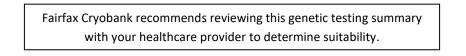


Donor 5813

Genetic Testing Summary



Last Updated: 11/15/18

Donor Reported Ancestry: Ecuadorian, Brazilian

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**

Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/ and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by gene sequencing in the CFTR gene	1/1250
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/1061
Expanded Genetic Disease Testing Panel attached- 289 diseases by gene sequencing	Carrier: Biotinidase Deficiency (BTD) 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: Fairfax Cryobank -

Physician: Report Generated: 2018-04-09 Donor 5813

DOB:

Gender: Male Ethnicity: Latin American Procedure ID: 115040 Kit Barcode: Specimen: Blood, #117572 Specimen Collection: 2018-03-23 Specimen Received: 2018-03-24 Specimen Analyzed: 2018-04-06

TEST INFORMATION

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

SUMMARY OF RESULTS: MUTATION(S) IDENTIFIED

Disease	Donor 5813	Partner Not Tested
Biotinidase Deficiency (BTD)Carrier (1 abnormal copy)O High ImpactMutation: c. 1330G>C (p.D444H)O Treatment BenefitsMethod: Genotyping & Sequencing		
	Reproductive Risk & Next Steps : partner testing.	Reproductive risk detected. Consider

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

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

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

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





ADDITIONAL RESULTS: NO INCREASED REPRODUCTIVE RISK

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

Disease (Gene)	Donor 5813	Partner Not Tested
Spinal Muscular Atrophy: SMN1 Linked (SMN1)*	SMN1 Copy Number: 2 or more copies Method: Genotyping & dPCR	

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

	Detection Rate	Pre-Test Carrier Risk	Post-Test Carrier Risk (2 SMN1 copies)	Post-Test Carrier Risk (3 SMN1 copies)
European	95%	1/35	1/632	1/3,500
Ashkenazi Jewish	rish 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.



Biotinidase Deficiency (BTD)

Biotinidase deficiency is an inherited disorder associated with skin and neurological problems if left untreated. This condition is caused by mutations in the BTD gene, which is typically responsible for extracting and recycling vitamin H (biotin) for use in various parts of the body. Affected individuals typically exhibit signs and symptoms within the first few months of life. Children with profound biotinidase deficiency often experience seizures, weak muscle tone (hypotonia), breathing problems, and delayed development. If left untreated, the disorder can lead to hearing and vision loss, problems with movement and balance (ataxia), skin rashes, hair loss (alopecia), and a fungal infection called candidiasis. Partial biotinidase deficiency is a milder form of the condition and affected children may experience hypotonia, skin rashes, and alopecia, but these symptoms often only appear during illness, infection, or other times of stress.

O High Impact

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

O Treatment Benefits

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

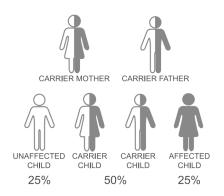
Clinical Information

- Physical Impairment
- Cognitive Impairment

Shortened Lifespan

Effective Treatment

Inheritance: Autosomal Recessive



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

Prognosis

Prognosis is generally favorable. Immediate treatment and lifelong management with biotin supplements can prevent many of the complications. However, if vision and hearing loss and developmental delay occur prior to treatment, they are usually irreversible, even with biotin therapy.

Treatment

Treatment involves immediate treatment and lifelong management with biotin supplements, which typically reverses and prevents most symptoms of this condition. Other treatment includes routine assessment of vision and hearing to ensure that a treated individual is not symptomatic.

Risk Information

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
General	78.32%	1/123	1/567

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



Methods and Limitations

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

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

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

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

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



CarrierMap[™]

Diseases & Mutations Assayed

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

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

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

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

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

3-Beta-Hydroxysteroid Dehydrogenase Deficiency (HSD3B2): Mutations (6): O 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): d' Genotyping | c.1155A>C (p.R385S), c.1310T>C (p.L437P) Sequencing | NM_020166:1-

3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCB Related (MCCC2): Mutations (8): O' 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.1437V) Sequencing | NM_022132:1-17

3-Methylglutaconic Aciduria: Type 3 (OPA3): Mutations (3): of 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): Ot 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): Or 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.I2949fs), c.5836T>C (p.W1946R), c.3161T>C (p.F1054S) Sequencing | NM_014363:2-10

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

Acrodermatitis Enteropathica (SLC39A4): Mutations (7): O' Genotyping | c. 1223-1227delCCGGG, c.968-971delAGTC, c.318C>A (p.N106K), c.599C>T (p.P200L), c.1120G>A (p.G374R), c.909G>C (p.Q303H), c.989G>A (p.G330D) Sequencing | NM_130849:1-12 Acute Infantile Liver Failure: TRMU Related (TRMU): Mutations (5): d' 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): of 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): O^{*} Genotyping | c.986C>T (p.A329V), c.872C>T (p.S291L), c.646G>A (p.G216R), c.632G>A (p.R211H), c.631C>T (p.R211C), c.596A>C (p.Q199P), c.536C>A (p.A179D), c.529G>A (p.V177M), c.467G>A (p.R156H), c.466C>T (p.R156C), c.454C>A (p.L152M), c.445C>T (p.R149W), c.419G>A (p.G140E), c.385G>A (p.V129M), c.320T>C (p.L107P), c.302G>A (p.R101Q), c.302G>T (p.R101L), c.301C>T (p.R101W), c.248C>A (p.A83D), c.220G>T (p.G74C), c.58G>A (p.G20R), c.43C>G (p.H15D) Sequencing | NM_000022:1-12

Alkaptonuria (HGD): Mutations (14): of 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): d^a Genotyping | SEA deletion, c.207C>A

(p.N69K), c.223G>C (p.D75H), c.2T>C, c.207C>G (p.N69K), c.340_351 delCTCCCCGCCGAG (p.L114_E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G Alpha-1-Antitrypsin Deficiency (SERPINA1): Mutations (4): of Genotyping |

c.226_228delTTC (p.76delF), c.1131 A>T (p.L377F), c.187C>T (p.R63C), c.1096G>A (p.E366K) Sequencing | NM_001127701:1-7

Alpha-Mannosidosis (MAN2B1): Mutations (3): O' Genotyping | c.2426T>C (p.L809P), c.2248C>T (p.R750W), c.1830+1G>C (p.V549_E610del) Sequencing | NM_000528:1-24 Alport Syndrome: COL4A3 Related (COL4A3): Mutations (3): of Genotyping | c.4420_4424delCTTTT, c.4441C>T (p.R1481X), c.4571C>G (p.S1524X) Sequencing | NM 000091:2-52

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

Amegakaryocytic Thrombocytopenia (MPL): Mutations (23): d' Genotyping | c.79+2T>A (IVS1+2T>A), c.127C>T (p.R43X), c.305G>C (p.R102P), c.823C>A (p.P275T), c.304C>T (p.R102C), c.376delT (F126Lfs), c.268C>T (p.R90X), c.235_236delCT (p.L79fs), c.367C>T (p.R123X), c.460T>C (p.W154R), c.1305G>C (p.W435C), c.770G>T (p.R257L), c.407C>T (p.P136L), c.407C>A (p.P136H), c.1781T>G (p.L594W), c.311T>C (p.F104S), c.556C>T (p.Q186X), c.1473G>A (p.W491X), c.1499delT (p.L500fs), c.769C>T (p.R257C), c.1904C>T (p.P635L), c.213-1G>A (IVS2-1G>A), c.1566-1G>T (IVS10-1G>T) Sequencing | NM_005373:1-

Andermann Syndrome (SLC12A6): Mutations (5): of Genotyping | c.2436delG (p.T813fsX813), c.901 delA, 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): of Genotyping | c.365G>A (p.W122X), c.871C>T (p.R291X), c.869C>G (p.T290S), c.703G>C (p.G235R), c.32T>C (p.111T), c.413G>T (p.G138V), c.57+1G>A, c.61C>T (p.R21X), c.263_266delAGAA (p.K88fs), c.77delA (p.E26fs), c.844delC (p.L282fs), c.466-2A>G, c.703G>A (p.G235R) Sequencing | NM_000045:1-8

Argininosuccinate Lyase Deficiency (ASL): Mutations (7): Or 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): O^{*} Genotyping | c. 1222delC (p.K409fs), c.296+1G>A (IVS3+1G>A), c.468delC, c.629-3C>A (IVS4-3C>A), c.743+2T>C (IVS6+2T>C), c.1123C>T (p.R375C), c.1303C>T (p.R435C), c.1094G>A (p.R365Q), c.1310G>A (p.C437Y), c.628G>A (p.E210K) Sequencing | NM_000103:2-10

Arthrogryposis, Mental Retardation, & Seizures (SLC35A3): Mutations (2): O Genotyping | c.1012A>G (p.S338G), c.514C>T (p.Q172X) Sequencing | NM_001271685:1-8 Asparagine Synthetase Deficiency (ASNS): Mutations (1): d^a Genotyping | c.1084T>G (p.F362V) Sequencing | NM_001673:3-13

Aspartylglycosaminuria (AGA): Mutations (7): d Genotyping | c.200_201 delAG, c.488G>C (p.C163S), c.214T>C (p.S72P), c.916T>C (p.C306R), c.904G>A (p.G302R), c.302C>T (p.A101V), c.179G>A (p.G60D) Sequencing | NM_000027:1-9

Ataxia with Vitamin E Deficiency (TTPA): Mutations (14): or 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^a 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): of 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.1222V), c.10036T>C (p.C3346R), c.383delC, c.4220T>G (p.L1407R), c.11612G>A (p.W3871X), c.5984A>G (p.E1995G), c.10637delT (p.V3546fs), c.3747T>G (p.C1249W), c.5750A>G (p.Q1917R), c.10865G>A (p.C3622Y), c.50C>T (p.A17V), c.8063G>T (p.C2688F), c.10402A>G (p.I3468V), c.1529delG (p.G510fs), c.657C>T (p.G219G), c.5513A>G (p.Y1838C), c.10856delA (p.K3619fs), c.5381-9T>G (IVS33-9T>G), c.3229-2A>C (IVS28-2A>C), c.10505A>T (p.E3502V), c.2269A>C (p.I757L), c.4165C>A (p.P1389T), c.10364delC (p.S3455fs), c.7350+653A>G (IVS46+653A>G) Sequencing | NM_138694:2-67

Bardet-Biedl Syndrome: BBS1 Related (BBS1): Mutations (3): Or 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): of Genotyping |

