

Donor 5189

Genetic Testing Summary

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

Last Updated: 09/06/18

Donor Reported Ancestry: German Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
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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 beta thalassemia, alpha thalassemia trait (aa/ and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by gene sequencing in the CFTR gene	1/1250
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Expanded genetic disease testing attached- 289 diseases by gene sequencing	Carrier: Usher Syndrome Type 3 (CLRN1) Negative for other genes sequenced	Carrier testing recommended for those using this donor

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

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



Carrier Map™

Partner Not Tested

Ordering Practice:

Practice Code:

Physician:

Report Generated: 2018-01-05 Report Updated: 2018-01-05 Donor 5189

OOB:
Gender: Male
Ethnicity: European
Procedure ID: 109739

Kit Barcode:

Specimen: Blood, #111705 Specimen Collection: 2017-12-14 Specimen Received: 2017-12-15 Specimen Analyzed: 2018-01-05

TEST INFORMATION

Test: CarrierMap $^{\text{SEQ}}$ (Genotyping &

Sequencing)

Panel: CarrierMap Expanded v3 -

Sequencing

Diseases Tested: 289 Genes Tested: 278 Genes Sequenced: 273

SUMMARY OF RESULTS: MUTATION(S) IDENTIFIED

Disease Donor 5189 Partner Not Tested

Usher Syndrome: Type 3 (CLRN1)

O High Impact

Carrier (1 abnormal copy)
Mutation: c.434-2A>T

Method: Sequencing

Reproductive Risk & Next Steps: Reproductive risk detected. Consider

partner testing

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

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

Assay performed by Reprogenetics

CLIA ID: 31 D 1054821

3 Regent Street, Livingston, NJ 07039

Lab Technician: Bo Chu

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





ADDITIONAL RESULTS: NO INCREASED REPRODUCTIVE RISK

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

Disease (Gene) Donor 5189 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

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

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





Usher Syndrome: Type 3 (CLRN1)

Usher syndrome type 3 causes progressive hearing and vision loss. In this disease, the CLRN1 gene responsible for nerve cell communication in the inner ear and retina is defective. Patients affected are born with normal vision and hearing, which deteriorate in the first few decades of life. Hearing loss typically begins after development of speech and progresses over time. Affected individuals also develop an eye disease called retinitis pigmentosa, which affects the light-sensitive tissue (retina) at the back of the eye, and causes night blindness and loss of peripheral and central vision. Affected individuals may also experience difficulty with balance and coordination due to abnormalities in the inner ear.

High Impact

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

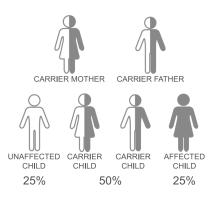
Clinical Information

Physical Impairment
 Cognitive Impairment
 Shortened Lifespan

Effective Treatment

Inheritance:

Autosomal Recessive



Prognosis

Prognosis for hearing and vision loss is generally unfavorable. Though affected individuals are born with normal hearing and vision, they experience progressive hearing and vision decline. Vision loss typically begins with night blindness, followed by loss of peripheral vision and eventually, loss of central vision. By middle age, most affected individuals have moderate to severe hearing loss and are legally blind. However, the rate of hearing and vision decline as well as the severity can vary among affected individuals. Usher syndrome type 3 does not cause other health problems and does not affect intelligence or life expectancy.

Treatment

There is no cure for Usher syndrome type 3. Treatments for hearing loss can include sign language, hearing aids and specialized instruction for the family on coping with hearing loss. Treatments for vision loss can include use of low vision aids and specialized instruction for the individual and family on coping with vision loss. Experimental therapies for retinitis pigmentosa, such as vitamin A palmitate supplements and lutein supplements can also be considered.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Ashkenazi Jewish	>99%	1/120	1/12000
Finnish	>99%	1/134	1/13400

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/usher-syndrome-type-3



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.



Carrier Map™

Diseases & Mutations Assayed

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

17-Alpha-Hydroxylase Deficiency (CYP17A1): Mutations (20): & Genotyping | c.157_159delTTC (p.53delF), c.316T>C (p.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): 07 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): σ Genotyping | c.293-13C>G

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

3-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD3B2): Mutations (6): 0° 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): σ 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): σ^a 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): & 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): o* 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): σ Genotyping | c.2593G>T (p.G865X), c.2211delT Sequencing | NM_000253:2-19

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

Acute Infantile Liver Failure: TRMU Related (TRMU): Mutations (5): σ 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): σ Genotyping | c.372delCATGCCCGCCTGGAACTT, c.832A>G (p.M278V), c.926A>G (p.Q309R), c.442C>T (p.R148X), c.532G>T (p.G178C) Sequencing | NM_004035:1-14

Adenosine Deaminase Deficiency (ADA): Mutations (22): ♂ Genotyping | c,986C>T (p.A329V), c.872C>T (p.S291L), c.646G>A (p.G216R), c.632G>A (p.R211H), c.631C>T (p.R211C), c.596A>C (p.Q199P), c.536C>A (p.A179D), c.529G>A (p.V177M), c.467G>A (p.R156H), c.466C>T (p.R156C), c.454C>A (p.L152M), c.445C>T (p.R149W), c.419G>A (p.G140E), c.385G>A (p.V129M), c.320T>C (p.L107P), c.302G>A (p.R101Q), c.302G>T (p.R101U), 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): O 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_351delCTCCCGGCGAG (p.L114_E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G

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

 $\label{eq:Alpha-Mannosidosis (MAN2B1): Mutations (3): \mathcal{O} Genotyping | c.2426T>C (p.1809P), c.2248C>T (p.R750W), c.1830+1G>C (p.V549_E610del) Sequencing | NM_000528:1-24$

Alport Syndrome: COL4A3 Related (COL4A3): Mutations (3): of 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 (5): & Genotyping | c.3713C>G (p.S1238X), c.4129C>T (p.R1377X), c.4715C>T (p.P1572L), c.4923C>A (p.C1641X), c.3601G>A (p.G1201S) Sequencing | NM_000092:2-48

Andermann Syndrome (SLC12A6): Mutations (5): σ 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): of Genotyping | c.859G>C (p.A287P), c.1615G>A (p.G539R), c.1475T>A (p.V492E), c.1370G>A (p.R457H) Sequencing | NM_000941:2-16

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

Argininosuccinate Lyase Deficiency (ASL): Mutations (7): 6° 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): d' Genotyping | c.1222delC, 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' Genotyping | c.1012A>G (p.S338G), c.514C>T (p.Q172X) Sequencing | NM_001271685:1-8

Asparagine Synthetase Deficiency (ASNS): Mutations (1): o' Genotyping | c.1084T>G

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

(p.F362V) Sequencing | NM_001673:3-13

Ataxia with Vitamin E Deficiency (TTPA): Mutations (14): 6° 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): o' Genotyping | c.5895insA (p.L1966fsX1969), c.9689delA (p.D3230fs), c.107C>T (p.T36M), c.1486C>T (p.R496X), c.10412T>G (p.V3471G), c.10658T>C (p.I3553T), c.10174C>T (p.Q3392X), c.9530T>C (p.I3177T), c.9053C>T (p.S3018F), c.8870T>C (p.I2957T), c.8011C>T (p.R2671X), c.6992T>A (p.I2331K), c.5221G>A (p.V1741M), c.4991C>T (p.S1664F), c.3761_3762delCCinsG (p.A1254fs), c.2414C>T (p.P805L), c.664A>G (p.I222V), c.10036T>C (p.C3346R), c.383delC, c.4220T>G (p.L1407R), c.11612G>A (p.W3871X), c.5984A>G (p.E1995G), c.10637delT (p.V3546fs), c.3747T>G (p.C1249W), c.5750A>G (p.Q1917R), c.10865G>A (p.C3622Y), c.50C>T (p.A17V), c.8063G>T (p.C2688F), c.10402A>G (p.I3468V), c.1529delG (p.G510fs), c.657C>T (p.G219G), c.5513A>G (p.Y1838C), c.1085delA (p.K3619fs), c.5381-9T>G (IVS33-9T>G), c.3229-2A>C (IVS28-2A>C), c.10505A>T (p.E3502V), c.2269A>C (p.I757L), c.4165C>A (p.P1389T), c.10364delC (p.S3455fs), c.7350+653A>G (IVS46+653A>G) Sequencing | NM_138694:2-67

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

Bardet-Biedl Syndrome: BBS10 Related (BBS10): Mutations (3): O Genotyping





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

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

Bardet-Biedl Syndrome: BBS12 Related (BBS12): Mutations (5): 0° 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): O⁷ 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): σ Genotyping | c.124_127delTTCT (p.F42Lfs), c.17_18delCT, c.20delA (p.E7Gfs), c.217insA (p.S73Kfs),

c.223+702_444+342del620insAAGTAGA, c.230delC, c.25_26delAA, c.315+1G>A, c.315+2T>C, c.316-197C>T, c.316-146T>G, c.315+745C>G, c.316-1G>A, c.316-1G>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, c.4delG [p.V2Cfs], c.51delC [p.K18Rfs], c.93-21G>A, c.92+1G>A, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>T, c.-50A>C, c.-78a>g, c.-79A>G, c.81A>G, c.52A>T (p.K18X), c.-137c>g, c.-138c>t, c.-151C>T, c.118C>T (p.Q40X), c.169G>C (p.G57R), c.295G>A (p.V99M), c.415G>C (p.A139P), c.47G>A (p.W16X), c.48G>A (p.W16X), c.801>a, c.2T>C, c.75T>A (p.G25G), c.444+111A>G, c.-29G>A, c.68_74delAAGTTGG, c.92G>C (p.R31T), c.92+1G>T, c.93-15T>G, c.93-1G>C, c.112delT, c.113G>A (p.W38X), c.114G>A (p.W38X), c.126delC, c.444+113A>G, c.250delG, c.225delC, c.383_385delAGG (p.Q128_A129delQAinsP), c.321_322insG (p.N109fs), c.316-1G>T, c.316-2A>C, c.287_288insA (p.197fs), c.271G>T (p.E91X), c.203_204delTG (p.V68Afs), c.154delC (p.P52fs), c.135delC (p.F46fs), c.92+2T>A, c.92+2T>C, c.90C>T (p.G30G), c.84_85insC (p.129fs), c.59A>G (p.N120S), c.46delT (p.W16Gfs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.27SG (p.M1R), 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° Genotyping | c.739C>T (p.R247W), c.745C>T (p.R249W) Sequencing | NM_000520:1-14

Beta-Ketothiolase Deficiency (ACAT1): Mutations (19): of 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.27>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.Y414Ifs), c.1240_1251delTATCTCCACGTC (p.Y414_V417del), c.278A>G (p.Y93C), c.595G>A (p.V199M), c.933delT (p.S311Rfs) Sequencing | NM_000060:1-4

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

Canavan Disease (ASPA): Mutations (8): O* 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.27>C (p.M1T), c.79G>A (p.G27R) Sequencing | NM_000049:1-6

Carnitine Palmitoyltransferase IA Deficiency (CPT1A): Mutations (10): of 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 (20): O* 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.R151C), c.520G>A (p.E174K), c.1148T>A (p.F383Y), c.1342T>C (p.F448L) Sequencing | NM_000098:1-5

