



Partner Information:  
**Not Tested**

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CAP#: 8042697  
CLIA#: 05D2043189  
Laboratory Director:  
Dr. Hanlin (Harry) Gao  
Report Date: **Feb 27, 2024**

Accession:  
**FT-7195888**

Test#: FT-TS14759365  
Specimen Type: Saliva Swab  
Collected: Not Provided

Accession:  
**N/A**

## FINAL RESULTS



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

## TEST PERFORMED

### Monash Beacon Expanded Male Carrier Screening Panel v2.1

(363 Gene Panel; gene sequencing  
with deletion and duplication analysis)

Condition and Gene	Inheritance		Partner
<b>Mitochondrial complex III deficiency</b> <i>BCS1L</i>	AR	 Carrier c.205C>T (p.Arg69Cys)	N/A

## INTERPRETATION:

### Notes and Recommendations:

- Based on these results, this individual is positive for a carrier mutation in 1 gene. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- 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. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- 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.
- Genetic counseling is recommended. Contact your physician about the available options for genetic counseling.



## MITOCHONDRIAL COMPLEX III DEFICIENCY

Patient		Partner
Result	⊕ Carrier	N/A
Variant Details	<b>BCS1L</b> (NM_004328.5) c.205C>T (p.Arg69Cys)	N/A

### *What is Mitochondrial complex III deficiency?*

The BCS1L gene is involved in the assembly of complex III of the mitochondrial respiratory chain. The BCS1L gene is associated with three conditions: generalized mitochondrial complex III deficiency, Björnstad syndrome, and GRACILE syndrome.

- Generalized mitochondrial complex III deficiency is a multi-system condition characterized by abnormalities of the brain, kidney, liver, heart, and muscles. This condition typically is severe and begins in infancy, but can be milder and/or later onset in rare cases.
- Björnstad syndrome is an extremely rare inherited disorder characterized by the presence of hair shafts that are abnormally flattened and twisted (pili torti). Individuals with this disorder may usually have dry, fragile, lusterless, and/or coarse scalp hair. In addition, most individuals with Björnstad syndrome experience deafness due to sensorineural hearing loss. When sensorineural deafness is present in such cases, it is congenital or within the first year of life. As an affected child ages, deafness may lead to speech difficulties. Underdevelopment of the ovaries of affected females or hypogonadism has occurred in association with Björnstad syndrome. Symptoms associated with this finding may include a delay in the development in secondary sexual characteristics.
- GRACILE syndrome is characterized by growth retardation, aminoaciduria, cholestasis, iron overload, lactic acidosis, and early death. This is a severe condition that begins before birth. Infants with this condition do not survive past the newborn period.

### *What is my risk of having an affected child?*

Mitochondrial complex III deficiency is inherited in an autosomal recessive manner. 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 effective treatments for any of these three conditions, which are often fatal in early childhood. Treatment is primarily supportive.

### *What mutation was detected?*

The detected heterozygous variant was NM\_004328.5:c.205C>T (p.Arg69Cys). This variant, p.Arg69Cys, has previously been reported as compound heterozygous in four unrelated individuals with BCS1L-related disorder, one of the patients also carried one truncating variant in NLGN4 gene (PubMed: [27959697](#), [31435670](#), [34662929](#)). Functional studies displayed that this variant impaired the respiratory capacity of the cell in vitro (PubMed: [31435670](#)). The laboratory classifies this variant as likely pathogenic.



## GENES TESTED:

### Monash Beacon Expanded Male Carrier Screening Panel v2.1 - 363 Genes

This analysis was run using the Monash Beacon Expanded Male Carrier Screening Panel v2.1 gene list. 363 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, ACADM, 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, CHRNG, 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, CYP1B1, 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, FKRP, FKTN, FOXRED1, FTCD, FUCA1, G6PC, GAA, GALC, GALNS, GALT, GAMT, GBA, GBE1, GCDH, GDAP1, GDF5, GFM1, GJB2, GJB6, GLB1, 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, MFSDB, 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, PROP1, PSAP, PTS, PUS1, QDPR, RAB23, RAG1, RAG2, RAPSN, RARS2, RAX, RDH12, RMRP, RNASEH2B, RPE65, RPGRIP1L, RTGL1, SACS, SAMD9, SAMHD1, SCO2, SEPS2, 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, TSM, 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, 99.60% and 99.54% 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