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

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

Bardet-Biedl Syndrome: BBS12 Related (BBS12): Mutations (5): & 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): O' 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): O" 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): O[®] 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): J G 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.51 delC (p.K18Rfs), c.93-21 G>A, c.92+1G>A, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>T, c.-50A>C, c.-78a>g, c.-79A>G, c.-81A>G, c.52A>T (p.K18X), c.-137c>g, c.-138c>t, c.-151C>T, c.118C>T (p.Q40X), c.169G>C (p.G57R), c.295G>A (p.V99M), c.415G>C (p.A139P), c.47G>A (p.W16X), c.48G>A (p.W16X), c.-80t>a, c.2T>C, c.75T>A (p.G25G), c.444+111A>G, c.-29G>A, c.68_74delAAGTTGG, c.92G>C (p.R31T), c.92+1G>T, c.93-15T>G, c.93-1G>C, c.112delT, c.113G>A (p.W38X), c.114G>A (p.W38X), c.126delC, c.444+113A>G, c.250delG, c.225delC, c.383_385delAGG (p.Q128_A129delQAinsP), c.321_322insG (p.N109fs), c.316-1G>T, c.316-2A>C, c.287_288insA (p.L97fs), c.271 G>T (p.E91X), c.203_204delTG (p.V68Afs), c.154delC (p.P52fs), c.135delC (p.F46fs), c.92+2T>A, c.92+2T>C, c.90C>T (p.G30G), c.84_85insC (p.L29fs), c.59A>G (p.N20S), c.46delT (p.W16Gfs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.2T>G, c.1A>G (p.M1V), c.-137c>t, c.-136C>G, c.-142C>T, c.-140c>t Sequencing | NM_000518:1-3

Beta-Hexosaminidase Pseudodeficiency (HEXA): Mutations (2): O' Genotyping | c.739C>T (p.R247W), c.745C>T (p.R249W) Sequencing | NM_000520:1-14

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

Biotinidase Deficiency (BTD): Mutations (21): of Genotyping |

c.98_104delGCGGCTGinsTCC (p.C33FfsX68), c.1368A>C (p.Q456H), c.755A>G (p.D252G), c.1612C>T (p.R538C), c.235C>T (p.R79C), c.100G>A (p.G34S), c.1330G>C (p.D444H), c.511G>A (p.A171T), c.1207T>G (p.F403V), c.470G>A (p.R157H), c.1595C>T (p.T532M), c.1489C>T (p.P497S), c.341G>T (p.G114V), c.1052delC (p.T351fs), c.393delC (p.F131Lfs), c. 1049delC (p.A350fs), c. 1239delC (p.Y414lfs), c. 1240_1251 delTATCTCCACGTC (p.Y414_V417del), c.278A>G (p.Y93C), c.595G>A (p.V199M), c.933delT (p.S311Rfs) Sequencing | NM_000060:1-4

Bloom Syndrome (BLM): Mutations (25): Or Genotyping |

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

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

Carnitine Palmitoyltransferase IA Deficiency (CPT1A): Mutations (10): 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 (19): of Genotyping | c.109_110insGC, c.1238_1239delAG, c.1737delC, c.1923_1935delGAAGGCCTTAGAA, c.534_558delGAACCCTGCAAAAAGTGACACTATCinsT, c.1649A>G (p.Q550R), c.1883A>C (p.Y628S), c.359A>G (p.Y120C), c.983A>G (p.D328G), c.149C>A (p.P50H), c.1810C>T (p.P604S), c.1891C>T (p.R631C), c.338C>T (p.S113L), c.370C>T (p.R124X), c.680C>T (p.P227L), c.1646G>A (p.G549D), c.452G>A (p.R151Q), c.520G>A (p.E174K), c.1148T>A (p.F383Y) Sequencing | NM_000098:1-5

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

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

Cerebrotendinous Xanthomatosis (CYP27A1): Mutations (14): Or 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): of 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): or Genotyping | c.1024G>A (p.G342R), c.894G>A (p.Q298X), c.883C>T (p.H295Y), c.652C>T (p.R218X) Sequencing | NM_001127605:2-10

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

Chronic Granulomatous Disease: CYBA Related (CYBA): Mutations (12): O' 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^a 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): 0³ 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): O' 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): O^{*} Genotyping | c.6578T>G (p.L2193R), c.7051C>T (p.R2351X), c.4471G>T (p.E1491X), c.2911C>T (p.R971X), c.7934G>A (p.G2645D), c.10888C>T (p.Q3630X), c.8459T>C (p.I2820T), c.9259_9260insT (p.I3087fs), 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): O^{*} 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): o" Genotyping | c.884G>A (p.R295H) Sequencing | NM_002435:1-8

Congenital Disorder of Glycosylation: Type 1C: ALG6 Related (ALG6): Mutations (4): d^a 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): O' 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): O' Genotyping | c.178+1_178+2insT (IVS2+3insT), c.201_202delCT, c.466-11T>A (IVS4-11T>A), c.64+1G>T (IVS1+1G>T), c.562C>T (p.R188C), c.772C>T (p.Q258X), c.545G>A (p.R182H), c.545G>T

CarrierMap[™]

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

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

Congenital Myasthenic Syndrome: DOK7 Related (DOK7): Mutations (6): Of Genotyping c.601C>T (p.R201X), c.539G>C (p.G180A), c.548_551 delTCCT (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): O Genotyping | c.264C>A (p.N88K), c.41T>C (p.L14P), c.807C>A (p.Y269X), c.548_549insGTTCT (p.L183fs), c.46_47insC (p.L16fs), c.133G>A (p.V45M), c.848T>C (p.L283P), c.484G>A (p.E162K), c.490C>T (p.R164C), c.-210A>G, c.193-15C>A (IVS1-15C>A) Sequencing | NM_005055:1-8

Congenital Neutropenia: Recessive (HAX1): Mutations (6): d' Genotyping | c. 121_125insG, c. 130_131 insA, c.431 insG, c.91 delG, c.256C>T (p.R86X), c.568C>T (p.Q190X) Sequencing | NM_006118:1-7

Corneal Dystrophy and Perceptive Deafness (SLC4A11): Mutations (8): of Genotyping | c.1459_1462delTACGinsA (p.487_488delYAinsT), c.2313_2314insTATGACAC,

c.554_561delGCTTCGCC (p.R185fs), c.2566A>G (p.M856V), c.1463G>A (p.R488K), c.2528T>C (p.L843P), c.637T>C (p.S213P), c.2321+1G>A Sequencing | NM_001174090:1-20

Corticosterone Methyloxidase Deficiency (CYP11B2): Mutations (3): O' Genotyping | c.1492A>G (p.T498A), c.541C>T (p.R181W), c.1382T>C (p.L461P) Sequencing | NM_000498:1-9

Crigler-Najjar Syndrome (UGT1A1): Mutations (11): O' Genotyping | c.508_513delTTC (p.170delF), c.1070A>G (p.Q357R), c.1021C>T (p.R341X), c.1124C>T (p.S375F), c.840C>A (p.C280X), c.991C>T (p.Q331X), c.923G>A (p.G308E), c.1198A>G (p.N400D), c.992A>G (p.Q331R), c.44T>G (p.L15R), c.524T>A (p.L175Q) Sequencing | NM_000463:1-5

Cystic Fibrosis (CFTR): Mutations (149): d' 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.S1251N), c.3846G>A (p.W1282X), c.3848G>T (p.R1283M), c.532G>A (p.G178R), c.988G>T (p.G330X), c.1090T>C (p.S364P), c.3302T>A (p.M1101K), c.617T>G (p.L206W), c.14C>T (p.P5L), c.19G>T (p.E7X), c.171G>A (p.W57X), c.313delA (p.1105fs), c.328G>C (p.D110H), c.580-1G>T, c.1055G>A (p.R352Q), c.1075C>A (p.Q359K), c.1079C>A (p.T360K), c.1647T>G (p.S549R), c.1976delA (p.N659fs), c.2290C>T (p.R764X), c.2737_2738insG (p.Y913X), c.3067_3072delATAGTG (p.I1023_V1024delT), c.3536_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.54-5940_273+10250del21080bp (p.S18fs), c.4364C>G (p.S1455X), c.4003C>T (p.L1335F), c.2538G>A (p.W846X), c.200C>T (p.P67L), c.4426C>T (p.Q1476X), c.1116+1G>A,

c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.263T>G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.658C>T (p.Q220X), c.868C>T (p.Q290X), c.1526delG (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_1611delAC (p.D537fs), c.3254A>G (p.H1085R), c.496A>G (p.K166E),

c.1408_1417delGTGATTATGG (p.V470fs), c.1585-8G>A, c.2909G>A (p.G970D), c.653T>A (p.L218X), c. 1175T>G (p.V392G), c.3139_3139+1delGG, c.3717+4A>G (IVS22+4A>G) Sequencing | NM_000492:1-27

Cystinosis (CTNS): Mutations (14): d' Genotyping | c. 18_21 delGACT, c.198_218delTATTACTATCCTTGAGCTCCC, c.283G>T (p.G95X), c.414G>A (p.W138X), c.506G>A (p.G169D), c.613G>A (p.D205N), c.473T>C (p.L158P), c.329G>T (p.G110V), c.416C>T (p.S139F), c.589G>A (p.G197R), c.969C>G (p.N323K), c.1015G>A (p.G339R), c.-39155_848del57119, c. 199_219delATTACTATCCTTGAGCTCCCC (p.167_P73del) Sequencing | NM 001031681:1,3-13

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

Cystinuria: Type I (SLC3A1): Mutations (10): d' Genotyping | c.1400T>C (p.M467T), c.2033T>C (p.L678P), c.542G>A (p.R181Q), c.1955C>G (p.T652R), c.1843C>A (p.P615T), c.1085G>A (p.R362H), c.1597T>A (p.Y533N), c.647C>T (p.T216M), c.808C>T (p.R270X), c.452A>G (p.Y151C) Sequencing | NM_000341:1-10

D-Bifunctional Protein Deficiency (HSD17B4): Mutations (6): O' 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^a Genotyping | c.215A>G (p.N72S), c.1144G>A (p.E382K) Sequencing | NM_000352:1-39

Du Pan Syndrome (GDF5): Mutations (4): O^{*} 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): of 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): O' Genotyping | c.2470_2471insG, c.5820G>A (p.P1940P), c.933C>A (p.Y311X), c.4039G>C (p.G1347R), c.8393T>A (p.M2798K), c.425A>G (p.K142R), C.8441-

14_8435delGCTCTTGGCTCCAGGACCCCT, c.4783-1G>A, c.7344G>A (p.V2448X), c.4991G>C (p.G1664A), c.497_498insA (p.V168GfsX179) Sequencing | NM_000094:1-118

Ehlers-Danlos Syndrome: Type VIIC (ADAMTS2): Mutations (2): of 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): O' Genotyping | c.919T>C (p.S307P), c.1694delC (p.A565VfsX23), c.734delT (p.L245fs), c.910-911 insA (p.R304fs), c.2635C>T (p.Q879X), c.1868T>C (p.L623Q), c.

