
	o ^r Taiwanese: Unknown	83.10%	Unknown
	o" US Amish: 1/16	86.84%	1/122
Polyglandular Autoimmune Syndrome: Type l	o ^a Finnish: 1/80	90.48%	1/840
	ơ" Iranian Jewish: 1/48	>99%	<1/4,800

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Disease	Carrier Rate	Detection Rate	Residual Risk
	o" Italian: Unknown	27.78%	Unknown
	o" Norwegian: 1/142	47.92%	1/273
	o" Sardinians: 1/61	81.82%	1/336
	o ^a United Kingdom: Unknown	70.00%	Unknown
	o [*] United States: Unknown	65.62%	Unknown
Pontocerebellar Hypoplasia: EXOSC3 Related	♂ General: Unknown	83.33%	Unknown
Pontocerebellar Hypoplasia: RARS2 Related	o ^a Sephardic Jewish: Unknown	>99%	Unknown
Pontocerebellar Hypoplasia: SEPSECS Related	0 ^a Iraqi Jewish: 1/42	>99%	<1/4,200
Pontocerebellar Hypoplasia: TSEN54 Related	o" European: 1/250	95.65%	1/5,750
Pontocerebellar Hypoplasia: VPS53 Related	0 [°] Moroccan Jewish: 1/37	>99%	<1/3,700
Pontocerebellar Hypoplasia: VRK 1 Related	o ^a Ashkenazi Jewish: 1/225	>99%	<1/22,50 0
Primary Carnitine Deficiency	o" European: 1/101	58.33%	1/242
	O [™] Faroese: 1/9	53.95%	1/20
	o" General: Unknown	20.22%	Unknown
Primary Ciliary Dyskinesia: DNAI1 Related	o" European: 1/211	52.38%	1/443
Primary Ciliary Dyskinesia: DNAI2 Related	o" Ashkenazi Jewish: 1/200	>99%	<1/20,00 0
Primary Congenital Glaucoma	o" Moroccan: Unknown	>99%	Unknown
	o" Saudi Arabian: 1/23	91.67%	1/276
	o" Turkish: 1/51	70.59%	1/173
Primary Hyperoxaluria: Type 1	o [*] Dutch: 1/174	62.12%	1/459
	o" General: 1/189	52.68%	1/399
Primary Hyperoxaluria: Type 2	o" General: Unknown	70.31%	Unknown
Primary Hyperoxaluria: Type 3	♂ Ashkenazi Jewish: Unknown	>99%	Unknown
	o" European: Unknown	25.00%	Unknown
Progressive Familial Intrahepatic Cholestasis: Type 2	ơ ^a European: Unknown	33.33%	Unknown
Propionic Acidemia: PCCA Related	o [*] Japanese: 1/102	86.67%	1/765
Propionic Acidemia: PCCB Related	o" General: 1/182	42.86%	1/319
	o" Greenlandic Inuit: 1/16	58.33%	1/38
	o ^a Japanese: 1/102	78.00%	1/464
	o" Korean: Unknown	56.25%	Unknown
	o" Latin American: 1/182	75.00%	1/728
	o" Spaniard: 1/182	52.38%	1/382
Pseudocholinesterase Deficiency	o" General: 1/33	65.00%	1/94
	0ª Iranian Jewish: 1/9	>99%	<1/900
Pycnodysostosis	o" Danish: Unknown	87.50%	Unknown
Pyruvate Carboxylase Deficiency	o" General: 1/251	62.50%	1/669
	o" Native American: 1/10	>99%	<1/1,000
Pyruvate Dehydrogenase Deficiency	o'' General: Unknown	50.00%	Unknown