**CEP290:** Copy number analysis for exons 8-13 and exons 39-42 may have reduced sensitivity in the CEP290 gene. Confirmation of these exons are limited to individuals with a positive personal history of CEP290-related conditions and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **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 on 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. **DDX11:** Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in the DDX11 gene. **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:** Significant interference from highly homologous regions in exons 1-2 of the HBA1 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. **HBA2:** Significant interference from highly homologous regions in exons 1-2 of the HBA2 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. **HSD17B4:** Copy number analysis for exons 4-6 may have reduced sensitivity in the HSD17B4 gene. Confirmation of these exons are limited to individuals with a positive personal history of D-bifunctional protein deficiency and Perrault syndrome and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **LMBRD1:** Copy number analysis for exons 9-12 may have reduced sensitivity in the LMBRD1 gene. Confirmation of these exons are limited to individuals with a positive personal history of combined methylmalonic aciduria and homocystinuria and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **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.



**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 (including exons 4-5), copy number analysis may have reduced sensitivity for partial gene deletions/duplications of *TRDN*. Confirmation of 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. **TYR:** Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 4-5 of the TYR gene (NM\_000372.5). **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:

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**Yan Meng, Ph.D., CGMB, FACMG** on 2/27/2024  
Laboratory Director, Fulgent

## DISCLAIMER:

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This test was developed and its performance characteristics determined by **Fulgent Therapeutics, LLC**. 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](mailto: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>ACADM</i>	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	General Population	1 in 69	98%	1 in 3,401	1 in 938,676
			Caucasian / European Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			East Asian Population	1 in 198	99%	1 in 19,701	<1 in 10 million
			Native American Population	1 in 43	96%	1 in 1,051	1 in 180,772
<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



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ASPA	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
ASS1	Citruinemia	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
ATM	Ataxia-telangiectasia	AR	General Population	1 in 100	92%	1 in 1,239	1 in 495,600
ATP6V1B1	Renal tubular acidosis with deafness	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ATP7B	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
BBS1	Bardet-Biedl syndrome type 1	AR	General Population	1 in 367	99%	1 in 36,601	<1 in 10 million
BBS10	Bardet-Biedl syndrome type 10	AR	General Population	1 in 395	99%	1 in 39,401	<1 in 10 million
BBS12	Bardet-Biedl syndrome type 12	AR	General Population	1 in 791	99%	1 in 79,001	<1 in 10 million
BBS2	BBS2-related ciliopathies	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
BCKDHA	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 18,040
BCKDHB	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
BCS1L	Mitochondrial complex III deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BLM	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
BSND	Bartter syndrome type 4a	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CAPN3	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
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
CBS	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
CC2D2A	Joubert syndrome 9	AR	General Population	1 in 201	99%	1 in 20,001	1 in 16,080,804
CCDC103	Primary ciliary dyskinesia, type 17	AR	General Population	1 in 316	98%	1 in 15,751	<1 in 10 million
CCDC39	Primary ciliary dyskinesia, type 14	AR	General Population	1 in 211	98%	1 in 10,501	1 in 8,862,844
CCDC88C	Congenital hydrocephalus 1	AR	General Population	1 in 137	99%	1 in 13,601	1 in 7,453,348
CDH23	Usher syndrome, type 1D	AR	General Population	1 in 285	90%	1 in 2,841	1 in 11,364
CEP290	CEP290-related Ciliopathies	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CFTR	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
CHRNE	Congenital myasthenic syndrome	AR	General Population	1 in 408	99%	1 in 40,701	<1 in 10 million
CHRNA3	Multiple pterygium syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CHST6	Macular corneal dystrophy, CHST6-related	AR	General Population	1 in 79	99%	1 in 7,801	1 in 2,465,116
CIITA	Bare lymphocyte syndrome, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CLN3	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
CLN5	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
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General Population	<1 in 500	92%	1 in 6,239	<1 in 10 million
CLN8	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
CLRN1	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
CNGB3	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
COL27A1	Steel syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
COL4A3	Alport syndrome, COL4A3-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
			Ashkenazi Jewish Population	1 in 188	98%	1 in 9,351	1 in 7,031,952
COL4A4	Alport syndrome, COL4A4-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
COL7A1	Dystrophic epidermolysis bullosa	AR	General Population	1 in 196	97%	1 in 6,501	1 in 5,096,784
COX15	Mitochondrial complex IV deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>	CRB1-related retinopathy	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 methyloxidase 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 1,464,244 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>	Dihydropolipoamide 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 56	91%	1 in 612	1 in 137,113
<i>DUOXA2</i>	Congenital hypothyroidism, DUOXA2-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
<i>ERCC2</i>	ERCC2-related disorders	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>	ERCC6-related disorders	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 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





Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>	EVC-related bone growth disorders	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>	EVC2-related bone growth disorders	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>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 Population	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>	FKRP Alpha-dystroglycanopathies	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>FKTN</i>	FKTN Alpha-dystroglycanopathies	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>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
<i>GAA</i>	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
<i>GALC</i>	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
<i>GALNS</i>	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR	General Population	1 in 224	97%	1 in 7,434	1 in 6,660,864
<i>GALT</i>	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
<i>GAMT</i>	Guanidinoacetate methyltransferase deficiency	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
<i>GBA</i>	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
<i>GBE1</i>	Glycogen storage disease IV	AR	General Population	1 in 387	99%	1 in 38,601	<1 in 10 million
<i>GCDH</i>	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
<i>GDAP1</i>	Charcot-Marie-Tooth disease, GDAP1-related	AR	General Population	1 in 152	99%	1 in 15,101	1 in 9,181,408
<i>GDF5</i>	Du Pan Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GFM1</i>	Combined oxidative phosphorylation deficiency, GFM1-related	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>GJB2</i>	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
<i>GJB6</i>	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
<i>GLB1</i>	GLB1-related disorders	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 4,980,200
<i>GLDC</i>	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
<i>GLE1</i>	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
<i>GNE</i>	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
<i>GNPTAB</i>	Mucopolidosis II & III	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNPTG</i>	Mucopolidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNS</i>	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GSS</i>	Glutathione synthetase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>GUCY2D</i>	Leber congenital amaurosis 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GUSB</i>	Mucopolysaccharidosis type VII	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>HADHA</i>	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
<i>HADHB</i>	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
<i>HAX1</i>	Severe congenital neutropenia, HAX1-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>HBA1</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>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
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
			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>HBB</i>	Hemoglobin E thalassemia	AR	General Population	1 in 158
<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



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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 Caucasian / European Population	1 in 434 1 in 345	98% 98%	1 in 21,651 1 in 17,201	<1 in 10 million <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 Puerto Rican Population	1 in 354 1 in 21	98% 98%	1 in 17,651 1 in 1,001	<1 in 10 million 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>	Hydroletharus syndrome	AR	General Population Finnish Population	<1 in 500 1 in 50	98% 98%	1 in 24,951 1 in 2,451	<1 in 10 million 1 in 490,200
<i>IDUA</i>	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	General Population Caucasian / European Population	<1 in 500 1 in 153	95% 95%	1 in 9,981 1 in 3,041	<1 in 10 million 1 in 1,861,092
<i>IVD</i>	Isovaleric Acidemia	AR	General Population African/African American Population Caucasian / European Population East Asian Population	1 in 167 1 in 100 1 in 115 1 in 407	90% 90% 90% 90%	1 in 1,661 1 in 991 1 in 1,141 1 in 4,061	1 in 1,109,548 1 in 396,400 1 in 524,860 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>	KCNJ11-related hyperinsulinism	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
<i>LAMA2</i>	Muscular dystrophy, LAMA2-related	AR	General Population Caucasian / European Population	<1 in 500 1 in 125	99% 99%	1 in 49,901 1 in 12,401	<1 in 10 million 1 in 6,200,500
<i>LAMA3</i>	Junctional epidermolysis bullosa 2	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
<i>LDLRAP1</i>	Familial Hypercholesterolemia	AR	General Population Amish Population Caucasian / European Population French Canadian Population	1 in 8 1 in 2 1 in 7 1 in 8	99% 99% 99% 99%	1 in 701 1 in 101 1 in 601 1 in 701	1 in 22,432 1 in 808 1 in 16,828 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 Caucasian / European Population Iranian Jewish Population	1 in 211 1 in 161 1 in 32	99% 99% 99%	1 in 21,001 1 in 16,001 1 in 3,101	<1 in 10 million 1 in 4,973,248 1 in 396,928
<i>LMBRD1</i>	Methylmalonic aciduria and homocystinuria, cbIF 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 Ashkenazi Jewish Population	1 in 500 1 in 180	98% 98%	1 in 24,951 1 in 8,951	<1 in 10 million 1 in 6,444,720
<i>LPL</i>	Familial lipoprotein lipase deficiency	AR	General Population French Canadian Population	1 in 500 1 in 46	99% 99%	1 in 49,901 1 in 4,501	<1 in 10 million 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 Faroese Population French Canadian Population	1 in 447 1 in 21 1 in 22	98% 98% 98%	1 in 22,301 1 in 1,001 1 in 1,051	<1 in 10 million 1 in 84,084 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 Caucasian / European Population	1 in 354 1 in 274	99% 99%	1 in 35,301 1 in 27,301	<1 in 10 million <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>	Mucopolipidosis IV	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 100	99% 99%	1 in 29,901 1 in 9,901	<1 in 10 million 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 Bukharan/Kurdish Jewish Population	<1 in 500 1 in 20	99% 99%	1 in 49,901 1 in 1,901	<1 in 10 million 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