1858_1879delTTGGGCCGACTGGGCGGCCTC (p.L620_L626del), c.1886+5G>T, c.1098+1G>A, c.1018C>T (p.R340X) Sequencing | NM_153717:2-21

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

Enhanced S-Cone (NR2E3): Mutations (5): of Genotyping | c.932G>A (p.R311Q), c.227G>A (p.R76Q), c.119-2A>C, c.226C>T (p.R76W), c.747+1G>C (IVS5+1G>C) Sequencing | NM 016346:1-8

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

Familial Chloride Diarrhea (SLC26A3): Mutations (6): or Genotyping | c.344delT (p.11151), c.559G>T (p.G187X), c.951 delGGT (p.V318del), c.1386G>A (p.W462X), c.371 A>T (p.H124L), c.2023_2025dupATC (p.1675L) Sequencing | NM_000111:2-21

Familial Dysautonomia (IKBKAP): Mutations (4): d^a 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.1387 delF), 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): o" Genotyping | c.776A>G (p.H259R), c.36C>A (p.Y12X), C.C761T (p.P254L), c.G-134T, c.844G>A (p.E282K), c.440T>C (p.L147P) Sequencing | NM_000525:1

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

Fanconi Anemia: Type A (FANCA): Mutations (10): 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): of 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): of Genotyping | c.1480+1G>C, c.307+1G>C, c.1794_1803delCTGGATCCGT (p.W599Pfs), c.637_643delTACCGCC (p.Y213K+4X), c.925-2A>G Sequencing | NM_004629:1-14

Fanconi Anemia: Type J (BRIP1): Mutations (1): O' Genotyping | c.2392C>T (p.R798X) Sequencing | NM_032043:2-20

Fumarase Deficiency (FH): Mutations (1): 0^a Genotyping | c.1431_1433insAAA Sequencing | NM 000143:1-10

GM1-Gangliosidoses (GLB1): Mutations (17): d' 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),

CarrierMap™

c.1369C>T (p.R457X), c.145C>T (p.R49C), c.202C>T (p.R68W), c.245C>T (p.T82M), c.601C>T (p.R201C), c.622C>T (p.R208C), c.1370G>A (p.R457Q), c.176G>A (p.R59H), c.367G>A (p.G123R), c.152T>C (p.I51T), c.1771T>A (p.Y591N), c.1577_1578insG Sequencing | NM 000404:1-16

GRACILE Syndrome (BCS1L): Mutations (12): d^a 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): of 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): of 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): d^a Genotyping | c.1153G>T (p.E385X), c.857G>A (p.G286D), c.2002A>C (p.T668P), c.1700A>C (p.Y567S), c.1586C>T (p.T529M), c.1472delA (p.K491fs), c.913A>G (p.I305V), c.683_694delATCTCTGGGAGTinsCTC (p.N228_S232del5insTP), c.246A>G (p.182M), c.1161+6555_*9573del31670bp Sequencing | NM_000153:2-17

Glutaric Acidemia: Type I (GCDH): Mutations (8): O^a Genotyping | c. 1204C>T (p.R402W), c. 1262C>T (p.A421V), c.743C>T (p.P248L), c.1093G>A (p.E365K), c.877G>A (p.A293T), c.1083-2A>C (IVS10-2A>C), c.680G>C (p.R227P), c.1198G>A (p.V400M) Sequencing | NM 000159:2-12

Glutaric Acidemia: Type IIA (ETFA): Mutations (5): of Genotyping | c.797C>T (p.T266M), c.470T>G (p.V157G), c.346G>A (p.G116R), c.809_811 delTAG (p.V270_A271 delinsA), c.963+1delG Sequencing | NM_000126:1-12

Glutaric Acidemia: Type IIB (ETFB): Mutations (2): d^a 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): d' 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): O* 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): O' 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 (15): of Genotyping c.17_18delAG, c.4455delT (p.S1486fs), c.1222C>T (p.R408X), c.16C>T (p.Q6X), c.1384delG (p.V462X), c.2039G>A (p.W680X), c.2590C>T (p.R864X), c.2681+1G>A, c.3439A>G (p.R1147G), c.3682C>T (p.R1228X), c.3965delT (p.V1322AfsX27), c.3980G>A (p.W1327X), c.4260-12A>G (IVS32-12A>G), c.4342G>C (p.G1448R), c.2681+1G>T Sequencing NM 000642:2-34

Glycogen Storage Disease: Type IV (GBE1): Mutations (3): O' 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): d' 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): of 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): Or 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): d^a Genotyping | c.914_915delTT, c.122G>A (p.R41Q), c.208G>C (p.V70L), c.835G>A (p.E279K), c.561+1G>A, c.109G>T (p.E37X), c.561+1G>T Sequencing | NM_000191:1-9

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

Hemochromatosis: Type 3: TFR2 Related (TFR2): Mutations (4): O 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): of Genotyping | c. 19G>A (p.E7K) Sequencing | NM_000518:1-3

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

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

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

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

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

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

Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3): Mutations (6): o" 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): O' Genotyping | c.1470_1486dup16 (p.H497Qfs) Sequencing | NM_000195:3-20

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

Hermansky-Pudlak Syndrome: Type 4 (HPS4): Mutations (7): 0^a Genotyping | c.1876C>T (p.Q626X), c.526C>T (p.Q176X), c.957_958insGCTTGTCCAGATGGCAGGAAGGAG (p.E319_N320ins8), c.634C>T (p.R212X), c.397G>T (p.E133X), c.649G>T (p.E217X), c.2039delC (p.P680fs) Sequencing | NM_152841:1-12

Holocarboxylase Synthetase Deficiency (HLCS): Mutations (7): d^a Genotyping | c.1795+5G>A (IVS10+5G>A), c.780delG, c.710T>C (p.L237P), c.1522C>T (p.R508W), c.1648G>A (p.V550M), c.1513G>C (p.G505R), c.772_781 delACAAGCAAGG (p.T258fs) Sequencing | NM_001242785:4-12

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

Hurler Syndrome (IDUA): Mutations (8): 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): of Genotyping | c.1559delT, c.1133A>T (p.D378V), c.1001G>A (p.G334D), c.571G>A (p.E191K), c.979T>C (p.F327L) Sequencing | NM_000478:2-12

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

Infantile Cerebral and Cerebellar Atrophy (MED17): Mutations (1): O' Genotyping | c.1112T>C (p.L371P) Sequencing | NM_004268:1-12

Isolated Microphthalmia: VSX2 Related (VSX2): Mutations (4): 0^a 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): Or Genotyping | c.941C>T (p.A314V) Sequencing | NM_002225:1-12

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

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

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

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

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

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

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

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

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

Limb-Girdle Muscular Dystrophy: Type 2A (CAPN3): Mutations (6): O^a 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): O^a 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): 0^a Genotyping | c.848G>A (p.C283Y), c.787G>A (p.E263K), c.525delT (p.F175fsX), c.87_88insT (p.G30fs) Sequencing | NM_000231:2-8

Limb-Girdle Muscular Dystrophy: Type 2D (SGCA): Mutations (1): O' Genotyping | c.229C>T (p.R77C) Sequencing | NM_000023:1-9

Limb-Girdle Muscular Dystrophy: Type 2E (SGCB): Mutations (6): 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): O 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): d Genotyping | c.826C>A (p.L276I) Sequencing | NM_001039885:1-4

Lipoprotein Lipase Deficiency (LPL): Mutations (1): of 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

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

MTHFR Deficiency: Severe (MTHFR): Mutations (6): d^a 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): of 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): d^a 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): of Genotyping | c.1114G>T (p.E372X), c.548G>C (p.R183P), c.832G>A (p.G278S), c.970C>T (p.R324X), c.487G>T (p.E163X), c.853C>T (p.R285X) Sequencing | NM_183050:1-10

Maple Syrup Urine Disease: Type 2 (DBT): Mutations (15): 07 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.1401T) Sequencing | NM 001918:1-11

Maple Syrup Urine Disease: Type 3 (DLD): Mutations (8): O' Genotyping | c.104_105insA, c.685G>T (p.G229C), c.214A>G (p.K72E), c.1081A>G (p.M361V), c.1123G>A (p.E375K), c.1178T>C (p.I393T), c.1463C>T (p.P488L), c.1483A>G (p.R495G) Sequencing | NM 000108:1-14

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

Meckel Syndrome: Type 1 (MKS1): Mutations (5): d' Genotyping | c. 1408-35_1408-7del29 (p.G470fs), c.80+2T>C (IVS1+2T>C), c.1024+1G>A (IVS11+1G>A), c.417G>A (p.E139X), c.50insCCGGG (p.D19AfsX) Sequencing | NM_017777:1-18

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (ACADM): Mutations (8): 0^a Genotyping | c.985A>G (p.K329E), c.362C>T (p.T1211), c.595G>A (p.G199R), c.799G>A (p.G267R), c.199T>C (p.Y67H), c.262C>T (p.L88F), c.616C>T (p.R206C), c.617G>A (p.C206H) Sequencing | NM_001127328:1-12

Megalencephalic Leukoencephalopathy (MLC1): Mutations (6): d^a Genotyping | c. 176G>A (p.G59E), c.278C>T (p.S93L), c. 135_136insC (p.C46fsX), c.908_918delTGCTGCTGCTGCTGinsGCA (p.V303GfsX96), c.880C>T (p.P294S), c.178-10T>A Sequencing | NM_139202:2-12

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

Methylmalonic Acidemia: MMAA Related (MMAA): Mutations (14): 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): of 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): o" 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): of Genotyping | c.344G>A (p.C115Y) Sequencing | NM_004553:1-4

Mitochondrial DNA Depletion Syndrome: MNGIE Type (TYMP): Mutations (6): O 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): o" 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): d^{*} Genotyping | c.182G>A (p.R61H), c.788A>G (p.D263G), c.740G>A (p.R247H), c. 1331 G>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): Or Genotyping | c.205T>G (p.F69V), c.485C>T (p.S162F), c.1156C>T (p.R386C), c.901G>T (p.G301C), c.337A>T (p.I113F), c.178G>A (p.D60N) Sequencing | NM_000512:2-14

Morquio Syndrome: Type B (GLB1): Mutations (8): d' 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): o^a Genotyping | c.3503_3504delTC (p.L1168QfsX5), c.3565C>T (p.R1189X), c.1120T>C (p.F374L) Sequencing | NM_024312:1-21

Mucolipidosis: Type IV (MCOLN1): Mutations (5): of 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): Or Genotyping | c.463T>C (p.S155P) Sequencing | NM_182760:1-9