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): of Genotyping | n.71A>G, c.263G>T Sequencing | NR_003051:1

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

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

Cholesteryl Ester Storage Disease (LIPA): Mutations (4): O 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): σ^a 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): of 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): of 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): σ 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): 6th 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): o" 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): of 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): 3 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_208delTG (p.E70Afs), c.1550G>A (p.G517E), c.717+4A>T, c.429-1G>C, c.1660delC (p.R554fs), c.2046+3A>C, c.2084C>T (p.P695L) Sequencing | NM 002529:2-17

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





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

Congenital Myasthenic Syndrome: CHRNE Related (CHRNE): Mutations (12): of 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): O* Genotyping | c.601C>T (p.R201X), c.539G>C (p.G180A), c.548_551delTCCT (p.F183fs), c.1263_1264insC (p.S422fs), c.101-1G>T, c.331+1G>T Sequencing | NM_173660:3-7

Congenital Myasthenic Syndrome: RAPSN Related (RAPSN): Mutations (11): of 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): 0° 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): O* Genotyping | c.1459_1462delTACGinsA (p.487_488delYAinsT), c.2313_2314insTATGACAC, c.554_561 delGCTTCGCC (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): o' Genotyping | c.1492A>G (p.T498A), c.541C>T (p.R181W), c.1382T>C (p.L461P) Sequencing | NM_000498:1-9

Crigler-Naijar 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): of Genotyping | c.1029delC, c.1153_1154insAT, c.1477delCA, c.1519_1521delATC (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. 1911delG, c. 1923delCTCAAAACTinsA, c. 1973delGAAATTCAATCCTinsAGAAA, c. 2052delA (p. K684fs), c.2052insA (p.Q685fs), c.2051_2052delAAinsG (p.K684SfsX38), c.2174insA, c.261delTT, c.2657+5G>A, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2988+1G>A, c.3039delC, c.3140-26A>G, c.325delTATinsG, c.3527delC, c.3535delACCA, c.3691delT, c.3717+12191C>T, c.3744delA, c.3773_3774insT (p.L1258fs), c.442delA, c.489+1G>T, c.531delT, c.579+1G>T, c.579+5G>A (IVS4+5G>A), c.803delA (p.N268fs), c.805_806delAT (p.I269fs), c.933_935delCTT (p.311delF), c.946delT, c.1645A>C (p.S549R), c.2128A>T (p.K710X), c.1000C>T (p.R334W), c.1013C>T (p.T338I), c.1364C>A (p.A455E), c.1477C>T (p.Q493X), c.1572C>A (p.C524X), c.1654C>T (p.Q552X), c.1657C>T (p.R553X), c.1721C>A (p.P574H), c.2125C>T (p.R709X), c.223C>T (p.R75X), c.2668C>T (p.Q890X), c.3196C>T (p.R1066C), c.3276C>G (p.Y1092X), c.3472C>T (p.R1158X), c.3484C>T (p.R1162X), c.349C>T (p.R117C), c.3587C>G (p.S1196X), c.3712C>T (p.Q1238X), c.3764C>A (p.S1255X), c.3909C>G (p.N1303K), c.1040G>A (p.R347H), c.1040G>C (p.R347P), c.1438G>T (p.G480C), c.1558G>T (p.V520F), c.1624G>T (p.G542X), c.1646G>A (p.S549N), c.1646G>T (p.S549I), c.1652G>A (p.G551D), c.1675G>A (p.A559T), c.1679G>C (p.R560T), c.178G>T (p.E60X), c.254G>A (p.G85E), c.271G>A (p.G91R), c.274G>T (p.E92X), c.3209G>A (p.R1070Q), c.3266G>A (p.W1089X), c.3454G>C (p.D1152H), c.350G>A (p.R117H), c.3611G>A (p.W1204X), c.3752G>A (p.S1251 N), c.3846G>A (p.W1282X), c.3848G>T (p.R1283M), c.532G>A (p.G 178R), c.988G>T (p.G330X), c.1090T>C (p.S364P), c.3302T>A (p.M 1101 K), 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.11023_V1024delT), c.3536_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.54-5940_273+10250del21080bp (p.S18fs), c.4364C>G (p.S1455X), c.4003C>T (p.L1335F), c.2538G>A (p.W846X), c.200C>T (p.P67L), c.4426C>T (p.Q1476X), c.1116+1G>A, c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y),

c.263T>G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.658C>T (p.Q220X), c.868C>T (p.Q290X), c.1626delG (p.G509fs), c.2908+1085-3367+260del7201, c.11C>A (p.S4X), c.3878_3881 delTATT (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_1611 delAC (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.L18X), 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): Of Genotyping | c.18_21 delGACT,

Cystinosis (CINS): Mulations (14): 6' Genotyping | c.18_21delGAC1, c.198_218delTATTACTATCCTTGAGCTCCC, c.283G>T (p.G95X), c.414G>A (p.W138X), c.506G>A (p.G169D), c.613G>A (p.D205N), c.473T>C (p.L158P), c.329G>T (p.G110V), c.416C>T (p.S139F), c.589G>A (p.G197R), c.969C>G (p.N323K), c.1015G>A (p.G339R), c.39155_848del57119, c.199_219delATTACTATCCTTGAGCTCCCC (p.I67_P73del) Sequencing | NM_001031681:1,3-13

Cystinuria: Non-Type I (SLC7A9): Mutations (15): o* 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): of Genotyping | c.1400T>C (p.M467T), c.2033T>C (p.I.678P), 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): 6 Genotyping | c.46G>A (p.G16S), c.63G>T (p.L21F), c.422_423delAG, c.652G>T (p.V218L), c.1369A>T (p.N457Y), c.1369A>G (p.N457D) Sequencing | NM_000414:1-24

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

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

Dyskeratosis Congenita: RTEL1 Related (RTEL1): Mutations (5): 0° 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): σ 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): of 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 (IV\$5+1G>C) Sequencing | NM 016346:1-8

Ethylmalonic Aciduria (ETHE1): Mutations (4): σ 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): & 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.l675L) Sequencing | NM_000111:2-21

Familial Dysautonomia (IKBKAP): Mutations (4): of 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): o* Genotyping | c.3989-9G>A, c.4159_4161 delTTC (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): 3° 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): of 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): O' 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): O^{*} 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): of Genotyping | c.2392C>T (p.R798X) Sequencing | NM_032043:2-20

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

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





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

GRACILE Syndrome (BCS1L): Mutations (12): 6 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): & 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): 6" 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_S232del5insTP), c.246A>G (p.182M), c.1161+6555_*9573del31670bp Sequencing | NM_000153:2-17

Glutaric Acidemia: Type I (GCDH): Mutations (8): O* Genotyping | c.1204C>T (p.R402W), c.1262C>T (p.R421V), c.743C>T (p.P248L), c.1093G>A (p.E365K), c.877G>A (p.A293T), c.1083-2A>C (IVS10-2A>C), c.680G>C (p.R227P), c.1198G>A (p.V400M) Sequencing | NM_000159:2-12

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

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

Glutaric Acidemia: Type IIC (ETFDH): Mutations (8): of 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): o* 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): σ 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): of 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): O* 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): O* 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): σ 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): 0* Genotyping | c.2128_2130delTTC (p.710delF), c.1627A>T (p.K543X), c.1628A>C (p.K543T), c.148C>T (p.R50X), c.255C>A (p.Y85X), c.613G>A (p.G205S), c.2392T>C (p.W798R), c.1827G>A (p.K609K), c.632delG (p.S211fs), c.808C>T (p.R270X) Sequencing | NM_005609:1-20

Glycogen Storage Disease: Type VII (PFKM): Mutations (4): & Genotyping | c.450+1G>A, c.329G>T (p.R110L), c.283C>T (p.R95X), c.2214delC (p.P739Qfs) Sequencing | NM 001166686:2-25

Guanidinoacetate Methyltransferase Deficiency (GAMT): Mutations (4): of 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): O* Genotyping | c.959G>T (p.G320V) Sequencing | NM_213653:2-4

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

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

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

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

Hemoglobinopathy: Hb O (HBB): Mutations (1): σ 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): & Genotyping | c.3416delT (p.L1139fs) Sequencing | NM_014844:2-20

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

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

Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2): Mutations (1): o' Genotyping | c.283C>T (p.R95X) Sequencing | NM_005562:1-23

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

Hermansky-Pudlak Syndrome: Type 3 (HPS3): Mutations (4): σ 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): O* 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): of Genotyping | c.1795+5G>A (IV\$10+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): 0* 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.T191 M), c.341C>T (p.A114V), c.969G>A (p.W324X) Sequencing | NM_001178008:3-17

Hurler Syndrome (IDUA): Mutations (8): O* 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): O 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

Infantile Cerebral and Cerebellar Atrophy (MED17): Mutations (1): of Genotyping | c.1112T>C (p.1371P) Sequencing | NM_004268:1-12

| Isolated Microphthalmia: VSX2 Related (VSX2): Mutations (4): O* 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): σ Genotyping | c.941C>T (p.A314V) Sequencing | NM_002225:1-12

Joubert Syndrome (TMEM216): Mutations (2): 3° Genotyping | c.218G>T (p.R73L), c.218G>A (p.R73H) Sequencing | NM_001173991:1-5

 $\begin{tabular}{ll} Lamellar Ichthyosis: Type 1 (TGM1): M totalions (1): σ Genotyping | c.877-2A>G (IVS5-2A>G) Sequencing | NM_000359:2-15 \\ \end{tabular}$

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

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

Leber Congenital Amaurosis: GUCY2D Related (GUCY2D): Mutations (3): of Genotyping | c.1694T>C (p.F565S), c.2943delG (p.G982V), c.387delC (p.P130Lfx) Sequencing | N.M. 000180-2-19





Leber Congenital Amaurosis: LCA5 Related (LCA5): Mutations (3): of Genotyping | c.835C>T (p.Q279X), c.1476_1477insA (p.P493TfsX1), c.1151delC Sequencing | NM 001122769:2-8

Leber Congenital Amaurosis: RDH12 Related (RDH12): Mutations (6): 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): σ Genotyping | c.1061C>T (p.A354V) Sequencing | NM_133259:1-38

Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related (EIF2B5): Mutations (9): O^{*} 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): 0° 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.1152T), c.537-3C>A Sequencing | NM_000233:1-11

Limb-Girdle Muscular Dystrophy: Type 2A (CAPN3): Mutations (6): 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): & 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): σ ' 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): σ Genotyping | c.229C>T (p.R77C) Sequencing | NM_000023:1-9

Limb-Girdle Muscular Dystrophy: Type 2E (SGCB): Mutations (6): o* 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): 3° Genotyping | c.644G>A (p.G215E) Sequencing | NM_000237:1-10

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

 $\label{lem:Lysinuric Protein Intolerance (SLC7A7): Mutations (4): σ^a Genotyping $\mid c.1228C>T$ (p.R410X), c.726G>A (p.W242X), c.1384_1385insATCA (p.R462fs), c.895-2A>T Sequencing $\mid NM_001126105:3-11$ (p.R462fs), c.895-2A=T Sequencing $\mid NM_001126105$

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* 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): σ 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