CarrierMap™

Disease	Carrier Rate	Detection	Residual
		Rate	Risk
Renal Tubular Acidosis and Deafness	a' Colombian (Antioquia): Unknown	92.86%	Unknown
Retinal Dystrophies: RLBP1 Related	o" Newfoundlander: 1/106	>99%	<1/10,60 0
	o" Swedish: 1/84	>99%	<1/8,400
Retinal Dystrophies: RPE65 Related	o" Dutch: 1/32	>99%	<1/3,200
	o ^a North African Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: CERKL Related	o ^r Yemenite Jewish: Unknown	>99%	Unknown
Retinitis Pigmentosa: DHDDS Related	0 ^a Ashkenazi Jewish: 1/91	>99%	<1/9,100
Retinitis Pigmentosa: FAM 161 A Related	o ^a Ashkenazi Jewish: Unknown	>99%	Unknown
	♂ Non-Ashkenazi Jewish: 1/32	>99%	<1/3,200
Rhizomelic Chondrodysplasia Punctata: Type I	o [®] General: 1∕159	72.68%	1/582
Salla Disease	o [*] European: Unknown	33.33%	Unknown
	o" Scandinavian: 1/200	94.27%	1/3,491
Sandhoff Disease	o ^a Argentinian: Unknown	95.45%	Unknown
	o" Cypriot: 1/7	80.00%	1/35
	o ^r Italian: Unknown	29.17%	Unknown
	o ^r Spaniard: Unknown	64.29%	Unknown
Sanfilippo Syndrome: Type A	ð Australasian: 1/119	44.12%	1/213
	o [*] Dutch: 1/78	63.10%	1/211
	o" European: 1/159	35.16%	1/245
	o [*] United States: 1/159	32.14%	1/234
Sanfilippo Syndrome: Type B	ð Australasian: 1/230	28.00%	1/319
	o ^r Dutch: Unknown	42.31%	Unknown
	o" European: Unknown	52.38%	Unknown
	ð Japanese: 1/200	81.82%	1/1,100
Sanfilippo Syndrome: Type C	o ^a Dutch: 1/346	75.00%	1/1,384
	ď Greek: 1/415	25.00%	1/553
	o" Moroccan: Unknown	80.00%	Unknown
	o" Spaniard: Unknown	64.29%	Unknown
Sanfilippo Syndrome: Type D	ð General: 1/501	83.33%	1/3,006
Short-Chain Acyl-CoA Dehydrogenase Deficiency	o ^a Ashkenazi Jewish: 1/15	65.00%	1/43
Sickle-Cell Anemia	o" African American: 1/10	>99%	<1/1,000
	o ^r Hispanic American: 1/95	>99%	<1/9,500
Sjogren-Larsson Syndrome	o ^r Dutch: Unknown	25.86%	Unknown
	ơ¹ Swedish: 1/205	>99%	<1/20,50 0
Sly Syndrome	o" General: 1/251	35.71%	1/390
Smith-Lemli-Opitz Syndrome	o [®] Brazilian: 1/94	79.17%	1/451
	o [*] European: 1/71	84.72%	1/465
	o ⁷ Japanese: Unknown	71.43%	Unknown
	o [*] United States: 1/70	95.00%	1/1,400
Stargardt Disease	o [*] General: 1/51	17.51%	1/62
Stuve-Wiedemann Syndrome	ơ" Emirati: 1/70	>99%	<1/7,000