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

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

Nemaline Myopathy: NEB Related (NEB): Mutations (2): of 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.L41Dfs), c.1481delC, c.3325C>T (p.R1109X), c.3478C>T (p.R1160X), c.2335-1G>A

CarrierMap™

Sequencing | NM_004646:1-29

Nephrotic Syndrome: Type 2 (NPHS2): Mutations (27): 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.F185fsX186), c.538G>A (p.V180M), c.503G>A (p.R168H), c.502C>A (p.R168S), c.502C>T (p.R168C), c.479A>G (p.D160G), c.467delT (p.L156fsX180), c.467_468insT (p.L156fsX166), c.419delG (p.G140fsX180), c.413G>A (p.R138Q), c.412C>T (p.R138X), c.353C>T (p.P118L), c.274G>T (p.G92C), c.104_105insG (p.G35fsX69), c.85G>A (p.A29T) Sequencing | NM_014625:1-8

Neuronal Ceroid-Lipofuscinosis: CLN5 Related (CLN5): Mutations (7): O^a Genotyping | c.1175_1176delAT (p.Y392X), c.225G>A (p.W75X), c.835G>A (p.D279N), c.335G>A (p.R112H), c.377G>A (p.C126Y), c.1054G>T (p.E352X), c.1121A>G (p.Y374C) Sequencing | NM 006493:1-4

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

Neuronal Ceroid-Lipofuscinosis: CLN8 Related (CLN8): Mutations (4): O^a Genotyping | c.70C>G (p.R24G), c.789G>C (p.W263C), c.88G>C (p.A30P), c.610C>T (p.R204C) Sequencing I NM 018941:2-3

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

Neuronal Ceroid-Lipofuscinosis: PPT1 Related (PPT1): Mutations (8): of Genotyping | c.223A>C (p.T75P), c.364A>T (p.R122W), c.451C>T (p.R151X), c.29T>A (p.L10X), c.656T>A (p.L219Q), c.322G>C (p.G108R), c.236A>G (p.D79G), c.134G>A (p.C45Y) Sequencing NM 000310:1-9

Neuronal Ceroid-Lipofuscinosis: TPP1 Related (TPP1): Mutations (9): of 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): Or 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): O Genotyping | c.1829_1831delGCC (p.610delR), c.880C>A (p.Q294K), c.1280A>G (p.H427R) Sequencing | NM 000543:1-6

Niemann-Pick Disease: Type C1 (NPC1): Mutations (14): of Genotyping | c.2783A>C (p.Q928P), c.3263A>G (p.Y1088C), c.3467A>G (p.N1156S), c.3107C>T (p.T1036M), c.3182T>C (p.11061T), c.2974G>C (p.G992R), c.2932C>T (p.R978C), c.2848G>A (p.V950M), c.2665G>A (p.V889M), c.2324A>C (p.Q775P), c.1133T>C (p.V378A), c.530G>A (p.C177Y), c.337T>C (p.C113R), c.2974G>T (p.G992W) Sequencing | NM_000271:1-25

Niemann-Pick Disease: Type C2 (NPC2): Mutations (11): d' Genotyping | c.58G>T (p.E20X), c.436C>T (p.Q146X), c.358C>T (p.P120S), c.352G>T (p.E118X), c.332delA (p.N111 lfs), 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): O Genotyping | c.657_661delACAAA (p.K219fs) Sequencing | NM_002485:1-16

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutations (29): O" Genotyping | c.167delT, c.235delC, c.313_326delAAGTTCATCAAGGG, c.358delGAG (p.120delE), c.35delG (p.G12fs), c.370C>T (p.Q124X), c.427C>T (p.R143W), c.109G>A (p.V37I), c.231G>A (p.W77X), c.551G>C (p.R184P), c.71G>A (p.W24X), c.229T>C (p.W77R), c.269T>C (p.L90P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T (p.G12V), c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147K), c.-23+1G>A, c.550C>T (p.R184W), c.-259C>T Sequencing | NM 004004:1-2

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

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

Oculocutaneous Albinism: Type 1 (TYR): Mutations (27): O' Genotyping | c.272G>A (p.C91Y), c.242C>T (p.P81L), c.265T>C (p.C89R), c.1A>G (p.M1V), c.140G>A (p.G47D), c.325G>A (p.G109R), c.568delG (p.G191Dfs), c.707G>A (p.W236X), c.832C>T (p.R278X), c.1118C>A (p.T373K), c.229C>T (p.R77W), c.823G>T (p.V275F), c.32G>A (p.W11X), c.149C>T (p.S50L), c.1467_1468insT (p.A490Cfs), c.820-2A>G, c.892C>T (p.R298W), c.1064C>T (p.A355V), c.1090A>C (p.N364H), c.1150C>G (p.P384A), c.1184+1G>A, c.1309G>A (p.D437N), c.1469C>A (p.A490D), c.133_134insC (p.P45fs), c.710delA (p.D237fs), c.978delA (p.Q326fs), c.1138_1158delTCTGCCAACGATCCTATCTTC (p.S380_F386del) Sequencing | NM_000372:1-5

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

Oculocutaneous Albinism: Type 4 (SLC45A2): Mutations (2): d' Genotyping | c.469G>A (p.D157N), c.563G>T (p.G188V) Sequencing | NM_016180:1-7

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

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

Ornithine Translocase Deficiency (SLC25A15): Mutations (3): Or 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): of 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.T2511), 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): o^a 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^a Genotyping | c.1001+1G>A, c.1151A>G (p.E384G), c.1246A>C (p.T416P), c.2168A>G (p.H723R), c.707T>C (p.L236P), c.716T>A (p.V239D), c.919-2A>G Sequencing | NM_000441:1-21

Persistent Mullerian Duct Syndrome: Type I (AMH): Mutations (6): O' Genotyping | c.1144G>T (p.E382X), c.571C>T (p.R191X), c.1518C>G (p.H506Q), c.1574G>A (p.C525Y), c.17_18delTC, c.283C>T (p.R95X) Sequencing | NM_000479:1-4

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

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

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

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

Pontocerebellar Hypoplasia: RARS2 Related (RARS2): Mutations (3): O 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): of Genotyping | c.1001A>G (p.Y334C) Sequencing | NM_016955:1-11

Pontocerebellar Hypoplasia: TSEN54 Related (TSEN54): Mutations (3): O^a 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): & Genotyping | c.1072C>T (p.R358X), c.397C>T (p.R133C) Sequencing | NM_003384:2-13

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

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

Primary Ciliary Dyskinesia: DNAI1 Related (DNAI1): Mutations (5): of 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): of Genotyping | c.1494+1G>A, c.346-3T>G, c.787C>T (p.R263X), c.1304G>A (p.W435X) Sequencing | NM_023036:2-13

Primary Congenital Glaucoma (CYP1B1): Mutations (9): d^a Genotyping | c.1405C>T (p.R469W), c.1093G>T (p.G365W), c.155C>T (p.P52L), c.1064_1076delGAGTGCAGGCAGA (p.R355Hfs), c.1410_1422delCATTGGCGAAGAA (p.C470fs), c.862_863insC, c. 1199_1200insTCATGCCACC, c. 182G>A (p.G61E), c. 535delG (p.A 179fs) 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): of 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): of 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): of Genotyping | c.293A>G (p.D98G) Sequencing | NM_000055:2-4

Pycnodysostosis (CTSK): Mutations (2): 0^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): of Genotyping | c.395A>G (p.Y132C), c.1030C>T (p.P344S) Sequencing | NM_000925:1-10

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

Retinal Dystrophies: RLBP1 Related (RLBP1): Mutations (3): O' 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): of 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): O" 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): 3 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): or 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): o^a 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): O^a 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): of 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.2021G>A (p.R674H), c.889C>T (p.R297X), c.1928G>A (p.R643H), c.1927C>T (p.R643C), c.1562C>T (p.P521L), c.1444C>T (p.R482W), c.1693C>T (p.R565W), c.1694G>C (p.R565P), c.700C>T (p.R234C), c.1876C>T (p.R626X) Sequencing | NM_000263:2-6

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

Sanfilippo Syndrome: Type D (GNS): Mutations (5): d' Genotyping | c.1063C>T (p.R355X), c. 1168C>T (p.Q390X), c. 1226insG (p.R409fsX), c. 1138insGTCCT (p.D380fsX), c. 1169delA (p.Q390fsX) Sequencing | NM_002076:1-14

Short-Chain Acyl-CoA Dehydrogenase Deficiency (ACADS): Mutations (5): O" 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): d^{*} Genotyping | c.20A>T (p.E7V) Sequencing | NM 000518:1-3

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

Sly Syndrome (GUSB): Mutations (5): of 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): d' Genotyping | c.964-1G>C, c.356A>T (p.H119L), c.1054C>T (p.R352W), c.1210C>T (p.R404C), c.278C>T (p.T93M), c.1055G>A (p.R352Q), c.1139G>A (p.C380Y), c.1337G>A (p.R446Q), c.452G>A (p.W151X), c.453G>A (p.W151X), c.744G>T (p.W248C), c.976G>T (p.V326L), c.326T>C (p.L109P), c.470T>C (p.L157P), c.1342G>A (p.E448K), c.1228G>A (p.G410S), c.906C>G (p.F302L), c.725G>A (p.R242H), c.724C>T (p.R242C), c.506C>T (p.S169L), c.1A>G, c.670G>A (p.E224K), c.818T>G (p.V273G), c.203T>C (p.L68P), c.292C>T (p.Q98X), c.532A>T (p.1178F), c.545G>T (p.W182L), c.682C>T (p.R228W), c.575C>T (p.S192F), c.1295A>G (p.Y432C), c.1039G>A (p.G347S), c.1079T>C (p.L360P), c.1424T>C (p.F475S), c.1190C>T (p.S397L), c.1351T>C (p.C451R), c.853_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): O' Genotyping | DEL EXON 7, c.22_23insA, c.43C>T (p.Q15X), c.91_92insT, c.305G>A (p.W102X), c.400G>A (p.E134K), c.439_443delGAAGT, c.558delA, c.585_586insT, c.683T>A (p.L228X), c.734C>T (p.P245L), c.768_778dupTGCTGATGCTT, c.815A>G (p.Y272C), c.821C>T (p.T274I), c.823G>A (p.G275S), c.834+2T>G, c.835-18_835-12delCCTTTAT, c.835G>T, c.836G>T dPCR | DEL EXON 7

Stargardt Disease (ABCA4): Mutations (16): O^a 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_3211 insGT (p.S1071 Vfs), 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): of 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): O' Genotyping | c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.532C>T (p.R178C), c.533G>A (p.R178H), c.805G>A (p.G269S), c.1510C>T (p.R504C), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.1003A>T (p.I335F), c.910_912delTTC (p.305delF), c.749G>A (p.G250D), c.632T>C (p.F211S), c.629C>T (p.S210F), c.613delC, c.611A>G (p.H204R), c.598G>A (p.V200M), c.590A>C (p.K197T), c.571-1G>T, c.540C>G (p.Y180X), c.538T>C (p.Y180H), c.533G>T (p.R178L),

