

4978 Santa Anita Ave.
Temple City, CA 91780
(p) 626-350-0537 (f) 626-454-1667
info@fulgentgenetics.com
www.fulgentgenetics.com



Patient Information:



Partner Information:

Not Tested

Physician:

Galea, Melanie
Sonic Genetics
14 Giffnock Avenue
Macquarie Park, NSW 2113 AU
Phone: +02 9855 5369

Laboratory:

Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Jun 21, 2023**

Accession:

FT-5981100
Test#: FT-TS14588148
Specimen Type: Saliva Swab
Collected: May 02, 2023

Accession:

N/A

FINAL RESULTS

TEST PERFORMED



Carrier for **ONE** genetic condition
Genetic counseling is
recommended.

**Sonic Beacon Expanded
Carrier Screen v2.0 - Male**
(361 Gene Panel; gene sequencing
with deletion and duplication analysis)

Condition and Gene

Inheritance



Partner

Leber congenital amaurosis type 13
RDH12

AR

⊕ Carrier

N/A

c.601del (p.Cys201Alafs*77)

INTERPRETATION:


Notes and Recommendations:

- Based on these results, this individual is positive for a carrier mutation in 1 gene. The risk estimates below are quantified based on general population carrier frequencies. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions:
 - There is a 1/2000 chance of having a child affected with Leber congenital amaurosis type 13, a *RDH12*-related condition.
- Testing for copy number changes in the *SMN1* gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- This report does not include variants of uncertain significance.
- Genetic counseling is recommended. Contact your physician about the available options for genetic counseling.





LEBER CONGENITAL AMAUROSIS TYPE 13

Patient		Partner
Result	+ Carrier	N/A
Variant Details	RDH12 (NM_152443.3) c.601del (p.Cys201Alafs*77)	N/A

What is Leber congenital amaurosis type 13?

Leber congenital amaurosis is an eye disorder that primarily affects the retina, which is the specialized tissue at the back of the eye that detects light and color. People with this disorder typically have severe visual impairment beginning in infancy.

What is my risk of having an affected child?

Leber congenital amaurosis type 13 is inherited in an autosomal recessive manner. The risk for being a carrier for Leber congenital amaurosis type 13 is very low (carrier frequency less than 1/500). Individuals of Caucasian/European descent have an increased carrier risk of 1/456. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

There are no approved treatments for Leber congenital amaurosis, although this is an area of active research and clinical trials. Lifelong monitoring by an ophthalmologist is recommended.

What mutation was detected?

The detected heterozygous variant was NM_152443.3:c.601del (p.Cys201Alafs*77). This frameshift variant is predicted to introduce a premature stop codon and result in a truncated protein. This variant is expected to disrupt the last 116 (~37%) amino acids of the original protein. The truncated or altered region is critical to protein function as there are multiple pathogenic variants downstream of this position (PubMed: [20736127](#), [16269441](#), [24123792](#)). This variant was observed as heterozygous with c.437T>A (p.V146D) (LP) in one patient with Leber Congenital Amaurosis (PubMed: [26047050](#)). The laboratory classifies this variant as likely pathogenic.





GENES TESTED:

Sonic Beacon Expanded Carrier Screen v2.0 - Male - 361 Genes

This analysis was run using the Sonic Beacon Expanded Carrier Screen v2.0 - Male gene list consisting of 361 genes (v2, effective November 1st 2022). 361 genes were tested with 99.5% of targets sequenced at >20x coverage. For more gene specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

ABCA12, ABCA3, ABCA4, ABCB11, ABCC8, ACAD9, ACADVL, ACAT1, ACOX1, ACSF3, ADA, ADAMTS2, ADGRG1, ADK, AGA, AGL, AGPS, AGXT, AHI1, AIPL1, ALDH3A2, ALDOB, ALG6, ALMS1, ALPL, AMT, AQP2, ARG1, ARL13B, ARSA, ARSB, ASL, ASNS, ASPA, ASS1, ATM, ATP6V1B1, ATP7B, BBS1, BBS10, BBS12, BBS2, BCKDHA, BCKDHB, BCS1L, BLM, BSND, CAPN3, CASQ2, CBS, CC2D2A, CCDC103, CCDC39, CCDC88C, CDH23, CEP290, CFTR, CHRNE, CHRNA, CHST6, CIITA, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL27A1, COL4A3, COL4A4, COL7A1, COX15, CPS1, CPT1A, CPT2, CRB1, CRYL1, CTNS, CTSA, CTSC, CTSD, CTSK, CYBA, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP17B1, CYP21A2, CYP27A1, DBT, DCLRE1C, DDX11, DHCR7, DHDDS, DLD, DNAH5, DNAI1, DNAI2, DUOX2, DUOX2A, DYNC2H1, DYSF, EIF2AK3, EIF2B5, ELP1, ERCC2, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, F2, F5, FAH, FAM126A, FAM161A, FANCA, FANCC, FANCG, FH, FKBP, FKTN, FOXRED1, FTCD, FUCA1, G6PC, GAA, GALC, GALNS, GALT, GAMT, GBA, GBE1, GCDH, GADAP1, GDF5, GFM1, GJB2, GJB6, GJB1, GLDC, GLE1, GNE, GNPTAB, GNPTG, GNS, GSS, GUCY2D, GUSB, HADHA, HADHB, HAX1, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HJV, HLCS, HMGCL, HOGA1, HPS1, HPS3, HPS4, HSD17B4, HSD3B2, HYLS1, IDUA, IVD, IYD, JAK3, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LCA5, LDLRAP1, LHX3, LIFR, LIPA, LMBRD1, LOXHD1, LPL, LRP2, LRPPRC, LYST, MAN2B1, MANBA, MCOLN1, MCPH1, MED17, MESP2, MFSD8, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MPI, MPL, MPV17, MTHFR, MTMR2, MTRR, MTPP, MUT, MVK, MYO7A, NAGA, NAGLU, NAGS, NBN, NDRG1, NDUFAF2, NDUFAF5, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NEB, NEU1, NPC1, NPC2, NPHP1, NPHS1, NPHS2, NTRK1, OAT, OCA2, OPA3, OTOF, P3H1, PAH, PANK2, PC, PCCA, PCCB, PCDH15, PCNT, PDHB, PEX1, PEX10, PEX12, PEX2, PEX26, PEX6, PEX7, PFKM, PHGDH, PHYH, PKHD1, PLA2G6, PLOD1, PMM2, POLG, POLR1C, POMGNT1, POMT1, POMT2, POR, PPT1, PRF1, PROPI, PSAP, PTS, PUS1, QDPR, RAB23, RAG1, RAG2, RAPSN, RARS2, RAX, RDH12, RMRP, RNASEH2B, RPE65, RPGRIP1L, RTEL1, SACS, SAMD9, SAMHD1, SCD2, SEPPSECS, SERPINA1, SGCA, SGCB, SGCD, SGCG, SGSH, SH3TC2, SLC12A6, SLC17A5, SLC19A3, SLC1A4, SLC22A5, SLC25A13, SLC25A15, SLC26A2, SLC26A3, SLC35A3, SLC37A4, SLC39A4, SLC45A2, SLC46A1, SLC5A5, SLC7A7, SMARCAL1, SMN1, SMPD1, SPG11, SPINK5, STAR, SUMF1, SURF1, TCIRG1, TCTN2, TECPR2, TF, TG, TGM1, TH, TMEM216, TPO, TPP1, TRDN, TRIM32, TRMU, TSEN54, TSFM, TSHB, TTC37, TTPA, TYMP, TYR, TYRP1, UGT1A1, USH1C, USH1G, USH2A, VPS13A, VPS13B, VPS45, VPS53, VRK1, VSX2, WHRN, WRN, XPA, XPC, ZFYVE26