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Disease	Carrier Rate	Detection Rate	Residual Risk
	o'' General: Unknown	75.00%	Unknown
Sulfate Transporter-Related Osteochondrodysplasia	ơ ^r Finnish: 1/51	95.83%	1/1,224
	o' General: 1/100	70.00%	1/333
Tay-Sachs Disease	o" Argentinian: 1/280	82.35%	1/1,587
	o" Ashkenazi Jewish: 1/29	99.53%	1/6,177
	o" Cajun: 1/30	>99%	<1/3,000
	o" European: 1/280	25.35%	1/375
	o" General: 1/280	32.09%	1/412
	o" Indian: Unknown	85.71%	Unknown
	o" Iraqi Jewish: 1/140	56.25%	1/320
	o" Japanese: 1/127	82.81%	1/739
	o" Moroccan Jewish: 1/110	22.22%	1/141
	o [®] Portuguese: 1/280	92.31%	1/3,640
	o" Spaniard: 1/280	67.65%	1/865
	o" United Kingdom: 1/161	71.43%	1/564
Trichohepatoenteric Syndrome: Type 1	o" European: 1/434	42.86%	1/760
	o" South Asian: 1/434	66.67%	1/1,302
Tyrosine Hydroxylase Deficiency	o' General: Unknown	36.11%	Unknown
Tyrosinemia: Type I	♂ Ashkenazi Jewish: 1/158	>99%	<1/15,80 0
	o" European: 1/166	57.14%	1/387
	o" Finnish: 1/123	97.22%	1/4,428
	o" French Canadian: 1/64	96.30%	1/1,728
	o" Pakistani: Unknown	92.86%	Unknown
Tyrosinemia: Type II	o" General: 1/251	40.00%	1/418
Usher Syndrome: Type 1B	o' European: 1/166	39.29%	1/273
	o" General: 1/143	12.89%	1/164
	o'' North African: Unknown	66.67%	Unknown
	o" Spaniard: 1/152	12.16%	1/173
Usher Syndrome: Type 1C	o" Acadian: 1/82	98.86%	1/7,216
	o" French Canadian: 1/227	83.33%	1/1,362
Usher Syndrome: Type 1D	o' General: 1/296	23.17%	1/385
Usher Syndrome: Type 1F	♂ Ashkenazi Jewish: 1/126	93.75%	1/2,016
Usher Syndrome: Type 2A	o [*] Chinese: Unknown	83.33%	Unknown
	o" European: 1/136	40.00%	1/227
	o ^a French Canadian: Unknown	66.67%	Unknown
	o [*] General: 1/136	46.92%	1/256
	o'' Japanese: Unknown	55.56%	Unknown
	o ^a Non-Ashkenazi Jewish: Unknown	61.11%	Unknown
	o" Scandinavian: 1/125	39.22%	1/206
	o" Spaniard: 1/133	39.02%	1/218
Usher Syndrome: Type 3	o" Ashkenazi Jewish: 1/120	>99%	<1/12,00 0
	o" Finnish: 1/134	>99%	<1/13,40 0

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Disease	Carrier Rate	Detection Rate	Residual Risk
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	o" General: 1/87	65.28%	1/251
Walker-Warburg Syndrome	♂ Ashkenazi Jewish: 1/150	>99%	<1/15,00 0
Werner Syndrome	o" General: 1/224	31.25%	1/326
	o" Japanese: 1/87	65.62%	1/253
Wilson Disease	♂ [®] Ashkenazi Jewish: 1/100	>99%	<1/10,00 0
	o" Canarian: 1/26	68.75%	1/83
	o" Chinese: 1/51	55.97%	1/116
	o" Cuban: Unknown	22.22%	Unknown
	o" European: 1/93	41.64%	1/159
	o ^a Greek: 1/90	44.94%	1/163
	o" Korean: 1/88	51.53%	1/182
	o" Spaniard: 1/93	38.18%	1/150
Wolcott-Rallison Syndrome	o" Saudi Arabian: Unknown	66.67%	Unknown
Wolman Disease	o" Iranian Jewish: 1/33	>99%	<1/3,300
Xeroderma Pigmentosum: Group A	o" Japanese: 1/75	97.62%	1/3,150
	o" North African: Unknown	87.50%	Unknown
	ơ" Tunisian: 1/112	90.91%	1/1,232
Xeroderma Pigmentosum: Group C	o" Moroccan: 1/71	76.19%	1/298
	ơ" Tunisian: 1/51	>99%	<1/5,100
Zellweger Spectrum Disorders: PEX1 Related	o" European: 1/139	70.27%	1/468
	o" General: 1/139	67.84%	1/432
Zellweger Spectrum Disorders: PEX10 Related	o ^a Japanese: Unknown	40.74%	Unknown
Zellweger Spectrum Disorders: PEX2 Related	o ^a Ashkenazi Jewish: 1/123	>99%	<1/12,30 0
Zellweger Spectrum Disorders: PEX6 Related	o" General: 1/288	30.00%	1/411



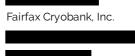


Patient Information Name: Donor 5427 Date of Birth: Sema4 ID: Client ID: Indication: Carrier Screening