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c.508C>T (p.R170W), c.409C>T (p.R137X), c.380T>G (p.L127R), c.346+1G>C, c.116T>G (p.L39R), c.78G>A (p.W26X), c.1A>G (p.M1V), c.1495C>T (p.R499C), c.459+5G>A (IVS4+5G>A), c.1422-2A>G, c.535C>T (p.H179Y), c.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.E114K), c.1432G>A (p.G478R), c.1178G>C (p.R393P), c.805+1G>C, c.1426A>T (p.R476X), c.623A>T (p.D208V), c.1537C>T (p.Q513X), c.1511G>T (p.R504L), c.1307_1308delTA (p.I436fs), c.571-8A>G, c.624_627delTCCT (p.D208fs), c.1211_1212delTG (p.L404fs), c.621T>G (p.D207E), c.1511G>A (p.R504H), c.1177C>T (p.R393X), c.2T>C (p.M1T), c.1292G>A (p.W431X), c.947_948insA (p.Y316fs), c.607T>G (p.W203G), c.1061_1063delTCT (p.F354_Y355delinsX), c.615delG (p.L205fs), c.805+2T>C, c.1123delG (p.E375fs), c.1121A>G (p.Q374R), c.1043_1046delTCAA (p.F348fs), c.1510delC (p.R504fs), c.1451T>C (p.L484P), c.964G>T (p.D322Y), c.1351C>G (p.L451V), c.571-2A>G (IVS5-2A>G) Sequencing | NM_000520:1-14

Trichohepatoenteric Syndrome: Type 1 (TTC37): Mutations (9): of 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): d^a Genotyping | c.698G>A (p.R233H) Sequencing | NM_199292:1-14

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

Usher Syndrome: Type 1C (USH1C): Mutations (6): d^a Genotyping | c.496+1G>A, c.238_239insC, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.36+1G>T, c.496+1G>T Sequencing | NM 153676:1-27

Usher Syndrome: Type 1D (CDH23): Mutations (14): Or Genotyping | c. 172C>T (p.Q58X), c.3367C>T (p.Q1123X), c.3617C>G (p.P1206R), c.3713_3714delCT (p.S1238fs), c.3880C>T (p.Q1294X), c.4069C>T (p.Q1357X), c.4488G>C (p.Q1496H), c.4504C>T (p.R1502X), c.5237G>A (p.R1746Q), c.5985C>A (p.Y1995X), c.6307G>T (p.E2103X), c.7549A>G (p.S2517G), c.8230G>A (p.G2744S), c.8497C>G (p.R2833G) Sequencing | NM_022124:2-68 Usher Syndrome: Type 1F (PCDH15): Mutations (7): of Genotyping | c.733C>T (p.R245X), c.2067C>A (p.Y684X), c.7C>T (p.R3X), c.1942C>T (p.R648X), c.1101 delT (p.A367fsX), c.2800C>T (p.R934X), c.4272delA (p.L1425fs) Sequencing | NM_001142763:2-35

Usher Syndrome: Type 2A (USH2A): Mutations (23): of Genotyping | c.14020A>G (p.R4674G), c.12067-2A>G, c.4338_4339delCT (p.C1447fs), c.2299delG (p.E767SfsX21), c.2276G>T (p.C759F), c.2209C>T (p.R737X), c.1256G>T (p.C419F), c.1000C>T (p.R334W), c.923_924insGCCA (p.H308fs), c.12708T>A (p.C4236X), c.13576C>T (p.R4526X), c.1840+1G>A, c.11328T>G (p.Y3776X), c.5329C>T (p.R1777W), c.9165_9168delCTAT (p.I3055MfsX2), c.9469C>T (p.Q3157X), c.1876C>T (p.R626X), c.7123delG (p.G2375fs), c.9492_9498delTGATGAG (p.D3165fs), c.6235A>T (p.K2079X), c.14403C>G (p.Y4801X), c.3788G>A (p.W1263X), c.11328T>A (p.Y3776X) Sequencing | NM_206933:2-72

Usher Syndrome: Type 3 (CLRN1): Mutations (5): 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): 0" 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): d^a 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): O^{*} 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^a 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): of 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): d^a Genotyping | c.172+2T>G, c.323G>T (p.C108F), c.374delC (p.T125fs), c.682C>T (p.R228X), c.619C>T (p.R207X), c.348T>A (p.Y116X), c.390-1G>C Sequencing | NM_000380:1-6

Xeroderma Pigmentosum: Group C (XPC): Mutations (5): d' Genotyping | c. 1735C>T (p.R579X), c.566_567delAT (p.Y189fs), c.413-9T>A, c.413-24A>G, c.1643_1644delTG (p.V548fs) Sequencing | NM_004628:1-16

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

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

Zellweger Spectrum Disorders: PEX6 Related (PEX6): Mutations (8): O" 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.

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

CarrierMap[™]

			-
Disease	Carrier Rate	Detection Rate	Residuc Risk
Alkaptonuria	o [®] Dominican: Unknown	>99%	Unknow
	o [*] Finnish: 1/251	60.00%	1/628
	o [*] Slovak: 1/69	59.38%	1/170
Alpha Thalassemia	o" General: 1/48	50.67%	1/97
Alpha-1-Antitrypsin Deficiency	o" European: 1/35	95.00%	1/700
	o" General: Unknown	95.00%	Unknow
Alpha-Mannosidosis	o" European: 1/354	30.23%	1/507
	o" General: 1/354	35.19%	1/546
Alport Syndrome: COL4A3 Related	o [*] Dutch: 1/409	22.73%	1/529
Alport Syndrome: COL4A4 Related	o" General: 1/409	23.33%	1/533
Amegakaryocytic Thrombocytopenia	♂ [®] Ashkenazi Jewish: 1/76	>99%	<1/7,60
	o" General: Unknown	64.81%	Unknow
Andermann Syndrome	o [*] French Canadian: 1/24	99.38%	1/3,88
Antley-Bixler Syndrome	o ^r General: Unknown	45.65%	Unknow
	o ^r Japanese: Unknown	60.47%	Unknow
Argininemia	o ^r Chinese: Unknown	40.00%	Unknow
	o ^a French Canadian: Unknown	75.00%	Unknow
	o ⁷ Japanese: Unknown	>99%	Unknow
Argininosuccinate Lyase Deficiency	o" European: 1/133	57.41%	1/312
	o" Saudi Arabian: 1/80	51.72%	1/166
Aromatase Deficiency	o ^r General: Unknown	25.00%	Unknow
Arthrogryposis, Mental Retardation, & Seizures	o ^a Ashkenazi Jewish: 1/205	>99%	<1/20,5 0
Asparagine Synthetase Deficiency	o ^r Iranian Jewish: 1/80	>99%	<1/8,00
Aspartylglycosaminuria	ơ [°] Finnish: 1/69	96.12%	1/1,78
Ataxia with Vitamin E Deficiency	o [*] European: 1/274	80.00%	1/1,37
	o" Italian: 1/224	97.73%	1/9,85
	♂ North African: 1/159	>99%	<1/15,9 0
Ataxia-Telangiectasia	o" Costa Rican: 1/100	68.52%	1/318
	o ^a North African Jewish: 1/81	96.97%	1/2,67
	o ^a Norwegian: 1/197	50.00%	1/394
	o ^r Sardinians: Unknown	85.71%	Unknow
	o ^a US Amish: Unknown	>99%	Unknow
Autosomal Recessive Polycystic Kidney Disease	o" Finnish: 1/45	84.21%	1/285
	ð ^a French: 1/71	62.50%	1/189
	o" General: 1/71	37.11%	1/113
Bardet-Biedl Syndrome: BBS1 Related	o" General: 1/376	70.27%	1/1,26
	o'' Northern European: 1∕376	85.90%	1/2,66
	o ^a Puerto Rican: Unknown	90.00%	Unknow
Bardet-Biedl Syndrome: BBS10 Related	o" General: 1/404	47.79%	1/774
Bardet-Biedl Syndrome: BBS11 Related	o ^a Bedouin: 1/59	>99%	<1/5,90
Bardet-Biedl Syndrome: BBS12 Related	o ^r General: Unknown	50.00%	Unknow