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 99.54% and 99.50% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory



regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes and Limitations

CFTR: Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. **CRYL1:** As mutations in the CRYL1 gene are not known to be associated with any clinical condition, sequence variants in this gene are not analyzed. However, to increase copy number detection sensitivity for large deletions including this gene and a neighboring gene on the panel (GJB6, also known as connexin 30), this gene was evaluated for copy number variation. **CYP11B1:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP11B2:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP21A2:** Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of the clinical findings, biochemical profile, and family history of each patient. CYP21A2 variants primarily associated with non-classic congenital adrenal hyperplasia (CAH) are not included in this analysis (PubMed: 23359698). The variants associated with non-classic disease, including but not limited to c.188A>T (p.His63Leu), c.844G>T (p.Val282Leu), c.1174G>A (p.Ala392Thr), and c.1360C>T (p.Pro454Ser) will not be reported. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH. **DUOX2:** The current testing method is not able to reliably detect variants in exons 6-8 of the DUOX2 gene (NM_014080.5) due to significant interference by the highly homologous gene, DUOX1. **F2:** The common risk allele NM_000506.5:c.*97G>A is not included in this analysis. **F5:** The common Factor 5 "Leiden" allele is not typically reported as this variant is associated with low disease penetrance. **GALT:** In general, the D2 "Duarte" allele is not reported if detected, but can be reported upon request. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state (PubMed: 25473725, 30593450). **GBA:** The current testing method may not be able to reliably detect certain pathogenic variants in the GBA gene due to homologous recombination between the pseudogene and the functional gene. **HBA1:** The phase of heterozygous alterations in the HBA1 gene cannot be determined, but can be confirmed through parental testing. **HBA2:** The phase of heterozygous alterations in the HBA2 gene cannot be determined, but can be confirmed through parental testing. **MTHFR:** As recommended by ACMG, the two common polymorphisms in the MTHFR gene - c.1286A>C (p.Glu429Ala, also known as c.1298A>C) and c.665C>T (p.Ala222Val, also known as c.677C>T) - are not reported in this test due to lack of sufficient clinical utility to merit testing (PubMed: 23288205). **NEB:** This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis. **NPHS2:** If detected, the variant NM_014625.3:c.686G>A (p.Arg229Gln) will not be reported as this variant is not significantly associated with disease when homozygous or in the compound heterozygous state with variants in exons 1-6 of NPHS2. **RNASEH2B:** All variants located in the last two exons of the HGMD transcript (NM_024570.4) should be classified as VUS. **SERPINA1:** If detected the variant NM_000295.5:c.863A>T (p.Glu288Val) will not be reported as this variant is associated with low disease penetrance and is not associated with severe early onset disease. **SMN1:** The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: 20301526). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration, but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported. **TRDN:** Due to high GC content of certain exons, copy number analysis may have reduced sensitivity for partial gene deletions/duplications of TRDN. Confirmation of

4978 Santa Anita Ave.
Temple City, CA 91780
(p) 626-350-0537 (f) 626-454-1667
info@fulgentgenetics.com
www.fulgentgenetics.com



partial gene deletions/duplications are limited to individuals with a positive personal history of cardiac arrhythmia and/or individuals carrying a pathogenic/likely pathogenic sequence variant. UGT1A1: Common variants in the *UGT1A1* gene (population allele frequency >5%) are typically not reported as they do not cause a Mendelian condition. WRN: Due to the interference by highly homologous regions within the WRN gene, our current testing method has less sensitivity to detect variants in exons 10-11 of WRN (NM_000553.6).

SIGNATURE:

A handwritten signature in black ink that reads "H. Gao".

Dr. Harry Gao, DABMG, FACMG on 6/21/2023 12:51 PM PDT
Electronically signed

DISCLAIMER:

This test was developed and its performance characteristics determined by **Fulgent Genetics**. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.





Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>ABCA12</i>	Congenital ichthyosis, ABCA12-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ABCA3</i>	Surfactant metabolism dysfunction, pulmonary 3	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
<i>ABCA4</i>	Stargardt disease	AR	General Population	1 in 51	98%	1 in 2,501	1 in 510,204
<i>ABCB11</i>	Progressive familial intrahepatic cholestasis	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
<i>ABCC8</i>	Familial hyperinsulinism	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 44	98%	1 in 2,151	1 in 378,576
			Finnish Population	1 in 25	98%	1 in 1,201	1 in 120,100
			Middle-Eastern Population	1 in 25	98%	1 in 1,201	1 in 120,100
<i>ACAD9</i>	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ACADVL</i>	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	General Population	1 in 118	93%	1 in 1,672	1 in 789,184
			Middle-Eastern Population	1 in 74	93%	1 in 1,044	1 in 309,024
			Native American Population	1 in 61	93%	1 in 858	1 in 209,352
			South Asian/Indian Population	1 in 73	93%	1 in 1,030	1 in 300,760
<i>ACAT1</i>	3-ketothiolase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ACOX1</i>	Peroxisomal acyl-CoA oxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ACSF3</i>	Combined malonic and methylmalonic aciduria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ADA</i>	Adenosine deaminase deficiency	AR	General Population	1 in 224	93%	1 in 3,187	1 in 2,855,552
<i>ADAMTS2</i>	Ehlers-Danlos syndrome, dermatosparaxis type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 248	98%	1 in 12,351	<1 in 10 million
<i>ADGRG1</i>	Bilateral frontoparietal polymicrogyria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ADK</i>	Hypermethioninemia due to adenosine kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>AGA</i>	Aspartylglucosaminuria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 71	98%	1 in 3,501	1 in 994,284
<i>AGL</i>	Glycogen storage disease type III	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Faroese Population	1 in 28	95%	1 in 541	1 in 60,592
			Inuit Population	1 in 25	95%	1 in 481	1 in 48,100
			North African Jewish Population	1 in 37	95%	1 in 721	1 in 106,708
<i>AGPS</i>	Rhizomelic chondrodysplasia punctata, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>AGXT</i>	Primary hyperoxaluria type 1	AR	General Population	1 in 120	99%	1 in 11,901	1 in 5,712,480
			Caucasian / European Population	1 in 173	99%	1 in 17,201	<1 in 10 million
<i>AHI1</i>	Joubert syndrome, AHI1-related	AR	General Population	1 in 448	99%	1 in 44,701	<1 in 10 million
<i>AIPL1</i>	Childhood-onset severe retinal dystrophy, AIPL1-related	AR	General Population	1 in 409	99%	1 in 40,801	<1 in 10 million
<i>ALDH3A2</i>	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>ALDOB</i>	Hereditary fructose intolerance	AR	General Population	1 in 122	99%	1 in 12,101	1 in 5,905,288
			African/African American Population	1 in 250	99%	1 in 24,901	<1 in 10 million
			Caucasian / European Population	1 in 67	99%	1 in 6,601	1 in 1,769,068
			Middle-Eastern Population	1 in 97	99%	1 in 9,601	1 in 3,725,188
<i>ALG6</i>	Congenital disorder of glycosylation type Ic	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ALMS1</i>	Alstrom syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ALPL</i>	Hypophosphatasia	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Caucasian / European Population	1 in 274	95%	1 in 5,461	1 in 5,985,256
			Mennonite Population	1 in 25	95%	1 in 481	1 in 48,100
<i>AMT</i>	Glycine encephalopathy	AR	General Population	1 in 373	98%	1 in 18,601	<1 in 10 million
			Finnish Population	1 in 117	98%	1 in 5,801	1 in 2,714,868
<i>AQP2</i>	Nephrogenic diabetes insipidus	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 169	95%	1 in 3,361	1 in 2,272,036
<i>ARG1</i>	Arginase deficiency	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
<i>ARL13B</i>	Joubert syndrome, ARL13B-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ARSA</i>	Metachromatic leukodystrophy	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Caucasian / European Population	1 in 78	99%	1 in 7,701	1 in 2,402,712
			Yemenite Jewish Population	1 in 75	99%	1 in 7,401	1 in 2,220,300
<i>ARSB</i>	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			Western Australian Population	1 in 283	98%	1 in 14,101	<1 in 10 million
<i>ASL</i>	Argininosuccinate lyase deficiency	AR	General Population	1 in 132	90%	1 in 1,311	1 in 692,208
<i>ASNS</i>	Asparagine synthetase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Iranian Jewish Population	1 in 80	99%	1 in 7,901	1 in 2,528,320
<i>ASPA</i>	Canavan disease	AR	General Population	1 in 300	97%	1 in 9,968	<1 in 10 million
			Ashkenazi Jewish Population	1 in 55	96%	1 in 1,351	1 in 297,220
<i>ASS1</i>	Citrullinemia	AR	General Population	1 in 119	96%	1 in 2,951	1 in 1,404,676
			East Asian Population	1 in 132	96%	1 in 3,276	1 in 1,729,728