Specimen Information

Specimen Type: Purified DNA Date Collected: 12/02/2021 Date Received: 12/07/2021 Final Report: 12/18/2021

Referring Provider



Custom Carrier Screen (5 genes)

with Personalized Residual Risk

SUMMARY OF RESULTS AND RECOMMENDATIONS

Positive	⊖ Negative
Carrier of Nephrotic Syndrome (<i>NPHS2</i> -Related) / Steroid- Resistant Nephrotic Syndrome (AR)	Negative for all other genes tested: DHCR7, PPT1, SLC26A4, and CYP21A2
Associated gene(s): NPHS2	To view a full list of genes and diseases tested
Variant(s) Detected: c.686G>A, p.R229Q, Pathogenic, Heterozygous (one copy)	please see Table 1 in this report

AR=Autosomal recessive; XL=X-linked

Recommendations

- Testing the partner for the above positive disorder(s) and genetic counseling are recommended.
- Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder.

Interpretation of positive results

Nephrotic Syndrome (NPHS2-Related) / Steroid-Resistant Nephrotic Syndrome (AR)

Results and Interpretation

A heterozygous (one copy) pathogenic missense variant, c.686G>A, p.R229Q, was detected in the *NPHS2* gene (NM_014625.3). Please note that this is a mild variant that is only expected to cause disease when found in trans with one of a specific set of variants that occurs in exons 7 or 8. Please see the disease interpretation below for additional information. Homozygotes are not expected to be affected, unless this variant is part of a more complex allele. When this variant is present in trans with a pathogenic variant, it is considered to be causative for an *NPHS2*-related disorder. Therefore, this individual is expected to be at least a carrier for an *NPHS2*-related disorder. Heterozygous carriers are not expected to exhibit symptoms of this disease.

What is Nephrotic Syndrome (NPHS2-Related) / Steroid-Resistant Nephrotic Syndrome?

Pathogenic variants in the *NPHS2* gene cause two autosomal recessive, pan-ethnic disorders: steroid-resistant nephrotic syndrome and focal segmental glomerulosclerosis.

- Steroid-resistant nephrotic syndrome (SRNS) is a severe disorder with onset usually occurring during childhood. Patients lose protein in their urine, which results in progressive kidney failure. Death will occur without a kidney transplant, usually by adolescence; however, many patients are cured after kidney transplant.
- Focal segmental glomerulosclerosis (FSGS) is a type of scarring of the kidney, and is usually diagnosed in the patient's second or third decade of life. FSGS is more slowly progressing than SRNS and usually leads to end-stage renal disease by the ages of 10-50.



Carrier screening report Donor 5427 Date of Birth: Sema4 ID:

Mutations in *NPHS2* have been demonstrated to have a complex genotype-phenotype correlation. A common pathogenic variant, p.R229Q, causes FSGS when found in trans with a number of specific variants, including p.A284V, p.A288T, p.R291W, p.A297V, p.E310K, p.E310V, p.L327F, p.Q328R, and p.F344LfsX4. While all of the variants that are disease-causing when in trans with R229Q are located in exons 7 and 8, not all pathogenic variants in exons 7 and 8 cause disease when in trans with R229Q. Examples of variants in exons 7 and 8 that do not cause disease when in trans with R229Q are p.R286TfsX17, p.V290M, and p.A317LfsX31. Additionally, p.R229Q is not disease-causing in the homozygous state (PMID: 24509478 and 29660491).

Test description

This patient was tested for a panel of diseases using a combination of sequencing, targeted genotyping and copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see Table 1 for a list of genes and diseases tested with the patient's personalized residual risk. If personalized residual risk is not provided, please see the complete residual risk table at **go.sema4.com/residualrisk**. Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