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Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Bardet-Biedl Syndrome: BBS2 Related	O ^a Ashkenazi Jewish: Unknown	>99%	Unknown		♂ Moroccan Jewish: 1/234	>99%	<1/23,40 0
	ð General: 1/638	38.46%	1/1,037	Citrin Deficiency	♂ [*] Japanese: 1/70	>99%	<1/7,000
	o ^a Middle Eastern: Unknown	>99%	Unknown	Citrullinemia: Type I	o" European: 1/120	18.18%	1/147
Bare Lymphocyte Syndrome: Type II	o ^a General: Unknown	66.67%	Unknown		ð" General: 1/120	52.27%	1/251
Bartter Syndrome: Type 4A	ð General: 1/457	81.82%	1/2,514		o" Japanese: Unknown	64.71%	Unknown
Beta Thalassemia	o ^a African American: 1/75	84.21%	1/475		o ^r Mediterranean: 1/120	50.00%	1/240
	o" Indian: 1/24	74.12%	1/93	Classical Galactosemia	o ^a African American: 1/78	73.13%	1/290
	o" Sardinians: 1/23	97.14%	1/804		♂ Ashkenazi Jewish: 1/127	>99%	<1/12,70
	o" Spaniard: 1/51	93.10%	1/739			75 170/	0
Beta-Hexosaminidase Pseudodeficiency	♂ Ashkenazi Jewish: Unknown	>99%	Unknown		් Dutch: 1∕91 ඒ European: 1∕112	75.47% 88.33%	1/371 1/960
	o" General: Unknown	>99%	Unknown		o" General: 1/125	80.00%	1/625
Beta-Ketothiolase Deficiency	o ^a Japanese: Unknown	58.33%	Unknown		0" Irish: 1/76	91.30%	1/874
	o" Spaniard: Unknown	90.00%	Unknown		o" Irish Travellers: 1/14	>99%	<1/1,400
Biotinidase Deficiency	ð" General: 1/123	78.32%	1/567	Cockayne Syndrome: Type A	o" Christian Arab: Unknown	50.00%	Unknown
Bloom Syndrome	♂ Ashkenazi Jewish: 1/134	96.67%	1/4,020	Cockayne Syndrome: Type B	o" General: 1/378	19.30%	1/468
	o ^a European: Unknown	66.22%	Unknown	Cohen Syndrome	o" European: Unknown	19.05%	Unknown
	o ^a Japanese: Unknown	50.00%	Unknown		o" Finnish: 1/140	67.24%	1/427
Canavan Disease	o ^a Ashkenazi Jewish: 1/55	98.86%	1/4,840		♂ US Amish: 1/12	>99%	<1/1,200
	o ^a European: Unknown	53.23%	Unknown	Combined Pituitary Hormone	o" European: 1/45	93.29%	1/671
Carnitine Palmitoyltransferase IA Deficiency	o" General: Unknown	38.89%	Unknown	Deficiency: PROP1 Related	o" General: 1/45	82.35%	1/255
	o" Hutterite: 1/16	>99%	<1/1,600	Congenital Disorder of Glycosylation:	o ^a Danish: 1/71	90.00%	1/710
	0 [°] Japanese: 1/101	66.67%	1/303	Type 1A: PMM2 Related			
Carnitine Palmitoyltransferase II	o" Ashkenazi Jewish:	>99%	Unknown		o' Dutch: 1/68	39.29%	1/112
Deficiency	Unknown				o' European: 1/71	55.33%	1/159
	o" General: Unknown	71.43%	Unknown	Congenital Disorder of Glycosylation: Type 1B: MPI Related	o' French: Unknown	54.17%	Unknown
Carnitine-Acylcarnitine Translocase Deficiency	o ^r Asian: Unknown	95.45%	Unknown	Congenital Disorder of Glycosylation: Type 1C: ALG6 Related	o" French: Unknown	59.09%	Unknown
	o ^a General: Unknown	18.75%	Unknown		o ^a General: Unknown	86.21%	Unknown
Carpenter Syndrome	o ^a Brazilian: Unknown	40.00%	Unknown	Congenital Ichthyosis: ABCA12 Related		>99%	Unknown
	o" Northern European: Unknown	85.00%	Unknown	Congenitari (niniyosis, ADCA 12 Kelalea	o [®] South Asian: Unknown	66.67%	Unknown
Cartilage-Hair Hypoplasia	o' Finnish: 1/76	93.33%	1/1,140	Congenital Insensitivity to Pain with	o [®] Japanese: Unknown	56.52%	Unknown
	o" US Amish: 1/19	>99%	<1/1,900	Anhidrosis			
Cerebrotendinous Xanthomatosis	o" Dutch: Unknown	78.57%	Unknown		o ^a Moroccan Jewish: Unknown	>99%	Unknown
	o" Italian: Unknown	45.95%	Unknown	Congenital Lipoid Adrenal Hyperplasia	0 [°] Japanese: 1/201	51.11%	1/411
	o ^r Japanese: Unknown	92.86%	Unknown	- J. F	o " Korean: 1/251	63.64%	, 1/690
	o" Moroccan Jewish: 1/6	87.50%	1/48	Congenital Myasthenic Syndrome:	o [*] European Gypsy: 1/26	>99%	<1/2,600
Chediak-Higashi Syndrome	o" General: Unknown	19.64%	Unknown	CHRNE Related			, ,
Cholesteryl Ester Storage Disease	o" General: 1/101	68.97%	1/325		o" North African: Unknown	60.87%	Unknown
Choreoacanthocytosis	ơ" Ashkenazi Jewish: Unknown	66.67%	Unknown	Congenital Myasthenic Syndrome: DOK7 Related	o" European: 1/472	19.05%	1/583
Chronic Granulomatous Disease:	o ^a Iranian: Unknown	71.43%	Unknown		o" General: 1/472	18.75%	1/581
CYBA Related	0 [*] Japanese: 1/274	>99%	<1/27,40	Congenital Myasthenic Syndrome: RAPSN Related	o" General: 1/437	88.57%	1/3,824
	o" Korean: 1/105	>99%	0 <1/10,50 0		O ^a Non-Ashkenazi Jewish: Unknown	>99%	Unknown

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Congenital Neutropenia: Recessive	o ^a English: Unknown	11.76%	Unknown		o" Saudi Arabian: 1/38	>99%	<1/3,800
	o ^r Japanese: Unknown	22.22%	Unknown	Familial Dysautonomia	♂ Ashkenazi Jewish: 1/31	>99%	<1/3,100
	o ^r Turkish: Unknown	89.47%	Unknown	Familial Hyperinsulinism: Type 1:	o" Ashkenazi Jewish: 1/52	98.75%	1/4,160
Corneal Dystrophy and Perceptive Deafness	o' General: Unknown	71.43%	Unknown	ABCC8 Related	o" Finnish: 1/101	45.16%	1/184
Corticosterone Methyloxidase Deficiency	o" Iranian Jewish: 1/32	>99%	<1/3,200	Familial Hyperinsulinism: Type 2: KCNJ 11 Related	o" Arab: Unknown	40.00%	Unknown
Crigler-Najjar Syndrome	o" Sardinians: Unknown	80.00%	Unknown	Familial Mediterranean Fever	o" Arab: 1/4	51.18%	1/8
	o ^r Tunisian: Unknown	>99%	Unknown		o" Armenian: 1/5	94.51%	1/91
Cystic Fibrosis	o" African American: 1/62	69.99%	1/207		o" Ashkenazi Jewish: 1/81	39.52%	1/134
	o" Ashkenazi Jewish: 1/23	96.81%	1/721		o" Iraqi Jewish: 1/4	76.92%	1/17
	0 [°] Asian: 1/94	65.42%	1/272		o" Israeli Jewish: 1/5	62.26%	1/13
	o [*] European: 1/25	94.96%	1/496		o" Lebanese: 1/6	91.67%	1/72
	o" Hispanic American: 1/48	77.32%	1/212		o" North African Jewish: 1/5	95.69%	1/116
	o ^r Native American: 1/53	84.34%	1/338		o ^r Syrian: 1/6	85.14%	1/40
Cystinosis	o [®] Dutch: 1/194	73.08%	1/721		o [*] Turkish: 1/5	74.25%	1/19
	o" French Canadian: 1/40	75.00%	1/160	Fanconi Anemia: Type A	♂ Moroccan Jewish: 1/100	>99%	<1/10,00 0
	o' General: 1/194	54.51%	1/426		o ^a Spanish Gypsy: 1/67	>99%	<1/6,700
Cystinuria: Non-Type I	o" European: 1/42	61.11%	1/108	Fanconi Anemia: Type C	o ^a Ashkenazi Jewish: 1/101	>99%	<1/10,10
	o' General: 1/42	37.50%	1/67	Tuncolii Aneiniu. Type C	O Asilkendzi Jewisii. 17 101	27770	0
	ơ ^a Libyan Jewish: 1/26	93.48%	1/399		o' General: Unknown	30.00%	Unknown
	o" United States: 1/42	56.25%	1/96	Fanconi Anemia: Type G	o [*] Black South African:	81.82%	1/556
Cystinuria: Type I	o" European: 1/42	46.67%	1/79		1/101		
	o [*] Swedish: 1/159	55.88%	1/360		ơ' French Canadian: Unknown	87.50%	Unknown
D-Bifunctional Protein Deficiency	o'' General: 1/159	38.64%	1/259		o ^r Japanese: Unknown	75.00%	Unknown
Diabetes: Recessive Permanent Neonatal	o' General: Unknown	25.00%	Unknown		o ^r Korean: Unknown	66.67%	Unknown
Du Pan Syndrome	o ^a Pakistani: Unknown	>99%	Unknown	Fanconi Anemia: Type J	o ^r General: Unknown	86.36%	Unknown
Dyskeratosis Congenita: RTEL1 Related	o" Ashkenazi Jewish: 1/203	>99%	<1/20,30	Fumarase Deficiency	o ^r General: Unknown	30.00%	Unknown
	o ^a General: 1/501	50.00%	0	GM1-Gangliosidoses	o" Eurodescent Brazilian: 1/66	62.15%	1/174
Dystrophic Epidermolysis Bullosa:	o" Italian: Unknown	45.00%	Unknown		o" European: 1/194	50.00%	1/388
Recessive		10.0076	0		o" General: 1/194	20.00%	1/243
	o ^a Mexican American: 1/345	56.25%	1/789		♂ [®] Hispanic American: 1∕194	58.33%	1/466
Ehlers-Danlos Syndrome: Type VIIC	o ^a Ashkenazi Jewish: Unknown	>99%	Unknown		o ^r Japanese: Unknown	62.82%	Unknown
Ellis-van Creveld Syndrome: EVC	олкпоwn o ^a General: 1/123	32.14%	1/181	GRACILE Syndrome	o" Finnish: 1/109	97.22%	1/3,924
Related	O General: 1/123	32.14%	1/101	Galactokinase Deficiency	o" Japanese: 1/501	50.00%	1/1,002
Ellis-van Creveld Syndrome: EVC2 Related	o ^r General: Unknown	<10%	Unknown		0 ⁷ Roma: 1/51	>99%	<1/5,100
Enhanced S-Cone	o" Ashkenazi Jewish:	90.48%	Unknown	Gaucher Disease	♂ Ashkenazi Jewish: 1/15	87.16%	1/117
	Unknown	/0.40%	UNKNOWN		o" General: 1/112	31.60%	1/164
	o' General: Unknown	52.50%	Unknown		o" Spaniard: Unknown	44.29%	Unknown
Ethylmalonic Aciduria	o" Arab/Mediterranean:	29.17%	Unknown		o ^a Turkish: 1/236	59.38%	1/581
	Unknown			Gitelman Syndrome	♂ European: 1/100	35.00%	1/154
	o' General: Unknown	38.24%	Unknown		o ^a European Gypsy: Unknown	>99%	Unknown
Familial Chloride Diarrhea	o [*] Finnish: 1/51	>99%	<1/5,100		o" General: 1/101	30.00%	1/144
	o" Kuwaiti: 1/38	90.00%	1/380		o ^r Taiwanese: Unknown	64.29%	Unknown
	o [*] Polish: 1/224	45.24%	1/409		C Idimenese. Officiowi	J-T. 2 7 /0	CIRIOWI