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>ATM</i>	Ataxia-telangiectasia	AR	General Population	1 in 100	92%	1 in 1,239	1 in 495,600
<i>ATP6V1B1</i>	Renal tubular acidosis with deafness	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ATP7B</i>	Wilson disease	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Caucasian / European Population	1 in 42	98%	1 in 2,051	1 in 344,568
			Ashkenazi Jewish Population	1 in 70	98%	1 in 3,451	1 in 966,280
<i>BBS1</i>	Bardet-Biedl syndrome type 1	AR	General Population	1 in 367	99%	1 in 36,601	<1 in 10 million
<i>BBS10</i>	Bardet-Biedl syndrome type 10	AR	General Population	1 in 395	99%	1 in 39,401	<1 in 10 million
<i>BBS12</i>	Bardet-Biedl syndrome type 12	AR	General Population	1 in 791	99%	1 in 79,001	<1 in 10 million
<i>BBS2</i>	Bardet-Biedl syndrome 2	AR	General Population	1 in 621	99%	1 in 62,001	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	99%	1 in 10,601	1 in 4,537,228
<i>BBS2</i>	Retinitis Pigmentosa 74	AR	General Population	1 in 621	99%	1 in 62,001	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	99%	1 in 10,601	1 in 4,537,228
<i>BCKDHA</i>	Maple syrup urine disease type Ia	AR	General Population	1 in 321	98%	1 in 16,001	<1 in 10 million
			Mennonite Population	1 in 10	98%	1 in 451	1 in 18,404
<i>BCKDHB</i>	Maple syrup urine disease type Ib	AR	General Population	1 in 364	98%	1 in 18,151	<1 in 10 million
			Ashkenazi Jewish Population	1 in 97	98%	1 in 4,801	1 in 1,862,788
<i>BCS1L</i>	Björnstad syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>BCS1L</i>	GRACILE syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>BCS1L</i>	Mitochondrial complex III deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>BLM</i>	Bloom syndrome	AR	General Population	1 in 800	87%	1 in 6,147	<1 in 10 million
			Ashkenazi Jewish Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
<i>BSND</i>	Barter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CAPN3</i>	Limb-girdle muscular dystrophy type 2A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 103	98%	1 in 5,101	1 in 2,101,612
<i>CASQ2</i>	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
<i>CBS</i>	Homocystinuria due to cystathionine beta-synthase deficiency	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
			Caucasian / European Population	1 in 86	99%	1 in 8,501	1 in 2,924,344
			Middle-Eastern Population	1 in 21	99%	1 in 2,001	1 in 168,084
<i>CC2D2A</i>	Joubert syndrome 9	AR	General Population	1 in 201	99%	1 in 20,001	1 in 16,080,804
<i>CCDC103</i>	Primary ciliary dyskinesia, type 17	AR	General Population	1 in 316	98%	1 in 15,751	<1 in 10 million
<i>CCDC39</i>	Primary ciliary dyskinesia, type 14	AR	General Population	1 in 211	98%	1 in 10,501	1 in 8,862,844
<i>CCDC88C</i>	Congenital hydrocephalus 1	AR	General Population	1 in 137	99%	1 in 13,601	1 in 7,453,348
<i>CDH23</i>	Usher syndrome, type 1D	AR	General Population	1 in 285	90%	1 in 2,841	1 in 11,364
<i>CEP290</i>	Joubert syndrome 5	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
<i>CEP290</i>	Leber congenital amaurosis 10	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
<i>CEP290</i>	Bardet-Biedl syndrome 14	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
<i>CEP290</i>	CEP290-related disorders	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
<i>CEP290</i>	Senior-Løken syndrome 6	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
<i>CEP290</i>	Meckel syndrome 4	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
<i>CFTR</i>	Cystic Fibrosis	AR	General Population	1 in 32	99%	1 in 3,101	1 in 396,928
			African/African American Population	1 in 61	99%	1 in 6,001	1 in 1,464,244
			Ashkenazi Jewish Population	1 in 24	99%	1 in 2,301	1 in 220,896
			Caucasian / European Population	1 in 25	99%	1 in 2,401	1 in 240,100
			East Asian Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Latino Population	1 in 58	99%	1 in 5,701	1 in 1,322,632
<i>CHRNAE</i>	Congenital myasthenic syndrome	AR	General Population	1 in 408	99%	1 in 40,701	<1 in 10 million
<i>CHRNAE</i>	Multiple pterygium syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CHST6</i>	Macular corneal dystrophy, CHST6-related	AR	General Population	1 in 79	99%	1 in 7,801	1 in 2,465,116
<i>CIITA</i>	Bare lymphocyte syndrome, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CLN3</i>	Neuronal ceroid lipofuscinosis	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
			Finnish Population	1 in 72	98%	1 in 3,551	1 in 1,022,688
<i>CLN5</i>	Neuronal ceroid lipofuscinosis 5	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 115	95%	1 in 2,281	1 in 1,049,260
<i>CLN6</i>	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General Population	<1 in 500	92%	1 in 6,239	<1 in 10 million
<i>CLN8</i>	Neuronal ceroid lipofuscinosis, CLN8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 135	95%	1 in 2,681	1 in 1,447,740
<i>CLRN1</i>	Usher syndrome, type 3A	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 120	98%	1 in 5,951	1 in 2,856,480
			Finnish Population	1 in 70	98%	1 in 3,451	1 in 966,280
<i>CNGB3</i>	Achromatopsia	AR	General Population	1 in 87	99%	1 in 8,601	1 in 2,993,148
			Micronesian Population	1 in 2	99%	1 in 101	1 in 808