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



Genes and diseases tested

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

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

	Disease	Gene	Inheritance Pattern	Status	Detailed Summary
۲	Positive				
	Nephrotic Syndrome (<i>NPHS2</i> -Related) / Steroid-Resistant Nephrotic Syndrome	NPHS2	AR	Carrier	c.686G>A, p.R229Q, Pathogenic, Heterozygous (one copy)
Θ	Negative				
	Congenital Adrenal Hyperplasia due to 21- Hydroxylase Deficiency	CYP21A2	AR	Reduced Risk	CYP21A2 copy number: 2 CYP21A2 sequencing: Negative Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Non-Classic)): 1 in 200 Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Classic)): 1 in 1,300
	Neuronal Ceroid-Lipofuscinosis (PPT1-Related)	PPT1	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
	Pendred Syndrome	SLC26A4	AR	Reduced Risk	Personalized Residual Risk: 1 in 390
	Smith-Lemli-Opitz Syndrome	DHCR7	AR	Reduced Risk	Personalized Residual Risk: 1 in 750

AR=Autosomal recessive; XL=X-linked

Test methods and comments

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

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

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

Genotyping (Analytical Detection Rate >99%)

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

Multiplex Ligation-Dependent Probe Amplification (MLPA) (Analytical Detection Rate >99%)

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

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

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

For congenital adrenal hyperplasia, the copy number of the *CYP21A2* gene was analyzed. This analysis can detect large deletions typically due to unequal meiotic crossing-over between *CYP21A2* and the pseudogene *CYP21A1P*. Classic 30-kb deletions make up approximately 20% of *CYP21A2* pathogenic alleles. This test may also identify certain point mutations in *CYP21A2* caused by gene conversion events between



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CYP21A2 and *CYP21A1P*. Some carriers may not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *CYP21A2* gene on one chromosome and loss of *CYP21A2* (deletion) on the other chromosome. Analysis of *CYP21A2* is performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For spinal muscular atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or individuals that carry an intragenic mutation in *SMN1*. Please also note that 2% of individuals diagnosed with SMA have a causative *SMN1* variant that occurred *de novo*, and therefore cannot be picked up by carrier screening in the parents. Analysis of *SMN1* is performed in association with short-read sequencing of exons 2a-7, followed by confirmation using long-range PCR (described below). The presence of the c.*3+80T>G (chr5:70,247,901T>G) variant allele in an individual with Ashkenazi Jewish or Asian ancestry is typically indicative of a duplication of *SMN1*. When present in an Ashkenazi Jewish or Asian individual with two copies of *SMN1*, c.*3+80T>G is likely indicative of a silent (2+0) carrier. In individuals with two copies of *SMN1* with African American, Hispanic or Caucasian ancestry, the presence or absence of c.*3+80T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