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
Globoid Cell Leukodystrophy	o ^a Dutch: 1/137	60.98%	1/351	Hemochromatosis: Type 2A: HFE2	o ^r European: Unknown	69.23%	Unknown
	o" European: 1/150	26.47%	1/204	Related	-71.4.1.	70 700/	
	o [*] Japanese: 1/150	36.00%	1/234		o [*] Mediterranean: Unknown	72.73%	Unknown
Glutaric Acidemia: Type I	o [™] European: 1/164	57.78%	1/388	Hemochromatosis: Type 3: TFR2 Related	o" Italian: Unknown	73.21%	Unknown
	o" General: 1/164	25.51%	1/220	Hemoglobinopathy: Hb C	o ^a African American: 1/51	>99%	<1/5,100
	♂ US Amish: 1/12	>99%	<1/1,200	Hemoglobinopathy: Hb D	o ^a Canadian: 1/64	>99%	<1/6,400
Glutaric Acidemia: Type IIA	o" General: Unknown	71.43%	Unknown		o ^a Indian: 1/16	>99%	<1/1,600
Glutaric Acidemia: Type IIB	o" General: Unknown	33.33%	Unknown		o" Iranian: 1/11	>99%	<1/1,100
Glutaric Acidemia: Type IIC	o ^r Taiwanese: Unknown	>99%	Unknown	Hemoglobinopathy: Hb E	o ^r Cambodia: 1/4	>99%	<1/400
	o ^a Turkish: Unknown	80.00%	Unknown		o ^r Chinese: 1/13	>99%	, <1/1,300
Glycine Encephalopathy: AMT Related	o' General: Unknown	40.91%	Unknown		o ^a Indian: 1/10	>99%	<1/1,000
Glycine Encephalopathy: GLDC Related	o ^a Finnish: 1/118	78.00%	1/536		0" Thai: 1/9	>99%	<1/900
	o" General: 1/280	12.50%	1/320	Hemoglobinopathy: Hb O	o [*] African American: 1/87	>99%	<1/8,700
Glycogen Storage Disease: Type IA	o ^a Ashkenazi Jewish: 1/71	>99%	<1/7,100		o ^r Middle Eastern: Unknown	>99%	Unknown
	o [®] Chinese: 1/159	80.00%	1/795	Hereditary Fructose Intolerance	o" European: 1/81	72.73%	1/297
	o [®] European: 1/177	76.88%	1/765		o" Italian: 1/81	90.91%	1/891
	o [®] Hispanic American:	27.78%	1/245		o" Slavic: 1/81	>99%	<1/8,100
	1/177			Hereditary Spastic Paraplegia: TECPR2 Related	o" Bukharan Jewish: 1/75	>99%	<1/7,500
Chucagan Storaga Diragra: Tupa IB	0 ⁷ Japanese: 1/177	89.22%	1/1,641	Herlitz Junctional Epidermolysis	o ^a Pakistani: Unknown	>99%	Unknown
Glycogen Storage Disease: Type IB	o [*] Australian: 1/354	50.00%	1/708	Bullosa: LAMA3 Related			
	Ø [*] European: 1/354	45.74%	1/652	Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	o ^r European: Unknown	70.00%	Unknown
	o ⁷ Japanese: 1/354	39.13%	1/582		o" General: 1/781	52.27%	1/1,636
Glycogen Storage Disease: Type II	o ^a African American: 1/60 o ^a Chinese: 1/112	45.83% 72.00%	1/111 1/400	Herlitz Junctional Epidermolysis	o" Italian: Unknown	28.57%	Unknown
	o" European: 1/97	51.76%	1/201	Bullosa: LAMC2 Related			
	o [®] North African: Unknown	60.00%	Unknown	Hermansky-Pudlak Syndrome: Type 1	o" Puerto Rican: 1/22	94.95%	1/436
Glycogen Storage Disease: Type III	of Faroese: 1/30	>99%	<1/3,000	Hermansky-Pudlak Syndrome: Type 3	o" Ashkenazi Jewish: 1/235	>99%	<1/23,50
Glycogen Slorage Disease. Type III			1/264				0
	o ^a General: 1/159 o ^a North African Jewish:	39.81%			o [*] European: 1/434	12.50%	1/496
	1/35	>99%	<1/3,500	Hermansky-Pudlak Syndrome: Type 4	o" European: Unknown	54.17%	Unknown
Glycogen Storage Disease: Type IV	o ^a Ashkenazi Jewish: 1/35	>99%	<1/3,500	Holocarboxylase Synthetase Deficiency	o" European: 1/148	83.33%	1/888
	o' General: 1/461	18.60%	1/566		♂ Japanese: 1/159	76.92%	1/689
Glycogen Storage Disease: Type V	o" Caucasus Jewish: Unknown	>99%	Unknown	Homocystinuria Caused by CBS Deficiency	o" European: 1/224	64.29%	1/627
	o" European: 1/159	60.71%	1/405		o" Irish: 1/128	70.59%	1/435
	o" General: Unknown	74.10%	Unknown		o" Italian: 1/224	35.71%	1/348
	♂ Spaniard: 1/159	67.11%	1/483		o ^r Norwegian: 1/41	84.38%	1/262
	o ^r Yemenite Jewish: Unknown	75.00%	Unknown		o" Qatari: 1/22	>99%	<1/2,200
Glycogen Storage Disease: Type VII	o" Ashkenazi Jewish: 1/250	>99%	<1/25,00 0		o" Saudi Arabian: Unknown	92.31%	Unknown
Guanidinoacetate Methyltransferase Deficiency	o" General: Unknown	29.41%	Unknown	Hurler Syndrome	o" Czech: 1/190 o" European: 1/194	52.50% 81.71%	1/400 1/1,061
HMG-CoA Lyase Deficiency	o™ General: 1/159	40.00%	1/265		o ^r General: 1/194	62.50%	1/517
Time-con Lyuse Deliciency	o [®] Japanese: Unknown	30.00%	Unknown		o" Italian: 1/194	61.11%	1/499
			Unknown Unknown		o" Japanese: 1/194	23.68%	1/254
	of Portuguese: Unknown	86.36%			o [®] Moroccan Jewish: 1/194	92.31%	1/2,522
	o" Saudi Arabian: Unknown	93.33%	Unknown		o [®] Scandinavian: 1/194	79.41%	1/942
						/ 7.41/0	1/742

Disease	Carrier Rate	Detection Rate	Residual Risk	Disease	Carrier Rate	Detection Rate	Residual Risk
	o [*] Spaniard: 1/194	52.50%	1/408	Limb-Girdle Muscular Dystrophy: Type	o" Brazilian: Unknown	57.14%	Unknown
Hypophosphatasia	o [®] Canadian Amish: 1/26	>99%	<1/2,600	2E	-1 - 1 (500	05 000	1 /710
	o" European: 1/159	19.23%	1/197		o [*] European: 1/539	25.00%	1/719
	o ^a Japanese: Unknown	54.55%	Unknown		o' General: Unknown	12.50%	Unknown
Inclusion Body Myopathy: Type 2	o ^a General: Unknown	85.83%	Unknown		o" US Amish: Unknown	>99%	Unknown
	o" Iranian Jewish: 1/16	>99%	<1/1,600	Limb-Girdle Muscular Dystrophy: Type 2F	o" Brazilian: Unknown	>99%	Unknown
	o ^r Japanese: Unknown	71.88%	Unknown		o ^r General: Unknown	83.33%	Unknown
	o ^r Korean: Unknown	72.50%	Unknown	Limb-Girdle Muscular Dystrophy: Type	o ^r Brazilian: Unknown	34.62%	Unknown
Infantile Cerebral and Cerebellar Atrophy	o [™] Caucasus Jewish: 1/20	>99%	<1/2,000	21	ơ ^a Danish: 1/100	85.53%	1/691
Isolated Microphthalmia: VSX2 Related	o ^a Middle Eastern: Unknown	71.43%	Unknown		o" General: Unknown	43.18%	Unknown
Isovaleric Acidemia	o' General: 1/251	47.37%	1/477		o [*] German: 1/300	43.18% 82.50%	1/1,714
Joubert Syndrome	o ^a Ashkenazi Jewish: 1/92	>99%	<1/9,200	Lipoprotein Lipase Deficiency	o [®] French Canadian: 1/44	28.95%	1/62
Lamellar Ichthyosis: Type 1	o [*] Norwegian: 1/151	81.40%	1/812		o [®] General: Unknown		Unknown
Laryngoonychocutaneous Syndrome	o [®] Pakistani: Unknown	>99%	Unknown	Long-Chain 3-Hydroxyacyl-CoA		20.00% 88.98%	
, , , ,	ơ' European: 1/251	47.32%	1/476	Dehydrogenase Deficiency	o ^a European: 1/126		1/1,144
	o ^r Finnish: Unknown	>99%	Unknown		ơ' General: 1/126	56.25%	1/288
Related				Lysinuric Protein Intolerance	♂ Finnish: 1/123	>99%	<1/12,30 0
Leber Congenital Amaurosis: LCA5 Related	o ^a Pakistani: Unknown	83.33%	Unknown		o [*] Italian: 1/120	45.45%	1/220
Leber Congenital Amaurosis: RDH 12	o' General: 1/560	38.37%	1/909		o [*] Japanese: 1/115	37.93%	1/185
Related	,		,		o ^r North African: Unknown	>99%	Unknown
Leigh Syndrome: French-Canadian	o [*] French Canadian: 1/23	95.45%	1/506	MTHFR Deficiency: Severe	o [*] Bukharan Jewish: 1/39	>99%	<1/3,900
Leukoencephalopathy with Vanishing White Matter: EIF2B5 Related	o'' Cree: Unknown	>99%	Unknown	Malonyl-CoA Decarboxylase Deficiency	o" General: Unknown	33.33%	Unknown
	o ^a European: Unknown	65.22%	Unknown	Maple Syrup Urine Disease: Type 1A	o" US Amish: 1/10	97.73%	1/440
·/· · · · · · · · · · · · · · · · · · ·	o ^a Brazilian: Unknown	>99%	Unknown	Maple Syrup Urine Disease: Type 1B	o" Ashkenazi Jewish: 1/97	>99%	<1/9,700
Hormone Resistance)				Maple Syrup Urine Disease: Type 2	o' General: 1/481	42.31%	1/834
Limb-Girdle Muscular Dystrophy: Type 2A	o ^a Basque: 1/61	61.46%	1/158		o' Norwegian: 1/481	50.00%	1/962
	o" Croatian: 1/133	76.00%	1/554		d' Turkish: 1/112	58.33%	1/269
	o" European: 1/103	17.23%	1/124	Maple Syrup Urine Disease: Type 3	o" Ashkenazi Jewish: 1/94	>99%	<1/9,400
	o" General: 1/103	26.47%	1/140		o' General: Unknown	68.75%	Unknown
	o" Italian: 1/162	35.71%	1/252	Maroteaux-Lamy Syndrome	o [*] Argentinian: 1/274	75.00%	1/1,096
	o [®] Russian: 1/103	53.33%	1/221		o' General: 1/388	61.54%	1/1,009
	o" US Amish: Unknown	>99%	, Unknown		o [*] Spaniard: 1/274	29.17%	1/387
	o [®] Caucasus Jewish: 1/25	>99%	<1/2,500	Meckel Syndrome: Type 1	o [*] European: 1/212	72.22%	1/763
2B	· · · · · · · · · · · · · · · · · · ·		, ,		ơ Finnish: 1/48	>99%	<1/4,800
	o" Libyan Jewish: 1/19	>99%	<1/1,900	Medium-Chain Acyl-CoA	o [*] European: 1/50	90.91%	1/550
Limb-Girdle Muscular Dystrophy: Type 2C	o ^a European Gypsy: 1/50	>99%	<1/5,000	Dehydrogenase Deficiency	♂ Saudi Arabian: 1/68	95.00%	1/1,360
	o ^r General: Unknown	60.00%	Unknown		o [®] United Kingdom: 1/51	90.00%	1/510
	o ^r Tunisian: Unknown	>99%	Unknown	Megalencephalic	o" Japanese: Unknown	50.00%	Unknown
	o" Brazilian: Unknown	64.29%	Unknown	Leukoencephalopathy	·		
	C Europage 1/200	າງ າງ∾∕	1/270		o" Libyan Jewish: 1/40	>99%	<1/4,000
	o ^a European: 1/288	22.22%	1/370		o" Turkish: Unknown	20.00%	Unknown
	o ^a Finnish: 1/150	95.45%	1/3,300	Metachromatic Leukodystrophy	o [*] European: 1/150	43.88%	1/267
	o'' General: Unknown	26.09%	Unknown		o [*] Habbanite Jewish: 1/5	50.00%	1/10