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>COL27A1</i>	Steel syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>COL4A3</i>	Alport syndrome, COL4A3-related	AR	General Population Ashkenazi Jewish Population	1 in 267 1 in 188	98% 98%	1 in 13,301 1 in 9,351	<1 in 10 million 1 in 7,031,952
<i>COL4A4</i>	Alport syndrome, COL4A4-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
<i>COL7A1</i>	Dystrophic epidermolysis bullosa	AR	General Population	1 in 196	97%	1 in 6,501	1 in 5,096,784
<i>COX15</i>	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>CPS1</i>	Carbamoylphosphate synthetase I deficiency	AR	General Population	1 in 570	98%	1 in 28,451	<1 in 10 million
<i>CPT1A</i>	Carnitine palmitoyltransferase IA deficiency	AR	General Population Hutterite Population	1 in 354 1 in 16	90% 90%	1 in 3,531 1 in 151	1 in 4,999,896 1 in 9,664
<i>CPT2</i>	Carnitine palmitoyltransferase II deficiency	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 51	95% 95%	1 in 9,981 1 in 1,001	<1 in 10 million 1 in 204,204
<i>CRB1</i>	Leber congenital amaurosis 8	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
<i>CRB1</i>	Retinitis pigmentosa 12	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
<i>CRYL1</i>	GJB6-CRYL1 related nonsyndromic hearing loss	UK	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
<i>CTNS</i>	Cystinosis	AR	General Population British Population Moroccan Jewish Population	1 in 158 1 in 81 1 in 100	99% 99% 99%	1 in 15,701 1 in 8,001 1 in 9,901	1 in 9,923,032 1 in 2,592,324 1 in 3,960,400
<i>CTSA</i>	Galactosialidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>CTSC</i>	Papillon-Lefevre syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CTSD</i>	Neuronal ceroid lipofuscinosis, CTSD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>CTSK</i>	Pycnodysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYBA</i>	Chronic granulomatous disease	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
<i>CYP11A1</i>	Congenital adrenal insufficiency	AR	General Population	1 in 114	99%	1 in 11,301	1 in 5,153,256
<i>CYP11B1</i>	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR	General Population Moroccan Jewish Population	1 in 158 1 in 35	98% 98%	1 in 7,851 1 in 1,701	1 in 4,961,832 1 in 238,140
<i>CYP11B2</i>	Corticosterone methyl oxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYP17A1</i>	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYP1B1</i>	Primary congenital glaucoma	AR	General Population	1 in 50	99%	1 in 4,901	1 in 980,200
<i>CYP21A2</i>	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	General Population Inuit Population Middle-Eastern Population	1 in 61 1 in 9 1 in 35	99% 99% 99%	1 in 6,001 1 in 801 1 in 3,401	1 in 4,464,244 1 in 28,836 1 in 476,140
<i>CYP27A1</i>	Cerebrotendinous xanthomatosis	AR	General Population Moroccan Jewish Population	1 in 500 1 in 5	98% 98%	1 in 24,951 1 in 201	<1 in 10 million 1 in 4,020
<i>DBT</i>	Maple syrup urine disease, type II	AR	General Population	1 in 481	98%	1 in 24,001	<1 in 10 million
<i>DCLRE1C</i>	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>DDX11</i>	Warsaw breakage syndrome	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 68	99% 99%	1 in 49,901 1 in 6,701	<1 in 10 million 1 in 1,822,672
<i>DHCR7</i>	Smith-Lemli-Opitz syndrome	AR	General Population African/African American Population Ashkenazi Jewish Population	1 in 30 1 in 138 1 in 36	96% 96% 96%	1 in 726 1 in 3,426 1 in 876	1 in 87,120 1 in 1,891,152 1 in 126,144
<i>DHDDS</i>	Retinitis pigmentosa 59	AR	General Population Ashkenazi Jewish Population	1 in 296 1 in 118	98% 98%	1 in 14,751 1 in 5,851	<1 in 10 million 1 in 2,761,672
<i>DLD</i>	Dihydroliipoamide dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 107	98% 98%	1 in 24,951 1 in 5,301	<1 in 10 million 1 in 2,268,828
<i>DNAH5</i>	Primary ciliary dyskinesia, DNAH5-related	AR	General Population Ashkenazi Jewish Population	1 in 142 1 in 113	98% 99%	1 in 7,051 1 in 11,201	1 in 4,004,968 1 in 5,062,852
<i>DNAI1</i>	Primary ciliary dyskinesia, DNAI1-related	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
<i>DNAI2</i>	Primary ciliary dyskinesia, DNAI2-related	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
<i>DUOX2</i>	Congenital hypothyroidism, DUOX2-related	AR	General Population	1 in 366	91%	1 in 4,057	1 in 5,938,797
<i>DUOX2</i>	Congenital hypothyroidism, DUOX2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>DYNC2H1</i>	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	General Population	1 in 68	98%	1 in 3,351	1 in 924,876
<i>DYSF</i>	Limb-girdle muscular dystrophy type 2B	AR	General Population Japanese Population Libyan Jewish Population	<1 in 500 1 in 332 1 in 18	95% 95% 95%	1 in 9,981 1 in 6,621 1 in 341	<1 in 10 million 1 in 8,792,688 1 in 24,552
<i>EIF2AK3</i>	Wolcott-Rallison Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>EIF2B5</i>	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ELP1</i>	Familial Dysautonomia	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 31	99% 99%	1 in 29,901 1 in 3,001	<1 in 10 million 1 in 372,124



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>ERCC2</i>	Xeroderma pigmentosum, group D	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
<i>ERCC2</i>	Photosensitive trichothiodystrophy 1	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
<i>ERCC2</i>	Cerebrooculofacioskeletal syndrome 2	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
<i>ERCC5</i>	Xeroderma Pigmentosa, group G	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ERCC6</i>	De Sanctis-Cacchione syndrome	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
<i>ERCC6</i>	Cockayne syndrome type B	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
<i>ERCC8</i>	Cockayne syndrome type A	AR	General Population	1 in 822	98%	1 in 41,051	<1 in 10 million
<i>ESCO2</i>	Roberts syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ETFA</i>	Glutaric aciduria IIA	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ETFB</i>	Glutaric aciduria IIB	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ETFDH</i>	Glutaric aciduria IIC	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 1,080,696
<i>ETHE1</i>	Ethylmalonic encephalopathy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>EVC</i>	Weyers acrofacial dysostosis, EVC-related	AR	General Population	1 in 142	98%	1 in 7,051	1 in 4,004,968
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC</i>	Ellis-van Creveld syndrome, EVC-related	AR	General Population	1 in 142	98%	1 in 7,051	1 in 4,004,968
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC2</i>	Weyers acrofacial dysostosis, EVC2-related	AR	General Population	1 in 240	98%	1 in 11,951	<1 in 10 million
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC2</i>	Ellis-van Creveld syndrome, EVC2-related	AR	General Population	1 in 240	98%	1 in 11,951	<1 in 10 million
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EXOSC3</i>	Pontocerebellar hypoplasia type 1B	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>F2</i>	Prothrombin-related conditions	AR	General Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Caucasian / European Population	1 in 4	99%	1 in 301	1 in 4,816
<i>F5</i>	Factor V deficiency	AR	General Population	1 in 36	99%	1 in 3,501	1 in 504,144
			Caucasian / European Population	1 in 19	99%	1 in 1,801	1 in 136,876
			Latino Population	1 in 45	99%	1 in 4,401	1 in 792,180
			African/African American Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
			East Asian Population	1 in 222	99%	1 in 22,101	<1 in 10 million
			Native American Population	1 in 80	99%	1 in 7,901	1 in 2,528,320
<i>FAH</i>	Tyrosinemia, type 1	AR	General Population	1 in 99	95%	1 in 1,961	1 in 776,556
			Ashkenazi Jewish Population	1 in 150	95%	1 in 2,981	1 in 1,788,600
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 1,181,448
			French Canadian Population	1 in 66	95%	1 in 1,301	1 in 343,464
			South Asian/Indian Population	1 in 172	95%	1 in 3,421	1 in 2,353,648
<i>FAM126A</i>	Hypomyelinating leukodystrophy type 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FAM126A</i>	Hypomyelinating leukodystrophy type 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FAM161A</i>	Retinitis pigmentosa 28	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
<i>FANCA</i>	Fanconi anemia group A	AR	General Population	1 in 239	99%	1 in 23,801	<1 in 10 million
			Moroccan Jewish	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Indian Jewish Population	1 in 27	99%	1 in 2,601	1 in 280,908
<i>FANCC</i>	Fanconi anemia group C	AR	General Population	1 in 535	99%	1 in 53,401	<1 in 10 million
			Ashkenazi Jewish Population	1 in 99	99%	1 in 9,801	1 in 3,881,196
<i>FANCG</i>	Fanconi anemia group G	AR	General Population	1 in 632	90%	1 in 6,311	<1 in 10 million
<i>FH</i>	Fumarase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 99	99%	1 in 9,801	1 in 3,881,196
<i>FKRP</i>	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>FKRP</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FKTN</i>	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
			Japanese Population	1 in 82	99%	1 in 8,101	1 in 2,657,128
<i>FKTN</i>	Fukuyama congenital muscular dystrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
			Japanese Population	1 in 82	99%	1 in 8,101	1 in 2,657,128
<i>FKTN</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FOXRED1</i>	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FTCD</i>	Glutamate formiminotransferase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FUCA1</i>	Fucosidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>G6PC</i>	Glycogen storage disease, type 1a	AR	General Population	1 in 177	95%	1 in 3,521	1 in 2,492,868
			Ashkenazi Jewish Population	1 in 64	95%	1 in 1,261	1 in 322,816