 $\label{eq:maple-syrup-Urine Disease: Type 2 (DBT): Mutations (15): σ 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$

 $\label{eq:maple-syrup-Urine Disease: Type 3 (DLD): Mutations (8): σ^* Genotyping | c.104_105insA, c.685G>T (p.G229C), c.214A>G (p.K72E), c.1081A>G (p.M361V), c.1123G>A (p.E375K), c.1178T>C (p.1393T), c.1463C>T (p.P488L), c.1483A>G (p.R495G) Sequencing | NM_000108:1-14$

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): of Genotyping | c.985A>G (p.K329E), c.362C>T (p.T121I), c.583G>A (p.G195R), c.799G>A (p.G267R), c.199T>C (p.Y67H), c.262C>T (p.L88F), c.616C>T (p.R206C), c.617G>A (p.C206H) Sequencing | NM_001127328:1-12

Megalencephalic Leukoencephalopathy (MLC1): Mutations (6): of 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): ♂ Genotyping | c.1210+1G>A, c.465+1G>A (IVS2+1G>A), c.862A>C (p.T288P), c.1136C>T (p.P379L), c.1283C>T (p.P428L), c.827C>T (p.T276M), c.542T>G (p.1181S), c.1232C>T (p.T411I), c.769G>C (p.D257H), c.739G>A (p.G247R), c.641C>T (p.A214V), c.302G>A (p.G101D), c.293C>T (p.S98F), c.257G>A (p.R86Q), c.263G>A (p.G88D), c.1114C>T (p.R372W), c.292_293delTCinsCT (p.S98L), c.302G>T (p.G101V) Sequencing | NM_001085425:2-9

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

Methylmalonic Acidemia: MMAB Related (MMAB): Mutations (11): & Genotyping | c.700C>T (p.Q234X), c.656A>G (p.Y219C), c.572G>A (p.R191Q), c.571C>T (p.R191W), c.569G>A (p.R190H), c.568C>T (p.R190C), c.556C>T (p.R186W), c.403G>A (p.A135T), c.291-1G>A, c.287T>C (p.196T), 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^{n} 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): o* 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): σ 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): of Genotyping | c.182G>A (p.R61H), c.788A>G (p.D263G), c.740G>A (p.R247H), c.1331G>A (p.R444K), c.1364T>G (p.V455G), c.776_777insT (p.G259fs), c.1175C>T (p.A392V) Sequencing | NM_000183:2-16

Morquio Syndrome: Type A (GALNS): Mutations (6): of 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): of 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): O' 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): o' 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): σ Genotyping | c.463T>C (p.S155P) Sequencing | NM_182760:1-9

Muscle-Eye-Brain Disease (POMGNT1): Mutations (3): 0° 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): σ Genotyping | c.149G>A (p.R50Q) Sequencing | NM_002437:2-8

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

Nephrotic Syndrome: Type 1 (NPHS1): Mutations (5): O* Genotyping | c.121_122delCT (p.L41 Dfs), c.1481 delC, c.3325C>T (p.R1109X), c.3478C>T (p.R1160X), c.2335-1 G>A



Carrier Map™

Sequencing | NM_004646:1-29

Nephrotic Syndrome: Type 2 (NPHS2): Mutations (27): o' 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.F183fsX186), c.538G>A (p.V180M), c.503G>A (p.R168H), c.502C>A (p.R168S), c.502C>T (p.R168C), c.479A>G (p.D160G), c.467delT (p.L156fsX180), c.467_468insT (p.L156fsX166), c.419delG (p.G140fsX180), c.413G>A (p.R138Q), c.412C>T (p.R138X), c.353C>T (p.P118L), c.274G>T (p.G92C), c.104_105insG (p.G35fsX69), c.85G>A (p.A29T) Sequencing | NM_014625:1-8

Neuronal Ceroid-Lipofuscinosis: CLN5 Related (CLN5): Mutations (7): o* 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

 $\label{eq:Neuronal Ceroid-Lipofuscinosis: CLN6 Related (CLN6): Mutations (8): σ^{*} Genotyping | c.663C>G (p.Y221X), c.460_462delATC (p.1154del), c.368G>A (p.G123D), c.308G>A (p.R103Q), c.214G>T (p.E72X), c.200T>C (p.L67P), c.139C>T (p.L47F), c.17G>C (p.R6T) Sequencing | NM_017882:2-7$

Neuronal Ceroid-Lipofuscinosis: CLN8 Related (CLN8): Mutations (4): of 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): of 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.L19Q), 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): o* Genotyping | c.523-1G>A, c.509-1G>C, c.622C>T (p.R208X), c.851G>T (p.G284V), c.1340G>A (p.R477H), c.1094G>A (p.C365Y), c.1093T>C (p.C365R), c.857A>G (p.N286S), c.616C>T (p.R206C) Sequencing | NM_000391:1-13

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

Niemann-Pick Disease: Type B (SMPD1): Mutations (3): & Genotyping | c.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): of 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): of Genotyping | c.657_661 delACAAA (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.556>C (p.R184P), c.71G>A (p.W24X), c.229T>C (p.W77R), c.269T>C (p.L90P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T, c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147K), c.-23+1G>A, c.550C>T (p.R184W), c.-259C>T Sequencing | NM_004004:1-2

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

Oculocutaneous Albinism: Type 1 (TYR): Mutations (27): & Genotyping | c.272G>A (p.C91Y), c.242C>T (p.P81L), c.265T>C (p.C89R), c.1A>G (p.M1V), c.140G>A (p.G47D), c.325G>A (p.G109R), c.568delG (p.G191Dfs), c.707G>A (p.W236X), c.832C>T (p.R278X), c.1118C>A (p.T373K), c.229C>T (p.R77W), c.823G>T (p.V275F), c.32G>A (p.W11X), c.149C>T (p.S50L), c.1467_1468insT (p.A490C5), c.820-2A>G, c.892C>T (p.R298W), c.1064C>T (p.A355V), c.1090A>C (p.N364H), c.1150C>G (p.P384A), c.1184+1G>A, c.1309G>A (p.D437N), c.1469C>A (p.A490D), c.133_134insC (p.P45fs), c.710delA (p.D237fs), c.978delA (p.Q326fs), c.1138_1158delTCTGCCAACGATCCTATCTTC (p.S380_F386del) Sequencing | NM_000372:1-5

Oculocutaneous Albinism: Type 3 (TYRP1): Mutations (6): O* 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): σ^2 Genotyping | c.469G>A (p.D157N), c.563G>T (p.G188V) Sequencing | NM_016180:1-7

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

Omenn Syndrome: RAG2 Related (RAG2): Mutations (1): σ^{n} 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): of 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): of 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): O Genotyping | c.1001+1G>A, c.1151A>G (p.E384G), c.1246A>C (p.T416P), c.2168A>G (p.H723R), c.707T>C (p.L236P), c.716T>A (p.V239D), c.919-2A>G Sequencing | NM_000441:1-21

Persistent Mullerian Duct Syndrome: Type I (AMH): Mutations (6): & Genotyping | c.1144G>T (p.E382X), c.571C>T (p.R191X), c.1518C>G (p.H506Q), c.1574G>A (p.C525Y), c.17_18deITC, c.283C>T (p.R95X) Sequencing | NM_000479:1-4

Persistent Mullerian Duct Syndrome: Type II (AMHR2): Mutations (14): 0* Genotyping | c.232+1G>A, c.1330_1356delCTGGGCAATACCCCTACCTCGATGAG, 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): O' Genotyping | c.1066-11G>A (IVS10-11G>A), c.1315+1G>A (IVS12+1G>A), c.1241A>G (p.Y414C), c.1222C>T (p.R408W), c.754C>T (p.R252W), c.1223G>A (p.R408Q), c.473G>A (p.R158Q), c.782G>A (p.R261Q), c.814G>T (p.G272X), c.143T>C (p.L48S), c.194T>C (p.165T), 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): o* Genotyping | c.769C>T (p.R257X), c.254A>G (p.Y85C), c.1163_1164insA (p.M388lfsX36), c.967_979delCTGTCCCTCCGC (p.L323SfsX51), c.415C>T (p.R139X) Sequencing | NM_000383:1-14

Pontocerebellar Hypoplasia: EXOSC3 Related (EXOSC3): Mutations (4): of 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

 $\label{eq:posterior} \begin{tabular}{ll} Pontocerebellar Hypoplasia: RARS2 Related (RARS2): Mutations (3): σ^{n} Genotyping | c.35A>G (p.Q12R), c.110+5A>G, c.1024A>G (p.M342V) Sequencing | NM_020320:1-20 |$

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

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

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

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





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

Primary Ciliary Dyskinesia: DNAI1 Related (DNAI1): Mutations (5): c* 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): 0* 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): 0^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): O* Genotyping | c.508G>A (p.G170R), c.454T>A (p.F152I), c.731T>C (p.I244T), c.121G>A (p.G41R), c.198C>G (p.Y66X), c.245G>A (p.G82E), c.466G>A (p.G156R), c.613T>C (p.S205P), c.697C>T (p.R233C), c.698G>A (p.R233H), c.738G>A (p.W246X) Sequencing | NM_000030:1-11

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

Primary Hyperoxaluria: Type 3 (HOGA1): Mutations (2): O' 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' 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): of 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): ♂ Genotyping | c.293A>G (p.D98G) Sequencing | NM_000055:2-4

Pycnodysostosis (CTSK): Mutations (2): o^a Genotyping | c.990A>G (p.X330W), c.926T>C (p.L309P) 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): σ^a Genotyping | c.395A>G (p.Y132C), c.1030C>T (p.P344S) Sequencing | NM_000925:1-10

Renal Tubular Acidosis and Deafness (ATP6V1B1): Mutations (7): σ 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.1386fs) 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): 3° 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): σ 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): of Genotyping | c.124A>G (p.K42E) Sequencing | NM_024887:2-9

Retinitis Pigmentosa: FAM161A Related (FAM161A): Mutations (5): 0* 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): O* Genotyping | c.903+1G>C, c.649G>A (p.G217R), c.875T>A (p.L292X), c.40A>C (p.T14P), c.45_52insGGGACGCC (p.H18RfsX35), c.120C>G (p.Y40X), c.345T>G (p.Y115X), c.653C>T (p.A218V) Sequencing | NM_000288:1-10

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

Sandhoff Disease (HEXB): Mutations (14): 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): d* 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): o' Genotyping | c.2021 G>A (p.R674H), c.889C>T (p.R297X), c.1928G>A (p.R643H), c.1927C>T (p.R643C), c.1562C>T (p.F521L), 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): O* 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): of 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): of 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): O' 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): of 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.R242W), c.575C>T (p.S192F), c.1295A>G (p.Y432C), c.1039G>A (p.G437S), c.1079T>C (p.L360P), c.1424T>C (p.F475S), c.1190C>T (p.S397L), c.1351T>C (p.C451R), c.853_855delTTC (p.285delF), c.1327C>T (p.R443C), c.151C>T (p.P51S), c.296T>C (p.L99P), c.443T>G (p.L148R), c.502T>A (p.F168I), c.523G>C (p.D175H), c.536C>T (p.P179L), c.728C>G (p.P243R), c.852C>A (p.F284L), c.861C>A (p.N287K), c.970T>C (p.Y324H), c.1384T>C (p.Y462H), c.1406G>C (p.R469P), c.111G>A (p.W37X) Sequencing | NM_001360:3-9