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

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

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

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

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

Exceptions: ABCD1 (NM_000033.3) exons 8 and 9; ACADSB (NM_001609.3) chr10:124,810,695-124,810,707 (partial exon 9); ADA (NM_000022.2) exon 1; ADAMTS2 (NM_014244.4) exon 1; AGPS (NM_003659.3) chr2:178,257,512-178,257,649 (partial exon 1); ALDH7A1 (NM_001182.4) chr5:125,911,150-125,911,163 (partial exon 7) and chr5:125,896,807-125,896,821 (partial exon 10); ALMS1 (NM_015120.4) chr2:73,612,990-73,613,041 (partial exon 1); APOPT1 (NM_ 032374.4) chr14:104,040,437-104,040,455 (partial exon 3); CDAN1 (NM_138477.2) exon 2; CEP152 (NM_014985.3) chr15:49,061,146-49,061,165 (partial exon 14) and exon 22; CEP290 (NM_025114.3) exon 5, exon 7, chr12:88,519,017-88,519,039 (partial exon 13), chr12:88,514,049-88,514,058 (partial exon 15), chr12:88,502,837-88,502,841 (partial exon 23), chr12:88,481,551-88,481,589 (partial exon 32), chr12:88,471,605-88,471,700 (partial exon 40); CFTR (NM_000492.3) exon 10; COL4A4 (NM_000092.4) chr2:227,942,604-227,942,619 (partial exon 25); COX10 (NM_001303,3) exon 6; CYP11B1 (NM_000497,3) exons 3-7; CYP11B2 (NM_000498,3) exons 3-7; DNA/2 (NM_023036.4) chr17:72,308,136-72,308,147 (partial exon 12); DOK7 (NM_173660.4) chr4:3,465,131-3,465,161 (partial exon 1) and exon 2; DUOX2 (NM_014080.4) exons 6-8; EIF2AK3 (NM_004836.5 exon 8; EVC (NM_153717.2) exon 1; F5 (NM_000130.4) chr1:169,551,662-169,551,679 (partial exon 2); FH (NM_000143.3) exon 1; GAMT (NM_000156.5 exon 1; GLDC (NM_000170.2) exon 1; GNPTAB (NM_024312.4) chr17:4.837,000-4.837,400 (partial exon 2); GNPTG (NM_032520.4) exon 1; GHR (NM_000163,4) exon 3; GYS2 (NM_021957.3) chr12:21,699,370-21,699,409 (partial exon 12); HGSNAT (NM_152419.2) exon 1; IDS (NM_000202.6) exon 3; ITGB4 (NM_000213.4) chr17:73,749,976-73,750,060 (partial exon 33); JAK3 (NM_000215.3) chr19:17,950,462-17,950,483 (partial exon 10); LIFR (NM_002310.5 exon 19; LMBRD1 (NM_018368.3) chr6:70.459.226-70.459.257 (partial exon 5), chr6:70.447.828-70.447.836 (partial exon 7) and exon 12; LYST (NM_000081.3) chr1:235,944,158-235,944,176 (partial exon 16) and chr1:235,875,350-235,875,362 (partial exon 43); MLYCD (NM_012213.2) chr16:83,933,242-83,933,282 (partial exon 1); MTR (NM_000254.2) chr1 237,024,418-237,024,439 (partial exon 20) and chr1:237,038,019-237,038,029 (partial exon 24); NBEAL2 (NM_015175.2) chr3 47,021,385-47,021,407 (partial exon 1); NEB (NM_001271208.1 exons 82-105; NPC1 (NM_000271.4)) chr18:21,123,519-21,123,538 (partial exon 14); NPHP1 (NM_000272.3) chr2:110,937,251-110,937,263 (partial exon 3); OCRL (NM_000276.3) chrX:128,674,450-128,674,460 (partial exon 1); PHKB (NM_000293,2) exon 1 and chr16:47,732,498-47,732,504 (partial exon 30); PIGN (NM_176787.4)



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chr18:59,815,547-59,815,576 (partial exon 8); *PIP5K1C* (NM_012398.2) exon 1 and chr19:3637602-3637616 (partial exon 17); *POU1F1* (NM_000306.3) exon 5; *PTPRC* (NM_002838.4) exons 11 and 23; *PUS1* (NM_025215.5 chr12:132,414,446-132,414,532 (partial exon 2); *RPGRIP1L* (NM_015272.2) exon 23; *SGSH* (NM_000199.3) chr17:78,194,022-78,194,072 (partial exon 1); *SLC6A8* (NM_005629.3) exons 3 and 4; *ST3GAL5* (NM_003896.3) exon 1; *SURF1* (NM_003172.3) chr9:136,223,269-136,223,307 (partial exon 1); *TRPM6* (NM_017662.4) chr9:77,362,800-77,362,811 (partial exon 31); *TSEN54* (NM_207346.2) exon 1; *TYR* (NM_000372.4) exon 5; *VWF* (NM_000552.3) exons 24-26, chr12:6,125,675-6,125,684 (partial exon 30), chr12:6,121,244-6,121,265 (partial exon 33), and exon 34.

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

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

Next Generation Sequencing for SMN1

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

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

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

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

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

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

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

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

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

Residual Risk Calculations

Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >30,000 variants and genomic frequency data from >138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining the *a priori* risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate. They are provided only as a guide for



assessing approximate risk given a negative result, and values will vary based on the exact ethnic background of an individual. This report does not represent medical advice but should be interpreted by a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

Personalized Residual Risk Calculations

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

Sanger Sequencing (Confirmation method) (Accuracy >99%)

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

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

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

Please note these tests were developed and their performance characteristics were determined by Sema4 Opco, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

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