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
GAA	Pompe disease	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
			African/African American Population	1 in 60	98%	1 in 2,951	1 in 708,240
			East Asian Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
GALC	Krabbe disease	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			Israeli Druze Population	1 in 6	99%	1 in 501	1 in 12,024
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR	General Population	1 in 224	97%	1 in 7,434	1 in 6,660,864
GALT	Galactosemia	AR	General Population	1 in 110	99%	1 in 10,901	1 in 4,796,440
			African/African American Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Ashkenazi Jewish Population	1 in 127	99%	1 in 12,601	1 in 6,401,308
GAMT	Guanidinoacetate methyltransferase deficiency	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
GBA	Gaucher disease	AR	General Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			African/African American Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Ashkenazi Jewish Population	1 in 15	99%	1 in 1,401	1 in 84,060
GBE1	Glycogen storage disease IV	AR	General Population	1 in 387	99%	1 in 38,601	<1 in 10 million
GCDH	Glutaric aciduria, type I	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Amish Population	1 in 9	98%	1 in 401	1 in 14,436
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR	General Population	1 in 152	99%	1 in 15,101	1 in 9,181,408
GDF5	Du Pan Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GJB2	Nonsyndromic hearing loss 1A	AR	General Population	1 in 42	99%	1 in 4,101	1 in 688,968
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 240,100
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 168,084
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Latino Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 8,702,992
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
GLB1	GM1-gangliosidosis	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
GLB1	Mucopolysaccharidosis type IVB (Morquio syndrome B)	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
GLDC	Glycine encephalopathy, GLDC-related	AR	General Population	1 in 193	98%	1 in 9,601	1 in 7,411,972
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 5,429,268
GLE1	Lethal congenital contracture syndrome 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	<1 in 500	99%	1 in 49,901	1 in 99,802,000
			Iranian Jewish Population	1 in 11	99%	1 in 1,001	1 in 44,044
GNPTAB	Mucopolipidosis II alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTAB	Mucopolipidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTG	Mucopolipidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GSS	Glutathione synthetase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GUCY2D	Leber congenital amaurosis 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GUSB	Mucopolysaccharidosis type VII	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
HADHA	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHB	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HAX1	Severe congenital neutropenia, HAX1-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
HBA1	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>HBA2</i>	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
<i>HBB</i>	Sickle cell disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
<i>HBB</i>	Hemoglobin C disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
<i>HBB</i>	Beta thalassemia	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			African/African American Population	1 in 10	99%	1 in 901	1 in 36,040
			East Asian Population	1 in 50	99%	1 in 4,901	1 in 980,200
			Latino Population	1 in 128	99%	1 in 12,701	1 in 6,502,912
			Mediterranean Population	1 in 3	99%	1 in 201	1 in 2,412
			South Asian/Indian Population	1 in 25	99%	1 in 2,401	1 in 240,100
<i>HEXA</i>	Tay-Sachs disease	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 27	99%	1 in 2,601	1 in 280,908
			Moroccan Jewish Population	1 in 110	99%	1 in 10,901	1 in 4,796,440
<i>HEXB</i>	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million
<i>HGSNAT</i>	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	General Population	1 in 434	98%	1 in 21,651	<1 in 10 million
			Caucasian / European Population	1 in 345	98%	1 in 17,201	<1 in 10 million
<i>HJV</i>	Hemochromatosis, type 2A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
<i>HLCS</i>	Holocarboxylase synthetase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HMGCL</i>	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HOGA1</i>	Primary hyperoxaluria type III	AR	General Population	1 in 184	99%	1 in 18,301	<1 in 10 million
<i>HPS1</i>	Hermansky-Pudlak syndrome 1	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
			Puerto Rican Population	1 in 21	98%	1 in 1,001	1 in 84,084
<i>HPS3</i>	Hermansky-Pudlak syndrome 3	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
<i>HPS4</i>	Hermansky-Pudlak syndrome 4	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HSD17B4</i>	D-bifunctional protein deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>HSD3B2</i>	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HYLS1</i>	Hydrolethals syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
<i>IDUA</i>	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Caucasian / European Population	1 in 153	95%	1 in 3,041	1 in 1,861,092
<i>IVD</i>	Isovaleric Acidemia	AR	General Population	1 in 167	90%	1 in 1,661	1 in 1,109,548
			African/African American Population	1 in 100	90%	1 in 991	1 in 396,400
			Caucasian / European Population	1 in 115	90%	1 in 1,141	1 in 524,860
			East Asian Population	1 in 407	90%	1 in 4,061	1 in 6,611,308
<i>IYD</i>	Thyroid dysmorphogenesis, IYD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>JAK3</i>	Severe combined immunodeficiency, JAK3-related	AR	General Population	1 in 299	99%	1 in 29,801	<1 in 10 million
<i>KCNJ11</i>	Congenital hyperinsulinism	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
			Caucasian / European Population	1 in 232	99%	1 in 23,101	<1 in 10 million
<i>KCNJ11</i>	Permanent neonatal diabetes mellitus	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
			Caucasian / European Population	1 in 232	99%	1 in 23,101	<1 in 10 million
<i>LAMA2</i>	Muscular dystrophy, LAMA2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
<i>LAMA3</i>	Junctional epidermolysis bullosa, LAMA3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMA3</i>	Laryngo-onycho-cutaneous syndrome	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMB3</i>	Junctional epidermolysis bullosa, LAMB3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMC2</i>	Junctional epidermolysis bullosa, LAMC2-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LCA5</i>	Leber congenital amaurosis 5	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>LDLRAP1</i>	Familial Hypercholesterolemia	AR	General Population	1 in 8	99%	1 in 701	1 in 22,432
			Amish Population	1 in 2	99%	1 in 101	1 in 808
			Caucasian / European Population	1 in 7	99%	1 in 601	1 in 16,828
			French Canadian Population	1 in 8	99%	1 in 701	1 in 22,432
<i>LHX3</i>	Combined pituitary hormone deficiency 3	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
<i>LIFR</i>	Stuve-Wiedemann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>LIPA</i>	Lysosomal acid lipase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 112	99%	1 in 11,101	1 in 4,973,248
			Iranian Jewish Population	1 in 26	99%	1 in 2,501	1 in 260,104
<i>LMBRD1</i>	Methylmalonic aciduria and homocystinuria, cblF type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>LOXHD1</i>	Nonsyndromic hearing loss 77	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 180	98%	1 in 8,951	1 in 6,444,720
<i>LPL</i>	Familial lipoprotein lipase deficiency	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			French Canadian Population	1 in 46	99%	1 in 4,501	1 in 828,184
<i>LRP2</i>	Donnai-Barrow syndrome	AR	General Population	1 in 214	99%	1 in 10,651	1 in 9,117,256
<i>LRPPRC</i>	Leigh syndrome with Complex IV deficiency	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
			Faroese Population	1 in 21	98%	1 in 1,001	1 in 84,084
			French Canadian Population	1 in 22	98%	1 in 1,051	1 in 92,488
<i>LYST</i>	Chediak-Higashi syndrome	AR	General Population	<1 in 500	90%	1 in 4,991	1 in 9,982,000
<i>MAN2B1</i>	Alpha-Mannosidosis	AR	General Population	1 in 354	99%	1 in 35,301	<1 in 10 million
			Caucasian / European Population	1 in 274	99%	1 in 27,301	<1 in 10 million
<i>MANBA</i>	Beta-Mannosidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MCOLN1</i>	Mucopolidosis IV	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<i>MCPH1</i>	Primary microcephaly 1, recessive	AR	General Population	1 in 147	99%	1 in 14,601	1 in 8,585,388
<i>MED17</i>	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Bukharan/Kurdish Jewish Population	1 in 20	99%	1 in 1,901	1 in 152,080
<i>MESP2</i>	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MFSD8</i>	Neuronal ceroid lipofuscinosis, MFSD8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>MKS1</i>	Bardet-Biedl syndrome 13	AR	General Population	1 in 260	98%	1 in 12,951	<1 in 10 million
			Finnish Population	1 in 47	98%	1 in 2,301	1 in 432,588
<i>MKS1</i>	Joubert syndrome 28	AR	General Population	1 in 260	98%	1 in 12,951	<1 in 10 million
			Finnish Population	1 in 47	98%	1 in 2,301	1 in 432,588
<i>MKS1</i>	Meckel syndrome 1	AR	General Population	1 in 260	98%	1 in 12,951	<1 in 10 million
			Finnish Population	1 in 47	98%	1 in 2,301	1 in 432,588
<i>MLC1</i>	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Libyan Jewish Population	1 in 40	99%	1 in 3,901	1 in 624,160
<i>MLYCD</i>	Malonyl-CoA decarboxylase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MMAA</i>	Methylmalonic aciduria, cblA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
<i>MMAB</i>	Methylmalonic aciduria, cblB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
<i>MMACHC</i>	Methylmalonic aciduria and homocystinuria, cblC type	AR	General Population	1 in 134	90%	1 in 1,331	1 in 713,416
<i>MMADHC</i>	Methylmalonic aciduria and homocystinuria, cblD type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MPI</i>	Congenital disorder of glycosylation type Ib	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MPL</i>	Congenital amegakaryocytic thrombocytopenia	AR	General Population	1 in 102	98%	1 in 5,051	1 in 2,060,808
			Ashkenazi Jewish Population	1 in 55	98%	1 in 2,701	1 in 594,220
<i>MPV17</i>	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR	General Population	<1 in 500	96%	1 in 12,476	<1 in 10 million
			Native American Population	1 in 20	96%	1 in 476	1 in 38,080
<i>MTHFR</i>	Homocystinuria, MTHFR-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>MTMR2</i>	Charcot-Marie-Tooth disease, type 4B1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MTRR</i>	Homocystinuria-megaloblastic anemia, cobalamin E type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MTTP</i>	Abetalipoproteinemia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 180	98%	1 in 8,951	1 in 6,444,720
<i>MUT</i>	Methylmalonic aciduria-methylmalonyl-CoA mutase deficiency	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<i>MVK</i>	Hyperimmunoglobulinemia D syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MVK</i>	Mevalonate kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MYO7A</i>	Usher syndrome, type 1B	AR	General Population	1 in 206	98%	1 in 10,251	1 in 8,446,824
			East Asian Population	1 in 62	98%	1 in 3,051	1 in 756,648
<i>MYO7A</i>	Non-syndromic hearing loss, MYO7A-related	AR	General Population	1 in 206	98%	1 in 10,251	1 in 8,446,824
			East Asian Population	1 in 62	98%	1 in 3,051	1 in 756,648
<i>NAGA</i>	Schindler disease types 1 and 3	AR	General Population	1 in 94	99%	1 in 9,301	1 in 3,497,176