Spinal Muscular Atrophy: SMN1 Linked (SMN1): Mutations (19): of 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.1228X), c.734C>T (p.P2451), c.768_778dupTGCTGATGCTT, c.815A>G (p.Y272C), c.821C>T (p.T274I), c.823G>A (p.G275S), c.834+2T>G, c.835-18_835-12delCCTTTAT, c.835G>T, c.836G>T dPCR | DEL EXON 7

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

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

Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2): Mutations (7): o' 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): 6 Genotyping | c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.532C>T (p.R178C), c.533G>A (p.R178H), c.805G>A (p.G269S), c.1510C>T (p.R504C), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.1003A>T (p.I335F), c.910_912delTTC (p.305delF), c.749G>A (p.G250D), c.632T>C (p.F211S), c.629C>T (p.S210F), c.613delC, c.611A>G (p.H204R), c.598G>A (p.V200M), c.590A>C (p.K197T), c.571-1G>T, c.540C>G (p.Y180X), c.538T>C (p.Y180H), c.533G>T (p.R178L),





c.508C>T (p.R170W), c.409C>T (p.R137X), c.380T>G (p.L127R), c.346+1G>C, c.116T>G (p.L39R), c.78G>A (p.W26X), c.1A>G (p.M1V), c.1495C>T (p.R499C), c.459+5G>A (IVS4+5G>A), c.1422-2A>G, c.535C>T (p.H179Y), c.1141 delG (p.V381fs), c.796T>G (p.W266G), c.155C>A (p.S52X), c.426delT (p.F142fs), c.413-2A>G, c.570+3A>G, c.536A>G (p.H179R), c.1146+1G>A, c.736G>A (p.A246T), c.1302C>G (p.F434L), c.778C>T (p.P260S), c.1008G>T (p.Q336H), c.1385A>T (p.E462V), c.964G>A (p.D322N), c.340G>A (p.E144K), 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.G513X), c.1511G>T (p.R504L), c.1307_1308delTA (p.1436fs), c.571-8A>G, c.624_627delTCCT (p.D208fs), c.1211_1212delTG (p.L404fs), c.621T>G (p.D207E), c.1511G>A (p.R504H), c.1177C>T (p.R393X), c.21>C (p.M1T), c.1292G>A (p.W431X), c.947_948insA (p.Y316fs), c.607T>G (p.W203G), c.1061_1063delTCT (p.F354_Y355delinsX), c.615delG (p.L205fs), c.805+2T>C, c.1123delG (p.E375fs), c.1121A>G (p.Q374R), c.1043_1046delTCAA (p.F348fs), c.1510delC (p.R504fs), c.1451T>C (p.L484P), c.964G>T (p.D322Y), c.1351C>G (p.L451V), c.571-2A>G (IVS5-2A>G) Sequencing | NM_000520:1-14

Trichohepatoenteric Syndrome: Type 1 (TTC37): Mutations (9): o' Genotyping | c.3847G>A (p.D1283N), c.751G>A (p.G251R), c.2251C>T (p.Q751X), c.439C>T (p.Q147X), c.2808G>A (p.W936X), c.2515+1G>C, c.4620+1G>C, c.1632+1delG, c.2578-7delTTTTT Sequencing | NM_014639:4-43

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

 $\label{thm:continuity} \textbf{Tyrosinemia: Type I (FAH): } \mbox{Mutations (10): σ' Genotyping | c.1062+5G>A, c.554-1G>T, c.607-6T>G, c.707-1G>C, c.782C>T (p.P261L), c.1069G>T (p.E357X), c.786G>A (p.W262X), c.698A>T (p.D233V), c.1009G>A (p.G337S), c.192G>T (p.Q64H) Sequencing | NM_000137:1-14$

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): & 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): of Genotyping | c.IVS5+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 (15): & Genotyping | c.172C>T (p.Q58X), c.3367C>T (p.Q1123X), c.3617C>G (p.P1206R), c.3713_3714delCT (p.S1238fs), c.3880C>T (p.Q1294X), c.4069C>T (p.Q1357X), c.4488G>C (p.Q1496H), c.4504C>T (p.R1502X), c.5237G>A (p.R1746Q), c.5985C>A (p.Y1995X), c.6307G>T (p.E2103X), c.7549A>G (p.S2517G), c.8230G>A (p.G2744S), c.8497C>G (p.R2833G), c.9524G>A (p.R3175H) Sequencing | NM_022124:2-68

Usher Syndrome: Type 1F (PCDH15): Mutations (7): of 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.1425fs) 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.E767SfsX21), c.2209C>T (p.R37X), c.1256G>T (p.C419F), c.1000C>T (p.R334W), c.923_924insGCCA (p.H308fs), c.12708T>A (p.C4236X), c.13576C>T (p.R4526X), c.1840+1G>A, c.11328T>G (p.Y3776X), c.5329C>T (p.R1777W), c.9165_9168delCTAT (p.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): of 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.G441D), c.1372T>C (p.F458L), c.1405C>T (p.R469W), c.1837C>T (p.R613W), c.553G>A (p.G185S), c.739A>C (p.K247Q), c.37C>T (p.Q13X), c.265C>T (p.P89S), c.272C>A (p.P91Q), c.364A>G (p.N122D), c.388_391delGAGA (p.E130fs), c.520G>A (p.V174M), c.856A>G (p.R286G), c.1606_1609delGCAG (p.A536fs), c.1531C>T (p.R511W), c.1512G>T (p.E504D), c.664G>A (p.G222R), c.685C>T (p.R229X), c.577G>C (p.G193R), c.881G>A (p.G294E), c.753-2A>C (IVS8-2A>C), c.1349G>A (p.R450H), c.1358G>A (p.R453Q), c.790A>G (p.K264E), c.1246G>A (p.A416T) Sequencing | NM_000018:1-20

Walker-Warburg Syndrome (FKTN): Mutations (5): O' 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): of 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): O* 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): Ø 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): of 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): σ Genotyping | c.172+2T>G, c.323G>T (p.C108F), c.374delC (p.T125fs), c.682C>T (p.R228X), c.619C>T (p.R207X), c.348T>A (p.Y116X), c.390-1G>C Sequencing | NM_000380:1-6

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

Zellweger Spectrum Disorders: PEX1 Related (PEX1): Mutations (3): of 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* Genotyping | c.764_765insA, c.874_875delCT Sequencing | NM_153818:2-6

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

Zellweger Spectrum Disorders: PEX6 Related (PEX6): Mutations (8): 0* Genotyping | c.1130+1G>A (IVS3+1G>A), c.1688+1G>A (IVS7+1G>A), c.1962-1G>A (p.L655fsX3), c.1301delC (p.S434Ffs), c.1601T>C (p.L534P), c.511insT (p.G171Wfs), c.802_815delGACGGACTGGCGCT (p.D268Cfs), c.1715C>T (p.T572I) Sequencing | NM_000287:1-17





Residual Risk Information

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

may become available in the	future.		
Disease	Carrier Rate	Detection Rate	Residual Risk
11 -Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	♂ Moroccan Jewish: 1/39	91.67%	1/468
17-Alpha-Hydroxylase Deficiency	♂ Brazilian: Unknown	54.55%	Unknown
	o [*] Japanese: Unknown	45.45%	Unknown
17-Beta-Hydroxysteroid Dehydrogenase Deficiency	♂ 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	♂ Argentinian: 1/4	<10%	1/4
	o' European: 1/16	<10%	1/16
3-Beta-Hydroxysteroid Dehydrogenase Deficiency	♂ General: Unknown	16.13%	Unknown
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCA Related	♂ European: 1/146	26.32%	1/198
	o' General: 1/112	37.50%	1/179
3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related	♂ 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	o⁴ Iraqi Jewish: 1/10	>99%	<1/1,000
3-Phosphoglycerate Dehydrogenase Deficiency	♂ 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	♂ Chinese: 1/183	<i>7</i> 8.95%	1/869
	o" East Asian: 1/180	64.20%	1/503
ARSACS	o' French Canadian: 1/22	95.45%	1/484
Abetalipoproteinemia	♂ Ashkenazi Jewish: 1/131	>99%	<1/13,10 0
Acrodermatitis Enteropathica	o ^r Arab: Unknown	40.00%	Unknown
	og Egyptian: Unknown	33.33%	Unknown
	of French: Unknown	27.78%	Unknown
	♂ Tunisian: Unknown	77.78%	Unknown
Acute Infantile Liver Failure: TRMU Related	♂ Yemenite Jewish: 1/40	71.43%	1/140
Acyl-CoA Oxidase I Deficiency	♂ General: Unknown	35.00%	Unknown
	♂ Japanese: Unknown	42.86%	Unknown
Adenosine Deaminase Deficiency	♂ General: 1/388	36.96%	1/615