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

Disease	Carrier Rate	Detection Rate	Residual Risk
Neuronal Ceroid-Lipofuscinosis: CLN5 Related	o" Finnish: 1/101	>99%	<1/10,10 0
Neuronal Ceroid-Lipofuscinosis: CLN6 Related	o" European: 1/159	36.36%	1/250
	o" General: 1/159	59.52%	1/393
	o [™] Portuguese: 1/128	81.00%	1/674
Neuronal Ceroid-Lipofuscinosis: CLN8 Related	o" Finnish: 1/135	>99%	<1/13,50 0
	o" Italian: 1/212	33.33%	1/318
	o" Turkish: Unknown	77.78%	Unknown
Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	o' General: 1/159	56.25%	1/363
Neuronal Ceroid-Lipofuscinosis: PPT1 Related	o" Finnish: 1/58	97.62%	1/2,436
	o" General: 1/159	72.50%	1/578
Neuronal Ceroid-Lipofuscinosis: TPP1 Related	o" Canadian: 1/159	67.50%	1/489
	♂ European: 1/159	75.00%	1/636
	o' General: 1/159	50.00%	1/318
	o" Newfoundlander: 1/43	85.29%	1/292
Niemann-Pick Disease: Type A	o" Ashkenazi Jewish: 1/101	95.00%	1/2,020
Niemann-Pick Disease: Type B	o [*] Czech: 1/276	83.33%	1/1,656
	o' General: Unknown	19.82%	Unknown
	o [®] North African: Unknown	86.67%	Unknown
	o ^r Spaniard: Unknown	38.10%	Unknown
Niemann-Pick Disease: Type C1	o'' Acadian: Unknown	>99%	Unknown
	o' General: 1/194	15.60%	1/230
	o ^r Japanese: Unknown	18.18%	Unknown
	o [®] Portuguese: 1/194	25.00%	1/259
Niemann-Pick Disease: Type C2	o" General: 1/194	75.00%	1/776
Nijmegen Breakage Syndrome	0 ⁷ Eastern European: 1/155	>99%	<1/15,50 0
Nonsyndromic Hearing Loss and Deafness: GJB2 Related	0° Ashkenazi Jewish: 1/20	95.83%	1/480
	o" Chinese: 1/100	82.26%	1/564
	o" European: 1/53	82.47%	1/302
	o" Ghanaian: Unknown	90.91%	Unknown
	o" Indian: Unknown	66.98%	Unknown
	o" Israeli: 1/16	93.10%	1/232
	O ^r Japanese: 1/75	75.00%	1/300
	o" Roma: Unknown	>99%	Unknown
	o [*] United States: 1/34	45.22%	1/62
Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related	♂ Ashkenazi Jewish: 1/180	>99%	<1/18,00 0
Nonsyndromic Hearing Loss and Deafness: MYO15A Related	0" Balinese: 1/6	>99%	<1/600
	0 [°] Pakistani: 1/77	24.00%	1/101
Oculocutaneous Albinism: Type 1	o" European: 1/101	26.32%	1/137
	o [*] Hutterite: 1/7	>99%	<1/700
	o [®] Moroccan Jewish: 1/30	71.88%	1/107

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

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Disease	Carrier Rate	Detection Rate	Residual Risk
	o" Italian: Unknown	27.78%	Unknown
	o" Norwegian: 1/142	47.92%	1/273
	o ^a Sardinians: 1/61	81.82%	1/336
	o ⁿ United Kingdom: Unknown	70.00%	Unknown
	o ^r United States: Unknown	65.62%	Unknown
Pontocerebellar Hypoplasia: EXOSC3 Related	o'' General: Unknown	83.33%	Unknown
Pontocerebellar Hypoplasia: RARS2 Related	♂ Sephardic Jewish: Unknown	>99%	Unknown
Pontocerebellar Hypoplasia: SEPSECS Related	0" Iraqi Jewish: 1/42	>99%	<1/4,200
Pontocerebellar Hypoplasia: TSEN54 Related	ơ¹ European: 1∕250	95.65%	1/5,750
Pontocerebellar Hypoplasia: VPS53 Related	o [®] Moroccan Jewish: 1/37	>99%	<1/3,700
Pontocerebellar Hypoplasia: VRK 1 Related	o" Ashkenazi Jewish: 1/225	>99%	<1/22,50 0
Primary Carnitine Deficiency	o" European: 1/101	58.33%	1/242
	o [*] Faroese: 1/9	53.95%	1/20
	o' General: Unknown	20.22%	Unknown
Primary Ciliary Dyskinesia: DNA11 Related	o" European: 1/211	52.38%	1/443
Primary Ciliary Dyskinesia: DNAI2 Related	♂ Ashkenazi Jewish: 1/200	>99%	<1/20,00 0
Primary Congenital Glaucoma	o" Moroccan: Unknown	>99%	Unknown
	o" Saudi Arabian: 1/23	91.67%	1/276
	o ^a Turkish: 1/51	70.59%	1/173
Primary Hyperoxaluria: Type 1	o'' Dutch: 1/174	62.12%	1/459
	o" General: 1/189	52.68%	1/399
Primary Hyperoxaluria: Type 2	o' General: Unknown	70.31%	Unknown
Primary Hyperoxaluria: Type 3	o" Ashkenazi Jewish: Unknown	>99%	Unknown
	o" European: Unknown	25.00%	Unknown
Progressive Familial Intrahepatic Cholestasis: Type 2	0 ⁷ European: Unknown	33.33%	Unknown
Propionic Acidemia: PCCA Related	o ^a Japanese: 1/102	86.67%	1/765
Propionic Acidemia: PCCB Related	o" General: 1/182	42.86%	1/319
	o" Greenlandic Inuit: 1/16	58.33%	1/38
	o ^r Japanese: 1/102	78.00%	1/464
	o'' Korean: Unknown	56.25%	Unknown
	o" Latin American: 1/182	75.00%	1/728
	o" Spaniard: 1/182	52.38%	1/382
Pseudocholinesterase Deficiency	o" General: 1/33	65.00%	1/94
	o" Iranian Jewish: 1/9	>99%	<1/900
Pycnodysostosis	o" Danish: Unknown	87.50%	Unknown
Pyruvate Carboxylase Deficiency	o" General: 1/251	62.50%	1/669
	o" Native American: 1/10	>99%	<1/1,000
Pyruvate Dehydrogenase Deficiency	o' General: Unknown	50.00%	Unknown

CarrierMap™

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

Disease	Carrier Rate	Detection Rate	Residual Risk
	o" General: Unknown	75.00%	Unknown
Sulfate Transporter-Related Osteochondrodysplasia	o" Finnish: 1/51	95.83%	1/1,224
	o' General: 1/100	70.00%	1/333
Tay-Sachs Disease	o" Argentinian: 1/280	82.35%	1/1,587
	o" Ashkenazi Jewish: 1/29	99.53%	1/6,177
	o" Cajun: 1/30	>99%	<1/3,000
	o" European: 1/280	25.35%	1/375
	o' General: 1/280	32.09%	1/412
	o" Indian: Unknown	85.71%	Unknown
	o" Iraqi Jewish: 1/140	56.25%	1/320
	o [*] Japanese: 1/127	82.81%	1/739
	o [*] Moroccan Jewish: 1/110	22.22%	1/141
	o [*] Portuguese: 1/280	92.31%	1/3,640
	o" Spaniard: 1/280	67.65%	1/865
	o" United Kingdom: 1/161	71.43%	1/564
Trichohepatoenteric Syndrome: Type 1	o" European: 1/434	42.86%	1/760
	♂ South Asian: 1/434	66.67%	1/1,302
Tyrosine Hydroxylase Deficiency	o" General: Unknown	36.11%	Unknown
Tyrosinemia: Type I	♂ Ashkenazi Jewish: 1/158	>99%	<1/15,80 0
	o" European: 1/166	57.14%	1/387
	0" Finnish: 1/123	97.22%	1/4,428
	o ^r French Canadian: 1/64	96.30%	1/1,728
	o ^r Pakistani: Unknown	92.86%	Unknown
Tyrosinemia: Type II	o' General: 1/251	40.00%	1/418
Usher Syndrome: Type 1B	o" European: 1/166	39.29%	1/273
	o" General: 1/143	12.89%	1/164
	o" North African: Unknown	66.67%	Unknown
	o" Spaniard: 1/152	12.16%	1/173
Usher Syndrome: Type 1C	♂ Acadian: 1/82	98.86%	1/7,216
	o" French Canadian: 1/227	83.33%	1/1,362
Usher Syndrome: Type 1D	♂ General: 1/296	23.17%	1/385
Usher Syndrome: Type 1F	♂ Ashkenazi Jewish: 1/126	93.75%	1/2,016
Usher Syndrome: Type 2A	o ^r Chinese: Unknown	83.33%	Unknown
	o" European: 1/136	40.00%	1/227
	o ^a French Canadian: Unknown	66.67%	Unknown
	o" General: 1/136	47.69%	1/260
	o" Japanese: Unknown	55.56%	Unknown
	o ^a Non-Ashkenazi Jewish: Unknown	61.11%	Unknown
	o [*] Scandinavian: 1/125	40.52%	1/210
	o" Spaniard: 1/133	53.66%	1/287
Usher Syndrome: Type 3	o" Ashkenazi Jewish: 1/120	>99%	<1/12,00 0
	ơ" Finnish: 1/134	>99%	<1/13,40 0

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