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 346	99%	1 in 34,501	<1 in 10 million
			East Asian Population	1 in 298	99%	1 in 29,701	<1 in 10 million
NAGS	N-acetylglutamate synthase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
NBN	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR	General Population	1 in 22	98%	1 in 1,051	1 in 92,488
NDUFAF2	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
			Ashkenazi Jewish Population	1 in 290	98%	1 in 14,451	<1 in 10 million
NDUFS4	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFS4	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Hutterite Population	1 in 27	99%	1 in 2,601	1 in 280,908
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Bukharan/Kurdish Jewish Population	1 in 24	99%	1 in 2,301	1 in 220,896
NDUFS7	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NEB	Nemaline myopathy	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Amish Population	1 in 11	98%	1 in 501	1 in 22,044
			Ashkenazi Jewish Population	1 in 108	98%	1 in 5,351	1 in 2,311,632
			Finnish Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
NEU1	Sialidosis, type I and II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NPC1	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
NPC2	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
NPHP1	Joubert syndrome 4	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHP1	Nephronophthisis	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHP1	NPHP1-related disorders	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
NPHP1	Senior-Løken syndrome 1	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHS1	Congenital nephrotic syndrome, type 1	AR	General Population	1 in 289	98%	1 in 14,401	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
NPHS2	Congenital nephrotic syndrome, type 2	AR	General Population	1 in 289	98%	1 in 14,401	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
OAT	Gyrate atrophy of choroid and retina	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
OCA2	Oculocutaneous albinism type II	AR	General Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
OPA3	Costeff syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Iraqi Jewish Population	1 in 50	98%	1 in 2,451	1 in 490,200
OTOF	Nonsyndromic hearing loss, OTOF-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Spanish Population	1 in 106	99%	1 in 10,501	1 in 4,452,424
P3H1	Osteogenesis imperfecta, type VIII	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			West African Population	1 in 67	99%	1 in 6,601	1 in 1,769,068
			African American Population	1 in 250	99%	1 in 24,901	<1 in 10,000,000
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	General Population	1 in 93	99%	1 in 9,201	1 in 3,422,772
			Caucasian / European Population	1 in 63	99%	1 in 6,201	1 in 1,562,652
			Middle-Eastern Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
			South East Asian	1 in 59	99%	1 in 5,801	1 in 1,369,036
PANK2	Pantothenate kinase-associated neurodegeneration	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million
PC	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
PCCA	Propionic acidemia, PCCA-related	AR	General Population	1 in 224	96%	1 in 5,576	1 in 4,996,096
			Native American Population	1 in 85	96%	1 in 2,101	1 in 714,340
PCCB	Propionic acidemia, PCCB-related	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
			Native American Population	1 in 85	99%	1 in 8,401	1 in 2,856,340
PCDH15	Non-syndromic hearing loss, PCDH15-related	AR	General Population	1 in 395	98%	1 in 19,701	1 in 78,804
			Ashkenazi Jewish Population	1 in 72	98%	1 in 3,551	1 in 14,204
PCDH15	Usher syndrome, type 1F	AR	General Population	1 in 395	98%	1 in 19,701	1 in 78,804
			Ashkenazi Jewish Population	1 in 72	98%	1 in 3,551	1 in 14,204
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>PEX1</i>	Zellweger syndrome, PEX1-related	AR	General Population	1 in 147	95%	1 in 2,921	1 in 1,717,548
<i>PEX10</i>	Zellweger syndrome, PEX10-related	AR	General Population Japanese Population	1 in 500 1 in 354	95% 95%	1 in 9,981 1 in 7,061	<1 in 10 million 1 in 9,998,376
<i>PEX12</i>	Zellweger syndrome, PEX12-related	AR	General Population	1 in 373	95%	1 in 7,441	<1 in 10 million
<i>PEX2</i>	Zellweger syndrome, PEX2-related	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 123	95% 95%	1 in 9,981 1 in 2,441	<1 in 10 million 1 in 1,200,972
<i>PEX26</i>	Zellweger syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>PEX6</i>	Zellweger syndrome, PEX6-related	AR	General Population Yemenite Jewish Population	1 in 280 1 in 18	99% 99%	1 in 27,901 1 in 1,701	<1 in 10 million 1 in 122,472
<i>PEX7</i>	Rhizomelic chondrodysplasia punctata, type 1	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
<i>PFKM</i>	Glycogen storage disease VII	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 120	99% 99%	1 in 49,901 1 in 11,901	<1 in 10 million 1 in 5,712,480
<i>PHGDH</i>	Phosphoglycerate dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 280	98% 98%	1 in 24,951 1 in 13,951	<1 in 10 million 1 in 10 million
<i>PHYH</i>	Refsum disease	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>PKHD1</i>	Polycystic kidney disease, PKHD1-related	AR	General Population Ashkenazi Jewish Population	1 in 70 1 in 107	98% 98%	1 in 3,451 1 in 5,301	1 in 966,280 1 in 2,268,828
<i>PLA2G6</i>	Infantile neuroaxonal dystrophy	AR	General Population	1 in 500	97%	1 in 16,634	<1 in 10 million
<i>PLOD1</i>	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
<i>PMM2</i>	Congenital disorder of glycosylation type 1a	AR	General Population Ashkenazi Jewish Population Caucasian / European Population	1 in 63 1 in 57 1 in 71	99% 99% 99%	1 in 6,201 1 in 5,601 1 in 7,001	1 in 1,562,652 1 in 1,277,028 1 in 1,988,284
<i>POLG</i>	Ataxia neuropathy spectrum	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
<i>POLG</i>	Progressive external ophthalmoplegia	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
<i>POLG</i>	Myocerebrohepatopathy syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
<i>POLG</i>	POLG-related disorders	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
<i>POLG</i>	Alpers-Huttenlocher syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
<i>POLR1C</i>	Hypomyelinating Leukodystrophy, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>POLR1C</i>	Treacher Collins syndrome, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>POMGNT1</i>	Muscular dystrophy-dystroglycanopathy	AR	General Population Finnish Population	1 in 462 1 in 111	98% 98%	1 in 23,051 1 in 5,501	<1 in 10 million 1 in 2,442,444
<i>POMGNT1</i>	Retinitis pigmentosa 76	AR	General Population Finnish Population	1 in 462 1 in 111	98% 98%	1 in 23,051 1 in 5,501	<1 in 10 million 1 in 2,442,444
<i>POMGNT1</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>POMT1</i>	Muscular dystrophy-dystroglycanopathy, POMT1-related	AR	General Population	1 in 290	99%	1 in 28,901	<1 in 10 million
<i>POMT1</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>POMT2</i>	Muscular dystrophy-dystroglycanopathy, POMT2-related	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
<i>POMT2</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>POR</i>	Antley-Bixler syndrome	AR	General Population	1 in 159	98%	1 in 7,901	1 in 5,025,036
<i>PPT1</i>	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General Population Caucasian / European Population Finnish Population	1 in 368 1 in 488 1 in 75	98% 98% 98%	1 in 18,351 1 in 24,351 1 in 3,701	<1 in 10 million <1 in 10 million 1 in 1,110,300
<i>PRF1</i>	Hemophagocytic lymphohistiocytosis, familial, 2	AR	General Population	1 in 149	99%	1 in 14,801	1 in 8,821,396
<i>PROP1</i>	Combined pituitary hormone deficiency 2	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
<i>PSAP</i>	Metachromatic leukodystrophy due to saposin-b deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>PTS</i>	Tetrahydrobiopterin deficiency	AR	General Population	1 in 354	96%	1 in 8,826	<1 in 10 million
<i>PUS1</i>	Mitochondrial myopathy and sideroblastic anemia 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>QDPR</i>	Tetrahydrobiopterin deficiency, QDPR-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>RAB23</i>	Carpenter syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>RAG1</i>	Omenn syndrome, RAG1-related	AR	General Population	1 in 290	98%	1 in 14,451	1 in 16,763,160
<i>RAG2</i>	Omenn syndrome, RAG2-related	AR	General Population	1 in 137	98%	1 in 6,801	1 in 3,726,948
<i>RAPSN</i>	Congenital myasthenic syndrome, RAPSN-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>RAPSN</i>	Fetal akinesia deformation sequence	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>RARS2</i>	Pontocerebellar hypoplasia type 6	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>RAX</i>	Microphthalmia, isolated 3	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million