Disease	Carrier Rate	Detection Rate	Residual Risk
Alkaptonuria	of Dominican: Unknown	>99%	Unknown
	♂ Finnish: 1/251	60.00%	1/628
	♂ Slovak: 1/69	59.38%	1/170
Alpha Thalassemia	♂ General: 1/48	50.67%	1/97
Alpha-1-Antitrypsin Deficiency	♂ European: 1/35	95.00%	1/700
	o" General: Unknown	95.00%	Unknown
Alpha-Mannosidosis	o' European: 1/354	30.23%	1/507
	♂ 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	26.67%	1/558
Amegakaryocytic Thrombocytopenia	o" Ashkenazi Jewish: 1/76	>99%	<1/7,600
	o' General: Unknown	64.81%	Unknown
Andermann Syndrome	of French Canadian: 1/24	99.38%	1/3,888
Antley-Bixler Syndrome	of General: Unknown	45.65%	Unknown
	og Japanese: Unknown	60.47%	Unknown
Argininemia	of Chinese: Unknown	40.00%	Unknown
	♂ French Canadian: Unknown	75.00%	Unknown
	o' 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	♂ Iranian Jewish: 1/80	>99%	<1/8,000
Aspartylglycosaminuria	of Finnish: 1/69	96.12%	1/1,780
Ataxia with Vitamin E Deficiency	of 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
	♂ North African Jewish: 1/81	96.97%	1/2,673
	o' Norwegian: 1/197	50.00%	1/394
	og Sardinians: Unknown	85. <i>7</i> 1%	Unknown
	o US Amish: Unknown	>99%	Unknown
Autosomal Recessive Polycystic Kidney Disease	o⁴ Finnish: 1/45	84.21%	1/285
	of French: 1/71	62.50%	1/189
	♂ General: 1/71	<i>37</i> .11%	1/113
Bardet-Biedl Syndrome: BBS1 Related	♂ General: 1/376	70.27%	1/1,265
	♂ Northern European: 1/376	85.90%	1/2,666
	♂ Puerto Rican: Unknown	90.00%	Unknown
Bardet-Biedl Syndrome: BBS10 Related	of General: 1/404	47.79%	1/774
Bardet-Biedl Syndrome: BBS11 Related	of Bedouin: 1/59	>99%	<1/5,900
Bardet-Biedl Syndrome: BBS 12 Related	od 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	♂ Ashkenazi Jewish: Unknown	>99%	Unknown		♂ Moroccan Jewish: 1/234	>99%	<1/23,40
	♂ General: 1/638	38.46%	1/1,037	Citrin Deficiency	♂ Japanese: 1/70	>99%	<1/7,000
	♂ Middle Eastern: Unknown	>99%	Unknown	Citrullinemia: Type I	♂ European: 1/120	18.18%	1/147
Bare Lymphocyte Syndrome: Type II	♂ General: Unknown	66.67%	Unknown		o'' General: 1/120	52.27%	1/251
Bartter Syndrome: Type 4A	♂ General: 1/457	81.82%	1/2,514		♂ Japanese: Unknown	64.71%	Unknown
Beta Thalassemia	♂ African American: 1/75	84.21%	1/475		♂ Mediterranean: 1/120	50.00%	1/240
	♂ Indian: 1/24	74.12%	1/93	Classical Galactosemia	♂ African American: 1/78	73.13%	1/290
	♂ Sardinians: 1/23	97.14%	1/804		♂ Ashkenazi Jewish: 1/127	>99%	<1/12,70
	♂ Spaniard: 1/51	93.10%	1/739				0
Beta-Hexosaminidase Pseudodeficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown		o ^a Dutch: 1/91 o ^a European: 1/112	75.47% 88.33%	1/371 1/960
,	♂ General: Unknown	>99%	Unknown		o' General: 1/125	80.00%	1/625
Beta-Ketothiolase Deficiency	♂ Japanese: Unknown	58.33%	Unknown		o' Irish: 1/76	91.30%	1/874
Sold No. omnoted Sold one,	♂ 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	of Christian Arab: Unknown	50.00%	Unknown
Bloom Syndrome	o' Ashkenazi Jewish: 1/134	96.67%	1/4,020	Cockayne Syndrome: Type B	of General: 1/378	19.30%	1/468
bloom syndrome	of European: Unknown	66.22%	Unknown	Cohen Syndrome	of European: Unknown	19.05%	Unknown
	of Japanese: Unknown	50.00%	Unknown	Conen syndrome	of Finnish: 1/140	67.24%	1/427
Canavan Disease	of Ashkenazi Jewish: 1/55	98.86%	1/4,840		of US Amish: 1/12	>99%	<1/1,200
Canavan Disease	•			Combined Bitainer House	•		
C:4: P-l:4 41A	of European: Unknown	53.23%	Unknown	Combined Pituitary Hormone Deficiency: PROP1 Related	o [™] European: 1/45	93.29%	1/671
Carnitine Palmitoyltransferase IA Deficiency	♂ General: Unknown	38.89%	Unknown		♂ General: 1/45	82.35%	1/255
	♂ Hutterite: 1/16	>99%	<1/1,600	Congenital Disorder of Glycosylation: Type 1A: PMM2 Related	o' Danish: 1/71	90.00%	1/710
	♂ Japanese: 1/101	66.67%	1/303	Type 17 CTMINE Related	o' Dutch: 1/68	39.29%	1/112
Carnitine Palmitoyltransferase II Deficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown		of European: 1/71	55.33%	1/159
	♂ General: Unknown	71.43%	Unknown	Congenital Disorder of Glycosylation:	o' French: Unknown	54.17%	Unknown
Carnitine-Acylcarnitine Translocase Deficiency	♂ Asian: Unknown	95.45%	Unknown	Type 1B: MPI Related Congenital Disorder of Glycosylation:	o⁴ French: Unknown	59.09%	Unknown
,	♂ General: Unknown	18.75%	Unknown	Type 1C: ALG6 Related			
Carpenter Syndrome	♂ Brazilian: Unknown	40.00%	Unknown		o' General: Unknown	86.21%	Unknown
, ,	♂ Northern European:	85.00%	Unknown	Congenital Ichthyosis: ABCA 12 Related	♂ North African: Unknown	>99%	Unknown
	Unknown				o'' South Asian: Unknown	66.67%	Unknown
Cartilage-Hair Hypoplasia	♂ Finnish: 1/76	93.33%	1/1,140	Congenital Insensitivity to Pain with	o" Japanese: Unknown	56.52%	Unknown
	♂ US Amish: 1/19	>99%	<1/1,900	Anhidrosis	ا با بداد	. 000/	
Cerebrotendinous Xanthomatosis	o⁴ Dutch: Unknown	78.57%	Unknown		♂ Moroccan Jewish: Unknown	>99%	Unknown
	♂ Italian: Unknown	45.95%	Unknown	Congenital Lipoid Adrenal Hyperplasia	♂ Japanese: 1/201	51.11%	1/411
	♂ Japanese: Unknown	92.86%	Unknown		♂ Korean: 1/251	63.64%	1/690
	♂ Moroccan Jewish: 1/6	87.50%	1/48	Congenital Myasthenic Syndrome:	♂ European Gypsy: 1/26	>99%	<1/2,600
Chediak-Higashi Syndrome	♂ General: Unknown	19.64%	Unknown	CHRNE Related			
Cholesteryl Ester Storage Disease	♂ General: 1/101	68.97%	1/325		o' North African: Unknown	60.87%	Unknown
Choreoacanthocytosis	♂ Ashkenazi Jewish: Unknown	66.67%	Unknown	Congenital Myasthenic Syndrome: DOK7 Related	♂ European: 1/472	19.05%	1/583
Chronic Granulomatous Disease:	♂ Iranian: Unknown	71.43%	Unknown		o' General: 1/472	18.75%	1/581
CYBA Related		>99%		Congenital Myasthenic Syndrome: RAPSN Related	of General: 1/437	88.57%	1/3,824
	o [™] Japanese: 1/274 o [™] Korean: 1/105	>99% >99%	<1/27,40 0 <1/10,50		♂ Non-Ashkenazi Jewish: Unknown	>99%	Unknown





X Recomb	ine				Carrie	rMo	ap [™]
Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Congenital Neutropenia: Recessive	♂ English: Unknown	11.76%	Unknown		♂ Saudi Arabian: 1/38	>99%	<1/3,800
	♂ Japanese: Unknown	22.22%	Unknown	Familial Dysautonomia	♂ Ashkenazi Jewish: 1/31	>99%	<1/3,100
	o⁴ Turkish: Unknown	89.47%	Unknown	Familial Hyperinsulinism: Type 1:	o⁴ Ashkenazi Jewish: 1/52	98.75%	1/4,160
Corneal Dystrophy and Perceptive Deafness	og General: Unknown	71.43%	Unknown	ABCC8 Related	♂ Finnish: 1/101	45.16%	1/184
Corticosterone Methyloxidase Deficiency	♂ Iranian Jewish: 1/32	>99%	<1/3,200	Familial Hyperinsulinism: Type 2: KCNJ11 Related	♂ Arab: Unknown	40.00%	Unknown
Crigler-Najjar Syndrome	♂ Sardinians: Unknown	80.00%	Unknown	Familial Mediterranean Fever	♂ Arab: 1/4	51.27%	1/8
	o⁴ Tunisian: Unknown	>99%	Unknown		o⁴ Armenian: 1/5	94.51%	1/91
Cystic Fibrosis	♂ African American: 1/62	69.99%	1/207		o⁴ Ashkenazi Jewish: 1/81	40.95%	1/137
Cysiic (1610313	of Ashkenazi Jewish: 1/23	96.81%	1/721		of Iragi Jewish: 1/4	76.92%	1/17
	•						
	of Asian: 1/94	65.42%	1/272		♂ Israeli Jewish: 1/5	62.67%	1/13
	of European: 1/25	94.96%	1/496		♂ Lebanese: 1/6	91.67%	1/72
	♂ Hispanic American: 1/48	77.32%	1/212		♂ North African Jewish: 1/5	95.69%	1/116
	od Native American: 1/53	84.34%	1/338		♂ Syrian: 1/6	85.14%	1/40
Cystinosis	o [®] Dutch: 1/194	73.08%	1/721		♂ Turkish: 1/5	74.43%	1/20
	♂ French Canadian: 1/40	75.00%	1/160	Fanconi Anemia: Type A	♂ Moroccan Jewish: 1/100	>99%	<1/10,00
	♂ General: 1/194	54.51%	1/426		o⁴ Spanish Gypsy: 1/67	>99%	<1/6,700
Cystinuria: Non-Type I	♂ European: 1/42	61.11%	1/108	Fanconi Anemia: Type C	o Spanish Sypsy: 1/ 0/ o Ashkenazi Jewish: 1/101	>99%	<1/10,10
	♂ General: 1/42	37.50%	1/67	rancon Anemia. Type C	O Asirkeliazi sewisii. 17 101	- 7 7 70	0
	♂ Libyan Jewish: 1/26	93.48%	1/399		of General: Unknown	30.00%	Unknown
Cystinuria: Type I	o [™] United States: 1/42 o [™] European: 1/42	56.25% 46.67%	1/96 1/79	Fanconi Anemia: Type G	o ^a Black South African: 1/101	81.82%	1/556
Cysillolla. Type I	of Swedish: 1/159	55.88%	1/360		of French Canadian:	87.50%	Unknown
D-Bifunctional Protein Deficiency	of General: 1/159	38.64%	1/259		Unknown		
Diabetes: Recessive Permanent	o' General: Unknown	25.00%	Unknown		♂ Japanese: Unknown	75.00%	Unknown
Neonatal	O General, Officiowii	23.00%	Onknown		♂ Korean: Unknown	66.67%	Unknown
Du Pan Syndrome	♂ Pakistani: Unknown	>99%	Unknown	Fanconi Anemia: Type J	♂ General: Unknown	86.36%	Unknown
Dyskeratosis Congenita: RTEL1 Related	♂ Ashkenazi Jewish: 1/203	>99%	<1/20,30	Fumarase Deficiency	♂ General: Unknown	30.00%	Unknown
	♂ General: 1/501	50.00%	0 1/1,002	GM1-Gangliosidoses	♂ Eurodescent Brazilian: 1/66	62.15%	1/174
Dystrophic Epidermolysis Bullosa:	♂ Italian: Unknown	45.00%	Unknown		o' European: 1/194	50.00%	1/388
Recessive					o'' General: 1/194	20.00%	1/243
	o⁴ Mexican American: 1/345	56.25%	1/789		♂ Hispanic American: 1/194	58.33%	1/466
Ehlers-Danlos Syndrome: Type VIIC	♂ Ashkenazi Jewish:	>99%	Unknown		of Japanese: Unknown	62.82%	Unknown
	Unknown	00.140/	1 /101	GRACILE Syndrome	♂ Finnish: 1/109	97.22%	1/3,924
Ellis-van Creveld Syndrome: EVC Related	♂ General: 1/123	32.14%	1/181	Galactokinase Deficiency	♂ Japanese: 1/501	50.00%	1/1,002
Ellis-van Creveld Syndrome: EVC2 Related	o" General: Unknown	<10%	Unknown		o'' Roma: 1/51	>99%	<1/5,100
Enhanced S-Cone	o" Ashkenazi Jewish:	90.48%	Unknown	Gaucher Disease	♂ Ashkenazi Jewish: 1/15 ♂ General: 1/112	87.16% 31.60%	1/117 1/164
	Unknown	E0 500'			o Spaniard: Unknown	44.29%	Unknown
ed to to a set o	of General: Unknown	52.50%	Unknown		o⁴ Turkish: 1/236	59.38%	1/581
Ethylmalonic Aciduria	o" Arab/Mediterranean: Unknown	29.17%	Unknown	Gitelman Syndrome	♂ European: 1/100	35.00%	1/154
	♂ General: Unknown	38.24%	Unknown		o⁴ European Gypsy:	>99%	Unknown
Familial Chloride Diarrhea	♂ Finnish: 1/51	>99%	<1/5,100		Unknown		
	o³ Kuwaiti: 1/38	90.00%	1/380		o⁴ General: 1/101	30.00%	1/144
	o'' Polish: 1/224	45.24%	1/409		o⁴ Taiwanese: Unknown	64.29%	Unknown