<i>RDH12</i>	Leber congenital amaurosis type 13	AR	General Population Caucasian / European Population	<1 in 500 1 in 456	98% 98%	1 in 24,951 1 in 22,751	<1 in 10 million <1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>RMRP</i>	Metaphyseal dysplasia without hypotrichosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RMRP</i>	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Finnish Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>RMRP</i>	Anauxetic dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RMRP</i>	Cartilage-hair hypoplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RNASEH2B</i>	Aicardi Goutieres syndrome 2	AR	General Population	1 in 217	99%	1 in 10,801	1 in 9,375,268
<i>RPE65</i>	Retinitis pigmentosa 20	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
<i>RPE65</i>	Leber congenital amaurosis 2	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
<i>RPGRIP1L</i>	COACH syndrome	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
<i>RPGRIP1L</i>	Joubert syndrome 7	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
<i>RPGRIP1L</i>	Meckel syndrome 5	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
<i>RTEL1</i>	Dyskeratosis congenita type 5	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 203	99%	1 in 20,201	<1 in 10 million
<i>SACS</i>	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			French Canadian Population	1 in 19	95%	1 in 361	1 in 27,436
<i>SAMD9</i>	Normophosphatemic Familial Tumoral Calcinosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Yemeni Jewish Population	1 in 25	99%	1 in 2,401	1 in 240,100
<i>SAMHD1</i>	Aicardi-Goutieres syndrome	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>SCO2</i>	Mitochondrial complex IV deficiency	AR	General Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
<i>SEPSECS</i>	Pontocerebellar hypoplasia type 2D	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Moroccan/Iraqi Jewish Population	1 in 44	99%	1 in 4,301	1 in 756,976
<i>SERPINA1</i>	Alpha-1 antitrypsin deficiency	AR	General Population	1 in 33	95%	1 in 641	1 in 84,612
			Caucasian / European Population	1 in 19	95%	1 in 361	1 in 27,436
<i>SGCA</i>	Limb-girdle muscular dystrophy, type 2D	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 288	98%	1 in 14,351	<1 in 10 million
			Finnish Population	1 in 150	98%	1 in 7,451	1 in 4,470,600
<i>SGCB</i>	Limb-girdle muscular dystrophy, type 2E	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 406	98%	1 in 20,251	<1 in 10 million
<i>SGCD</i>	Limb-girdle muscular dystrophy, type 2F	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SGCG</i>	Limb-girdle muscular dystrophy, type 2C	AR	General Population	1 in 381	98%	1 in 19,001	<1 in 10 million
			Moroccan Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			Roma / Gypsy Population	1 in 96	98%	1 in 4,751	1 in 1,824,384
<i>SGSH</i>	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR	General Population	1 in 454	98%	1 in 22,651	<1 in 10 million
			Caucasian / European Population	1 in 253	98%	1 in 12,601	<1 in 10 million
<i>SH3TC2</i>	Charcot-Marie-Tooth disease, SH3TC2-related	AR	General Population	1 in 69	99%	1 in 6,801	1 in 1,877,076
<i>SLC12A6</i>	Andermann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			French Canadian Population	1 in 23	99%	1 in 2,201	1 in 202,492
<i>SLC17A5</i>	Sialic acid storage disorder	AR	General Population	<1 in 500	91%	1 in 5,545	<1 in 10 million
			Finnish Population	1 in 100	91%	1 in 1,101	1 in 440,400
<i>SLC19A3</i>	Biotin-responsive basal ganglia disease	AR	General Population	1 in 109	99%	1 in 5,401	1 in 2,354,836
<i>SLC1A4</i>	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>SLC22A5</i>	Systemic primary carnitine deficiency	AR	Ashkenazi Jewish Population	1 in 106	99%	1 in 10,501	1 in 4,452,424
			General Population	1 in 129	99%	1 in 12,801	1 in 6,605,316
			African/African American Population	1 in 86	99%	1 in 8,501	1 in 2,924,344
			East Asian Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			Faroese Population	1 in 9	99%	1 in 801	1 in 28,836
			Pacific Islander Population	1 in 37	99%	1 in 3,601	1 in 532,948
<i>SLC25A13</i>	Citrin deficiency	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			East Asian Population	1 in 65	95%	1 in 1,281	1 in 333,060
<i>SLC25A15</i>	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			French Canadian Population	1 in 37	99%	1 in 3,601	1 in 532,948
<i>SLC26A2</i>	Diastrophic dysplasia	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
<i>SLC26A2</i>	Achondrogenesis, type IB	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
SLC26A2	Multiple epiphyseal dysplasia	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Atelosteogenesis II	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
SLC26A3	Congenital secretory chloride diarrhea	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Middle-Eastern Population	1 in 57	98%	1 in 2,801	1 in 638,628
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 453	98%	1 in 22,601	<1 in 10 million
SLC37A4	Glycogen storage disease, type Ib	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Ashkenazi Jewish Population	1 in 71	95%	1 in 1,401	1 in 397,884
SLC39A4	Acrodermatitis enteropathica	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC45A2	Oculocutaneous albinism, type IV	AR	General Population	1 in 159	98%	1 in 7,901	1 in 5,025,036
			Japanese Population	1 in 146	98%	1 in 7,251	1 in 4,234,584
SLC46A1	Hereditary folate malabsorption	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Puerto Rican Population	1 in 500	99%	1 in 49,901	<1 in 10 million
SLC5A5	Thyroid dysmorphogenesis, SLC5A5-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC7A7	Lysinuric protein intolerance	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 1,181,448
			Japanese Population	1 in 119	95%	1 in 2,361	1 in 1,123,836
SMARCAL1	Schimke immunosseous dysplasia	AR	General Population	1 in 500	90%	1 in 4,991	1 in 9,982,000
SMN1	Spinal muscular atrophy	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
			African/African American Population	1 in 72	71%	1 in 246	1 in 70,848
			Ashkenazi Jewish Population	1 in 67	91%	1 in 734	1 in 196,712
			Caucasian / European Population	1 in 47	95%	1 in 921	1 in 173,148
			East Asian Population	1 in 59	93%	1 in 830	1 in 195,880
			Latino Population	1 in 68	90%	1 in 671	1 in 182,512
			Sephardic Jewish Population	1 in 34	96%	1 in 826	1 in 112,336
SMN1	Spinal muscular atrophy silent carrier	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
SMPD1	Niemann-Pick disease, type A/B	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
			Ashkenazi Jewish Population	1 in 115	95%	1 in 2,281	1 in 1,049,260
			Latino Population	1 in 106	95%	1 in 2,101	1 in 890,824
SPG11	SPG11-related Neuromuscular Disorders	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
SPINK5	Netherton syndrome	AR	General Population	1 in 224	99%	1 in 23,301	<1 in 10 million
			Ashkenazi Jewish Population	1 in 17	99%	1 in 1,601	1 in 108,868
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SUMF1	Multiple sulfatase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 320	98%	1 in 15,951	<1 in 10 million
SURF1	Charcot-Marie-Tooth disease, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SURF1	Leigh syndrome, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TCIRG1	Osteopetrosis 1	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TCTN2	Meckel syndrome 8	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ethiopian Jewish Population	1 in 42	99%	1 in 4,101	1 in 688,968
			Yemenite Jewish Population	1 in 78	99%	1 in 7,701	1 in 2,402,712
TCTN2	Joubert syndrome 24	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TECPR2	Spastic paraplegia 49	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TF	Atransferrinemia	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
TG	Thyroid dysmorphogenesis, TG-related	AR	General Population	1 in 241	99%	1 in 24,001	<1 in 10 million
TGM1	Congenital ichthyosis	AR	General Population	1 in 224	95%	1 in 4,461	1 in 3,997,056
TH	Segawa syndrome	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
TMEM216	Joubert syndrome 2	AR	General Population	1 in 141	98%	1 in 7,001	1 in 3,948,564
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
TMEM216	Meckel syndrome 2	AR	General Population	1 in 141	98%	1 in 7,001	1 in 3,948,564
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
TPO	Thyroid dysmorphogenesis, TPO-related	AR	General Population	1 in 373	99%	1 in 37,201	<1 in 10 million
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	General Population	1 in 252	97%	1 in 8,368	1 in 8,434,944
			French Canadian Population	1 in 53	97%	1 in 1,734	1 in 367,608
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
TRIM32	Limb-girdle muscular dystrophy, type 2H	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Hutterite Population	1 in 12	98%	1 in 551	1 in 26,448
TRIM32	Bardet-Biedl syndrome 11	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Hutterite Population	1 in 12	98%	1 in 551	1 in 26,448