Disease	Carrier Rate	Detection Rate	Residual Risk	Disease
Globoid Cell Leukodystrophy	o⁴ Dutch: 1/137	60.98%	1/351	Hemochromatosis: Ty
	of European: 1/150	26.47%	1/204	Related
	♂ Japanese: 1/150	36.00%	1/234	
Glutaric Acidemia: Type I	o' European: 1/164	57.78%	1/388	Hemochromatosis: Ty Related
	of General: 1/164	25.51%	1/220	Hemoglobinopathy:
	♂ US Amish: 1/12	>99%	<1/1,200	Hemoglobinopathy:
Glutaric Acidemia: Type IIA	of General: Unknown	71.43%	Unknown	, ,
Glutaric Acidemia: Type IIB	og General: Unknown	33.33%	Unknown	
Glutaric Acidemia: Type IIC	♂ Taiwanese: Unknown	>99%	Unknown	Hemoglobinopathy:
	♂ Turkish: Unknown	80.00%	Unknown	,
Glycine Encephalopathy: AMT Related	of General: Unknown	40.91%	Unknown	
Glycine Encephalopathy: GLDC telated	♂ Finnish: 1/118	78.00%	1/536	
	o" General: 1/280	12.50%	1/320	Hemoglobinopathy:
Glycogen Storage Disease: Type IA	♂ Ashkenazi Jewish: 1/71	>99%	<1/7,100	
	o' Chinese: 1/159	80.00%	1/795	Hereditary Fructose I
	o ^a European: 1/177	76.88%	1/765	
	♂ Hispanic American: 1/177	27.78%	1/245	Hereditary Spastic Po
	o ^a Japanese: 1/177	89.22%	1/1,641	Related
Glycogen Storage Disease: Type IB	of Australian: 1/354	50.00%	1/708	Herlitz Junctional Ep Bullosa: LAMA3 Rela
	o" European: 1/354	45.74%	1/652	Herlitz Junctional Ep
	♂ Japanese: 1/354	39.13%	1/582	Bullosa: LAMB3 Rela
Slycogen Storage Disease: Type II	♂ African American: 1/60	45.83%	1/111	
	o' Chinese: 1/112	72.00%	1/400	Herlitz Junctional Epi
	o' European: 1/97	51.76%	1/201	Bullosa: LAMC2 Rela
	♂ North African: Unknown	60.00%	Unknown	Hermansky-Pudlak S
Glycogen Storage Disease: Type III	o' Faroese: 1/30	>99%	<1/3,000	Hermansky-Pudlak S
	o" General: 1/159	39.81%	1/264	
	or North African Jewish: 1/35	>99%	<1/3,500	Hermansky-Pudlak S
Glycogen Storage Disease: Type IV	♂ Ashkenazi Jewish: 1/35	>99%	<1/3,500	Holocarboxylase Sy Deficiency
	♂ General: 1/461	18.60%	1/566	,
Slycogen Storage Disease: Type V	♂ Caucasus Jewish: Unknown	>99%	Unknown	Homocystinuria Cau Deficiency
	o" European: 1/159	60.71%	1/405	
	♂ General: Unknown	74.10%	Unknown	
	o ^a Spaniard: 1/159	67.11%	1/483	
	♂ Yemenite Jewish: Unknown	75.00%	Unknown	
Glycogen Storage Disease: Type VII	♂ Ashkenazi Jewish: 1/250	>99%	<1/25,00 0	
Guanidinoacetate Methyltransferase Deficiency	♂ General: Unknown	29.41%	Unknown	Hurler Syndrome
HMG-CoA Lyase Deficiency	♂ General: 1/159	40.00%	1/265	
	♂ Japanese: Unknown	30.00%	Unknown	
	o' Portuguese: Unknown	86.36%	Unknown	
	•			

Disease	Carrier Rate	Detection Rate	Residual Risk
Hemochromatosis: Type 2A: HFE2 Related	♂ European: Unknown	69.23%	Unknown
	♂ Mediterranean: Unknown	72.73%	Unknown
Hemochromatosis: Type 3: TFR2 Related	♂ Italian: Unknown	73.21%	Unknown
Hemoglobinopathy: Hb C	♂ African American: 1/51	>99%	<1/5,100
Hemoglobinopathy: Hb D	♂ Canadian: 1/64	>99%	<1/6,400
	o'' Indian: 1/16	>99%	<1/1,600
	♂ Iranian: 1/11	>99%	<1/1,100
Hemoglobinopathy: Hb E	♂ Cambodia: 1/4	>99%	<1/400
	♂ Chinese: 1/13	>99%	<1/1,300
	o'' Indian: 1/10	>99%	<1/1,000
	o'' Thai: 1/9	>99%	<1/900
Hemoglobinopathy: Hb O	♂ African American: 1/87	>99%	<1/8,700
	♂ Middle Eastern: Unknown	>99%	Unknown
Hereditary Fructose Intolerance	♂ European: 1/81	72.73%	1/297
	o⁴ Italian: 1/81	90.91%	1/891
	o ^a Slavic: 1/81	>99%	<1/8,100
Hereditary Spastic Paraplegia: TECPR2 Related	♂ Bukharan Jewish: 1/75	>99%	<1/7,500
Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related	o [™] Pakistani: Unknown	>99%	Unknown
Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	♂ European: Unknown	70.00%	Unknown
	♂ General: 1/781	52.27%	1/1,636
Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related	♂ Italian: Unknown	28.57%	Unknown
Hermansky-Pudlak Syndrome: Type 1	♂ Puerto Rican: 1/22	94.95%	1/436
Hermansky-Pudlak Syndrome: Type 3	♂ Ashkenazi Jewish: 1/235	>99%	<1/23,50 0
	♂ European: 1/434	12.50%	1/496
Hermansky-Pudlak Syndrome: Type 4	♂ European: Unknown	54.17%	Unknown
Holocarboxylase Synthetase Deficiency	♂ European: 1/148	83.33%	1/888
	♂ Japanese: 1/159	76.92%	1/689
Homocystinuria Caused by CBS Deficiency	o' European: 1/224	64.29%	1/627
	o ^a Irish: 1/128	70.59%	1/435
	o" Italian: 1/224	35.71%	1/348
	♂ Norwegian: 1/41	84.38%	1/262
	♂ Qatari: 1/22	>99%	<1/2,200
	♂ Saudi Arabian: Unknown	92.31%	Unknown
Hurler Syndrome	♂ Czech: 1/190	52.50%	1/400
	♂ European: 1/194	81.71%	1/1,061
	o⁴ General: 1/194	62.50%	1/517
	♂ Italian: 1/194	61.11%	1/499
	o⁴ Japanese: 1/194	23.68%	1/254
	o⁴ Moroccan Jewish: 1/194	92.31%	1/2,522
	o⁴ Scandinavian: 1/194	79.41%	1/942





Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
	♂ Spaniard: 1/194	52.50%	1/408	Limb-Girdle Muscular Dystrophy: Type	♂ Brazilian: Unknown	57.14%	Unknown
Hypophosphatasia	♂ Canadian Amish: 1/26	>99%	<1/2,600	2E	1.500	05.000/	1 /710
	♂ European: 1/159	19.23%	1/197		of European: 1/539	25.00%	1/719
	♂ Japanese: Unknown	54.55%	Unknown		of General: Unknown	12.50%	Unknown
Inclusion Body Myopathy: Type 2	♂ General: Unknown	85.83%	Unknown		of US Amish: Unknown	>99%	Unknown
	♂ Iranian Jewish: 1/16	>99%	<1/1,600	Limb-Girdle Muscular Dystrophy: Type 2F	od Brazilian: Unknown	>99%	Unknown
	♂ Japanese: Unknown	71.88%	Unknown		♂ General: Unknown	83.33%	Unknown
	♂ Korean: Unknown	72.50%	Unknown	Limb-Girdle Muscular Dystrophy: Type	♂ Brazilian: Unknown	34.62%	Unknown
Infantile Cerebral and Cerebellar Atrophy	o³ Caucasus Jewish: 1/20	>99%	<1/2,000	21	o⁴ Danish: 1/100	85.53%	1/691
Isolated Microphthalmia: VSX2 Related	♂ Middle Eastern: Unknown	71.43%	Unknown		o⊓ General: Unknown	43.18%	Unknown
Isovaleric Acidemia	♂ General: 1/251	47.37%	1/477		♂ 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	♂ Norwegian: 1/151	81.40%	1/812	,	♂ General: Unknown	20.00%	Unknown
Laryngoonychocutaneous Syndrome	♂ Pakistani: Unknown	>99%	Unknown	Long-Chain 3-Hydroxyacyl-CoA	♂ European: 1/126	88.98%	1/1,144
Leber Congenital Amaurosis: CEP290 Related	of European: 1/251	47.32%	1/476	Dehydrogenase Deficiency	, ,		
Leber Congenital Amaurosis: GUCY2D	o⁴ Finnish: Unknown	>99%	Unknown		of General: 1/126	56.25%	1/288
Related				Lysinuric Protein Intolerance	♂ Finnish: 1/123	>99%	<1/12,30 0
Leber Congenital Amaurosis: LCA5 Related	♂ Pakistani: Unknown	83.33%	Unknown		♂ Italian: 1/120	45.45%	1/220
Leber Congenital Amaurosis: RDH 12	♂ General: 1/560	38.37%	1/909		♂ Japanese: 1/115	37.93%	1/185
Related					♂ North African: Unknown	>99%	Unknown
Leigh Syndrome: French-Canadian	♂ French Canadian: 1/23	95.45%	1/506	MTHFR Deficiency: Severe	♂ Bukharan Jewish: 1/39	>99%	<1/3,900
Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related	♂ Cree: Unknown	>99%	Unknown	Malonyl-CoA Decarboxylase Deficiency	on General: Unknown	33.33%	Unknown
	♂ European: Unknown	65.22%	Unknown	Maple Syrup Urine Disease: Type 1A	♂ US Amish: 1/10	97.73%	1/440
Leydig Cell Hypoplasia (Luteinizing	♂ Brazilian: Unknown	>99%	Unknown	Maple Syrup Urine Disease: Type 1B	♂ Ashkenazi Jewish: 1/97	>99%	<1/9,700
Hormone Resistance)				Maple Syrup Urine Disease: Type 2	♂ General: 1/481	42.31%	1/834
Limb-Girdle Muscular Dystrophy: Type 2A	♂ Basque: 1/61	61.46%	1/158		♂ Norwegian: 1/481	50.00%	1/962
	♂ Croatian: 1/133	76.00%	1/554		♂ Turkish: 1/112	58.33%	1/269
	o' European: 1/103	17.23%	1/124	Maple Syrup Urine Disease: Type 3	♂ Ashkenazi Jewish: 1/94	>99%	<1/9,400
	o' General: 1/103	26.47%	1/140		♂ General: Unknown	68.75%	Unknown
	♂ Italian: 1/162	35.71%	1/252	Maroteaux-Lamy Syndrome	♂ Argentinian: 1/274	75.00%	1/1,096
	o [*] Russian: 1/103	53.33%	1/221		♂ General: 1/388	61.54%	1/1,009
	o⊓ US Amish: Unknown	>99%	Unknown		♂ Spaniard: 1/274	29.17%	1/387
Limb-Girdle Muscular Dystrophy: Type	♂ Caucasus Jewish: 1/25	>99%	<1/2,500	Meckel Syndrome: Type 1	♂ European: 1/212	72.22%	1/763
2B	,		, ,		♂ Finnish: 1/48	>99%	<1/4,800
	♂ Libyan Jewish: 1/19	>99%	<1/1,900	Medium-Chain Acyl-CoA Dehydrogenase Deficiency	o⁴ European: 1/50	90.91%	1/550
Limb-Girdle Muscular Dystrophy: Type 2C	♂ European Gypsy: 1/50	>99%	<1/5,000	Denyarogenase Deticiency	o" Saudi Arabian: 1/68	95.00%	1/1,360
	♂ General: Unknown	60.00%	Unknown		♂ United Kingdom: 1/51	90.00%	1/510
	♂ Tunisian: Unknown	>99%	Unknown	Megalencephalic	о" Japanese: Unknown	50.00%	Unknown
Limb-Girdle Muscular Dystrophy: Type 2D	o⁵ Brazilian: Unknown	64.29%	Unknown	Leukoencephalopathy	o ^a Libyan Jewish: 1/40	>99%	<1/4,000
	o" European: 1/288	22.22%	1/370		,		
	o' Finnish: 1/150	95.45%	1/3,300	AA-tkt-1-1-1-1-1-1	of Turkish: Unknown	20.00%	Unknown
	of General: Unknown	26.09%	Unknown	Metachromatic Leukodystrophy	of European: 1/150	43.88%	1/267
	Concidi. Onknown	20.07/0	CHANOWII		♂ Habbanite Jewish: 1/5	50.00%	1/10





Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Methylmalonic Acidemia: MMAA Related	♂ General: 1/274	63.51%	1/751	Neuronal Ceroid-Lipofuscinosis: CLN5 Related	o⁴ Finnish: 1/101	>99%	<1/10,10 0
Methylmalonic Acidemia: MMAB Related	o'' General: 1/396	71.25%	1/1,377	Neuronal Ceroid-Lipofuscinosis: CLN6 Related	o⁴ European: 1/159	36.36%	1/250
Methylmalonic Acidemia: MUT Related	d' General: 1/177	43.62%	1/314		♂ General: 1/159	59.52%	1/393
Methylmalonic Aciduria and	♂ Chinese: Unknown	61.39%	Unknown		♂ Portuguese: 1/128	81.00%	1/674
Homocystinuria: Type cblC	♂ General: 1/159	65.74%	1/464	Neuronal Ceroid-Lipofuscinosis: CLN8 Related	of Finnish: 1/135	>99%	<1/13,50 0
	o" Italian: Unknown	75.00%	Unknown		o⁴ Italian: 1/212	33.33%	1/318
	♂ Portuguese: Unknown	91.18%	Unknown		♂ Turkish: Unknown	77.78%	Unknown
Mitochondrial Complex I Deficiency: NDUFS6 Related	♂ Caucasus Jewish: 1/24	>99%	<1/2,400	Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	♂ General: 1/159	56.25%	1/363
Mitochondrial DNA Depletion Syndrome: MNGIE Type	♂ Ashkenazi Jewish: Unknown	>99%	Unknown	Neuronal Ceroid-Lipofuscinosis: PPT 1 Related	o⁴ Finnish: 1/58	97.62%	1/2,436
	♂ General: Unknown	47.37%	Unknown		♂ General: 1/159	72.50%	1/578
Maria de la	♂ Iranian Jewish: Unknown	>99%	Unknown Unknown	Neuronal Ceroid-Lipofuscinosis: TPP1 Related	♂ Canadian: 1/159	67.50%	1/489
Mitochondrial Myopathy and Sideroblastic Anemia	♂ Iranian Jewish: Unknown	>99%	Unknown		o³ European: 1/159	<i>7</i> 5.00%	1/636
Mitochondrial Trifunctional Protein	oʻ Japanese: Unknown	60.00%	Unknown		♂ General: 1/159	50.00%	1/318
Deficiency: HADHB Related					♂ Newfoundlander: 1/43	85.29%	1/292
Morquio Syndrome: Type A	♂ Colombian: 1/257	85.00%	1/1,713	Niemann-Pick Disease: Type A	♂ Ashkenazi Jewish: 1/101	95.00%	1/2,020
	o [®] European: 1/257	20.97%	1/325	Niemann-Pick Disease: Type B	♂ Czech: 1/276	83.33%	1/1,656
	of Finnish: 1/257	50.00%	1/514		♂ General: Unknown	19.82%	Unknown
	of Latin American: 1/257	36.11%	1/402		o North African: Unknown	86.67%	Unknown
Morquio Syndrome: Type B	of European: Unknown	83.33%	Unknown		o⁴ Spaniard: Unknown	38.10%	Unknown
Mucolipidosis: Type II/III	of General: 1/158	24.60%	1/210	Niemann-Pick Disease: Type C1	♂ Acadian: Unknown	>99%	Unknown
	♂ Japanese: 1/252 ♂ Korean: Unknown	51.25% 30.00%	1/517 Unknown		♂ General: 1/194	15.60%	1/230
	of Portuguese: 1/176	50.00%	1/352		og Japanese: Unknown	18.18%	Unknown
Mucolipidosis: Type IV	of Ashkenazi Jewish: 1/97	96.15%	1/2,522		of Portuguese: 1/194	25.00%	1/259
Multiple Pterygium Syndrome	of European: Unknown	41.67%	Unknown	Niemann-Pick Disease: Type C2	o' General: 1/194	<i>7</i> 5.00%	1/776
Mulliple Flerygium Syndrome	of Middle Eastern: Unknown	60.00%	Unknown	Nijmegen Breakage Syndrome	♂ Eastern European: 1/155	>99%	<1/15,50 0
	of Pakistani: Unknown	50.00%	Unknown	Nonsyndromic Hearing Loss and	♂ Ashkenazi Jewish: 1/20	95.83%	1/480
Multiple Sulfatase Deficiency	of Ashkenazi Jewish: 1/320	95.00%	1/6,400	Deafness: GJB2 Related	O / GIRONGEI SOWISH. 1/ 20	70.00%	17 400
mompro contract 2 charactery	of General: 1/501	18.18%	1/612		♂ Chinese: 1/100	82.26%	1/564
Muscle-Eye-Brain Disease	o' European: Unknown	54.17%	Unknown		o European: 1/53	82.47%	1/302
,	of Finnish: 1/112	97.37%	1/4,256		o⁴ Ghanaian: Unknown	90.91%	Unknown
	o' General: Unknown	23.53%	Unknown		♂ Indian: Unknown	66.98%	Unknown
	o" United States: Unknown	25.00%	Unknown		♂ Israeli: 1/16	93.10%	1/232
Navajo Neurohepatopathy	o' Navajo: 1/39	>99%	<1/3,900		♂ Japanese: 1/75	<i>7</i> 5.00%	1/300
Nemaline Myopathy: NEB Related	♂ Ashkenazi Jewish: 1/108	>99%	<1/10,80		o Roma: Unknown	>99%	Unknown
			0		♂ United States: 1/34	45.22%	1/62
Nephrotic Syndrome: Type 1	o' Finnish: 1/45 o' US Amish: 1/12	76.84% 50.00%	1/194 1/24	Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related	o⊓ Ashkenazi Jewish: 1/180	>99%	<1/18,00 0
Nephrotic Syndrome: Type 2	of Israeli-Arab: Unknown	55.56%	Unknown	Nonsyndromic Hearing Loss and	♂ Balinese: 1/6	>99%	<1/600
	♂ Pakistani: Unknown	20.00%	Unknown	Deafness: MYO 15A Related	o [™] Pakistani: 1/77	24.00%	1/101
	o ^a Polish: Unknown	16.18%	Unknown	Oculocutaneous Albinism: Type 1	o' European: 1/101	26.32%	1/137
	oʻ Saudi Arabian: Unknown	72.73%	Unknown	,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,	of Hutterite: 1/7	>99%	<1/700
					of Moroccan Jewish: 1/30	71.88%	1/107
							,





Detection

Rate

27.78%

47.92%

81.82%

70.00%

65.62%

83.33%

>99%

>99%

95.65%

>99%

>99%

58.33% 53.95%

20.22%

52.38%

>99%

>99% 91.67%

70.59% 62.12%

52.68% 70.31%

>99%

25.00%

33.33%

86.67%

42.86%

58.33%

78.00%

56.25%

75.00%

52.38%

65.00%

>99%

87.50%

62.50%

>99%

50.00%

Residual

Risk

Unknown

1/273

1/336

Unknown

Unknown

Unknown

Unknown

<1/4,200

1/5,750

<1/3,700

<1/22,50 0 1/242

1/20

Unknown

1/443

<1/20,00

Unknown

1/276 1/173

1/459 1/399

Unknown

Unknown

Unknown

Unknown

1/765

1/319

1/38

1/464

Unknown

1/728

1/382

1/94

<1/900

Unknown

1/669

<1/1,000

Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	
	♂ Puerto Rican: Unknown	91.67%	Unknown		♂ Italian: Unknown	
Oculocutaneous Albinism: Type 3	♂ Black South African: 1/47	94.74%	1/893		♂ Norwegian: 1/142	
Oculocutaneous Albinism: Type 4	o' Japanese: 1/146	58.33%	1/350		♂ Sardinians: 1/61	
Omenn Syndrome: DCLRE1C Related	of Apache: 1/29	>99% 97.22%	<1/2,900		♂ United Kingdom: Unknown	
0 0 1 04000111	o ^a Navajo: 1/29		1/1,044		♂ United States: Unknown	
Omenn Syndrome: RAG2 Related	o [™] Arab: Unknown	40.00%	Unknown	Pontocerebellar Hypoplasia: EXOSC3	o General: Unknown	
	♂ Non-Ashkenazi Jewish: Unknown	70.00%	Unknown	Related Pontocerebellar Hypoplasia: RARS2	o" Sephardic Jewish:	
Ornithine Translocase Deficiency	o' French Canadian: 1/20	95.00%	1/400	Related	Unknown	
	♂ Italian: Unknown	18.75%	Unknown	Pontocerebellar Hypoplasia: SEPSECS	♂ Iraqi Jewish: 1/42	
	♂ Japanese: Unknown	60.00%	Unknown	Related		
Osteopetrosis: TCIRG1 Related	♂ Ashkenazi Jewish: 1/350	>99%	<1/35,00 0	Pontocerebellar Hypoplasia: TSEN54 Related	o⁴ European: 1/250	
	o' Costa Rican: Unknown	>99%	Unknown	Pontocerebellar Hypoplasia: VPS53	♂ Moroccan Jewish: 1/37	
	o' General: 1/251	25.00%	1/335	Related	3.4.11	
POLG Related Disorders: Autosomal Recessive	og Belgian: Unknown	85.00%	Unknown	Pontocerebellar Hypoplasia: VRK1 Related	♂ Ashkenazi Jewish: 1/225	
	o'' Finnish: 1/140	>99%	<1/14,00	Primary Carnitine Deficiency	o⁴ European: 1/101	
	,		0		o⁴ Faroese: 1/9	
	o" General: Unknown	93.10%	Unknown		♂ General: Unknown	
	♂ Norwegian: Unknown	>99%	Unknown	Primary Ciliary Dyskinesia: DNA11 Related	♂ European: 1/211	
Papillon-Lefevre Syndrome	♂ General: Unknown	35.29%	Unknown		♂ Ashkenazi Jewish: 1/200	
	♂ Indian Jewish: Unknown	>99%	Unknown	Primary Ciliary Dyskinesia: DNAI2 Related	O Ashkendzi Jewish: 1/200	
	o⁴ Turkish: Unknown	50.00%	Unknown	Primary Congenital Glaucoma	♂ Moroccan: Unknown	
Pendred Syndrome	♂ European: 1/58	42.11%	1/100		♂ Saudi Arabian: 1/23	
	♂ Japanese: Unknown	45.83%	Unknown		♂ Turkish: 1/51	
	o [*] Pakistani: Unknown	29.82%	Unknown	Primary Hyperoxaluria: Type 1	o⁴ Dutch: 1/174	
Persistent Mullerian Duct Syndrome: Type I	of General: Unknown	28.12%	Unknown		♂ General: 1/189	
Persistent Mullerian Duct Syndrome:	o⁴ General: Unknown	78.12%	Unknown	Primary Hyperoxaluria: Type 2	♂ General: Unknown	
Туре II				Primary Hyperoxaluria: Type 3	O' Ashkenazi Jewish: Unknown	
Phenylalanine Hydroxylase Deficiency	o⁴ Arab: Unknown	46.08%	Unknown		♂ European: Unknown	
	♂ Ashkenazi Jewish: 1/224	44.44%	1/403	Progressive Familial Intrahepatic	o³ European: Unknown	
	od Brazilian: 1/71	56.41%	1/163	Cholestasis: Type 2		
	od Chinese: 1/51	76.57%	1/218	Propionic Acidemia: PCCA Related	o⁴ Japanese: 1/102	
	o' Cuban: 1/71	69.64%	1/234	Propionic Acidemia: PCCB Related	♂ General: 1/182	
	♂ European: 1/51	73.00%	1/189		♂ Greenlandic Inuit: 1/16	
	♂ French Canadian: 1/80	76.27%	1/337		♂Japanese: 1/102	
	♂ Iranian: 1/31	66.94%	1/94		♂ Korean: Unknown	
	♂ Korean: 1/51	57.58%	1/120		♂ Latin American: 1/182	
	o ^a Non-Ashkenazi Jewish: Unknown	63.64%	Unknown		of Spaniard: 1/182	
	o⁴ Slovakian Gypsy: 1/39	>99%	<1/3,900	Pseudocholinesterase Deficiency	♂ General: 1/33	
	♂ Spanish Gypsy: 1/4	93.75%	1/64		♂ Iranian Jewish: 1/9	
	o' Taiwanese: Unknown	83.10%	Unknown	Pycnodysostosis	o⁴ Danish: Unknown	
	o'' US Amish: 1/16	86.84%	1/122	Pyruvate Carboxylase Deficiency	♂ General: 1/251	
Polyglandular Autoimmune Syndrome: Type I	of Finnish: 1/80	90.48%	1/840	Pyruvate Dehydrogenase Deficiency	on Native American: 1/10 on General: Unknown	
'/5" '	♂ Iranian Jewish: 1/48	>99%	<1/4,800	, , , , , , , , , , , , , , , , , , , ,		





Carrier Rate of Colombian (Antioquia): Unknown of Newfoundlander: 1/106 of Swedish: 1/84	Detection Rate 92.86%	Residual Risk	Disease	Carrier Rate	Detection Rate	
Unknown O' Newfoundlander: 1/106	92.86%					Risk
o Newfoundlander: 1/106		Unknown		♂ General: Unknown	<i>7</i> 5.00%	Unknown
♂ Swedish: 1/84	>99%	<1/10,60	Sulfate Transporter-Related Osteochondrodysplasia	♂ Finnish: 1/51	95.83%	1/1,224
O' Swedish: 1/84		0		♂ General: 1/100	70.00%	1/333
	>99%	<1/8,400	Tay-Sachs Disease	♂ Argentinian: 1/280	82.35%	1/1,587
♂ Dutch: 1/32	>99%	<1/3,200		♂ Ashkenazi Jewish: 1/29	99.53%	1/6,177
♂ North African Jewish: Unknown	>99%	Unknown		♂ Cajun: 1/30	>99%	<1/3,000
ರ್ Yemenite Jewish: Unknown	>99%	Unknown		o' European: 1/280	25.35%	1/375
♂ Ashkenazi Jewish: 1/91	>99%	<1/9,100		♂ General: 1/280	32.09%	1/412
♂ Ashkenazi Jewish:	>99%	Unknown		o' Indian: Unknown	85. <i>7</i> 1%	Unknown
Unknown				♂ Iraqi Jewish: 1/140	56.25%	1/320
♂ Non-Ashkenazi Jewish: 1 /32	>99%	<1/3,200		o' Japanese: 1/127	82.81%	1/739
•	72 68%	1/582		o' Moroccan Jewish: 1/110	22.22%	1/141
o ceneral. Ty 107	7 2.00%	1,002		o' Portuguese: 1/280	92.31%	1/3,640
o European: Unknown	33.33%	Unknown		o' Spaniard: 1/280	67.65%	1/865
♂ Scandinavian: 1/200	94.27%	1/3,491		♂ United Kingdom: 1/161	71.43%	1/564
♂ Argentinian: Unknown	95.45%	Unknown	Trichohepatoenteric Syndrome: Type 1	o' European: 1/434	42.86%	1/760
o [®] Cypriot: 1/7	80.00%	1/35		o' South Asian: 1/434	66.67%	1/1,302
o ≀Italian: Unknown	29.17%	Unknown	Tyrosine Hydroxylase Deficiency	o' General: Unknown	36.11%	Unknown
o Spaniard: Unknown	64.29%	Unknown	Tyrosinemia: Type I	♂ Ashkenazi Jewish: 1/158	>99%	<1/15,80
♂ Australasian: 1/119	44.12%	1/213		~ E 1 /144	E7 140/	1 /207
o Dutch: 1/78	63.10%	1/211		. ,		1/387
♂ European: 1/159	35.16%	1/245		•		1/4,428
o United States: 1/159	32.14%	1/234		•		Unknown
♂ Australasian: 1/230	28.00%	1/319	Tyracinamia: Tyrac II			1/418
♂ Dutch: Unknown	42.31%	Unknown	, ,,	•		1/273
♂ European: Unknown	52.38%	Unknown	Osher Syndronie. Type 16	. ,		1/164
♂Japanese: 1/200	81.82%	1/1,100		,		Unknown
o Dutch: 1/346	75.00%	1/1,384				1/173
♂ Greek: 1/415	25.00%	1/553	Usher Syndrome: Type 1C			1/7,216
♂ Moroccan: Unknown	80.00%	Unknown	Osher Syndrome. Type TC	•		1/1,362
oਾ Spaniard: Unknown	64.29%	Unknown	Usher Syndrome: Type 1D	•		1/391
♂ General: 1/501	83.33%	1/3,006	, , , , ,	,		1/2,016
♂ Ashkenazi Jewish: 1/15	65.00%	1/43	Usher Syndrome: Type 2A	or Chinese: Unknown	83.33%	Unknown
♂ African American: 1/10	>99%	<1/1,000		♂ European: 1/136	40.00%	1/227
♂ Hispanic American: 1/95	>99%	<1/9,500		of French Canadian:	66.67%	Unknown
o³ Dutch: Unknown	25.86%	Unknown			46 02%	1/256
♂ Swedish: 1/205	>99%	<1/20,50		,		Unknown
♂ General: 1/251	35.71%	0 1/390		of Non-Ashkenazi Jewish:	61.11%	Unknown
♂ Brazilian: 1/94	79.17%	1/451		Unknown		
od European: 1/71	84.72%	1/465		of Scandinavian: 1/125	39.22%	1/206
් Japanese: Unknown	71.43%	Unknown		of Spaniard: 1/133	39.02%	1/218
♂ United States: 1/70	95.00%	1/1,400	Usher Syndrome: Type 3	o⁴ Ashkenazi Jewish: 1/120	>99%	<1/12,00 0
,		, ,		of Finnish: 1 /124	\000 %	<1/13,40
	Inknown Inknow	## Non-Ashkenazi Jewish:	## Non-Ashkenazi Jewish:	Inknown 7 Non-Adhkenazi Jewish:	Inthonorm	Infancoun



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	♂ General: 1/224	31.25%	1/326
	og Japanese: 1/87	65.62%	1/253
Wilson Disease	♂ Ashkenazi Jewish: 1/100	>99%	<1/10,00 0
	♂ Canarian: 1/26	68 <i>.</i> 75%	1/83
	♂ Chinese: 1/51	55.97%	1/116
	og Cuban: Unknown	22.22%	Unknown
	o European: 1/93	41.64%	1/159
	o'' Greek: 1/90	44.94%	1/163
	♂ Korean: 1/88	51.53%	1/182
	♂ Spaniard: 1/93	38.18%	1/150
Wolcott-Rallison Syndrome	og Saudi Arabian: Unknown	66.67%	Unknown
Wolman Disease	♂ Iranian Jewish: 1/33	>99%	<1/3,300
Xeroderma Pigmentosum: Group A	♂ Japanese: 1/75	97.62%	1/3,150
	o North African: Unknown	87.50%	Unknown
	♂ Tunisian: 1/112	90.91%	1/1,232
Xeroderma Pigmentosum: Group C	♂ Moroccan: 1/71	<i>7</i> 6.19%	1/298
	♂ Tunisian: 1/51	>99%	<1/5,100
Zellweger Spectrum Disorders: PEX1 Related	o⁴ European: 1/139	70.27%	1/468
	♂ General: 1/139	67.84%	1/432
Zellweger Spectrum Disorders: PEX10 Related	♂ Japanese: Unknown	40.74%	Unknown
Zellweger Spectrum Disorders: PEX2 Related	♂ Ashkenazi Jewish: 1/123	>99%	<1/12,30 0
Zellweger Spectrum Disorders: PEX6 Related	o'' General: 1/288	30.00%	1/411