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>TRMU</i>	Liver failure, acute infantile	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Yemeni Jewish Population	1 in 34	98%	1 in 1,651	1 in 224,536
<i>TSEN54</i>	Pontocerebellar hypoplasia type 2A	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>TSMF</i>	Combined oxidative phosphorylation deficiency, TSMF-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
<i>TSHB</i>	Congenital hypothyroidism, TSHB-related	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
<i>TTC37</i>	Trichohepatoenteric syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>TTPA</i>	Ataxia with isolated vitamin E deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 267	90%	1 in 2,661	1 in 2,841,948
<i>TYMP</i>	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>TYR</i>	Oculocutaneous albinism types 1A and 1B	AR	General Population	1 in 20	99%	1 in 1,901	1 in 152,080
<i>TYRP1</i>	Oculocutaneous albinism, type III	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			African Population	1 in 47	98%	1 in 2,301	1 in 432,588
<i>UGT1A1</i>	Crigler-Najjar syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>USH1C</i>	Usher syndrome, type IC	AR	General Population	1 in 353	90%	1 in 3,521	1 in 4,971,652
			French Canadian Population	1 in 227	90%	1 in 2,261	1 in 2,052,988
<i>USH1C</i>	Non-syndromic hearing loss, USH1C-related	AR	General Population	1 in 353	90%	1 in 3,521	1 in 4,971,652
			French Canadian Population	1 in 227	90%	1 in 2,261	1 in 2,052,988
<i>USH1G</i>	Usher syndrome type IG	AR	General Population	1 in 434	99%	1 in 43,301	<1 in 10 million
<i>USH2A</i>	Usher syndrome, type 2A	AR	General Population	1 in 126	96%	1 in 3,126	1 in 1,575,504
			Caucasian / European Population	1 in 73	96%	1 in 1,801	1 in 525,892
			Ashkenazi Jewish Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Iranian Jewish Population	1 in 60	99%	1 in 5,901	1 in 1,416,240
<i>VPS13A</i>	Choreoacanthocytosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>VPS13B</i>	Cohen syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>VPS45</i>	Severe congenital neutropenia, VPS45-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>VPS53</i>	Pontocerebellar hypoplasia type 2E	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Moroccan Jewish Population	1 in 37	98%	1 in 1,801	1 in 266,548
<i>VRK1</i>	Pontocerebellar hypoplasia type 1A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>VSX2</i>	Microphthalmia with or without coloboma	AR	General Population	1 in 91	98%	1 in 4,501	1 in 1,638,364
<i>WHRN</i>	Usher syndrome type 2D	AR	General Population	1 in 282	99%	1 in 28,101	<1 in 10 million
<i>WRN</i>	Werner syndrome	AR	General Population	1 in 308	98%	1 in 15,351	<1 in 10 million
			Caucasian / European Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Japanese Population	1 in 71	98%	1 in 3,501	1 in 994,284
<i>XPA</i>	Xeroderma pigmentosum, group A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
<i>XPC</i>	Xeroderma pigmentosum, group C	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ZFYVE26</i>	Spastic paraplegia 15	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million

* For genes that have tested negative

† The carrier frequency for heterozygous alpha thalassemia carriers ($\alpha\alpha/\alpha-$) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis ($\alpha\alpha/-$) is 1 in 1000.

Abbreviations: AR, autosomal recessive; XL, X-linked