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>MKS1</i>	MKS1-related ciliopathies	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
<i>MLC1</i>	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population Libyan Jewish Population	<1 in 500 1 in 40	99% 99%	1 in 49,901 1 in 3,901	<1 in 10 million 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 Ashkenazi Jewish Population	1 in 102 1 in 55	98% 98%	1 in 5,051 1 in 2,701	1 in 2,060,808 1 in 594,220
<i>MPV17</i>	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR	General Population Native American Population	<1 in 500 1 in 20	96% 96%	1 in 12,476 1 in 476	<1 in 10 million 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 Ashkenazi Jewish Population	<1 in 500 1 in 180	98% 98%	1 in 24,951 1 in 8,951	<1 in 10 million 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>	Mevalonate kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MYO7A</i>	MYO7A-related disorders	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 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
<i>NAGLU</i>	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	General Population Caucasian / European Population East Asian Population	<1 in 500 1 in 346 1 in 298	99% 99% 99%	1 in 49,901 1 in 34,501 1 in 29,701	<1 in 10 million <1 in 10 million <1 in 10 million
<i>NAGS</i>	N-acetylglutamate synthase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>NBN</i>	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
<i>NDRG1</i>	Charcot-Marie-Tooth disease, type 4D	AR	General Population	1 in 22	98%	1 in 1,051	1 in 92,488
<i>NDUFAF2</i>	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>NDUFAF5</i>	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population Ashkenazi Jewish Population	1 in 447 1 in 290	98% 98%	1 in 22,301 1 in 14,451	<1 in 10 million <1 in 10 million
<i>NDUFS4</i>	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>NDUFS4</i>	Mitochondrial complex I deficiency	AR	General Population Hutterite Population	<1 in 500 1 in 27	99% 99%	1 in 49,901 1 in 2,601	<1 in 10 million 1 in 280,908
<i>NDUFS6</i>	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population Bukharan/Kurdish Jewish Population	<1 in 500 1 in 24	99% 99%	1 in 49,901 1 in 2,301	<1 in 10 million 1 in 220,896
<i>NDUFS7</i>	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>NDUFV1</i>	Mitochondrial complex I deficiency, nuclear type 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>NEB</i>	Nemaline myopathy	AR	General Population Amish Population Ashkenazi Jewish Population Finnish Population	1 in 112 1 in 11 1 in 108 1 in 112	98% 98% 98% 98%	1 in 5,551 1 in 501 1 in 5,351 1 in 5,551	1 in 2,486,848 1 in 22,044 1 in 2,311,632 1 in 2,486,848
<i>NEU1</i>	Sialidosis, type I and II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>NPC1</i>	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
<i>NPC2</i>	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
<i>NPHP1</i>	NPHP1-related ciliopathies	AR	General Population Finnish Population	1 in 480 1 in 124	98% 98%	1 in 23,951 1 in 6,151	<1 in 10 million 1 in 3,050,896
<i>NPHS1</i>	Congenital nephrotic syndrome, type 1	AR	General Population Finnish Population	1 in 289 1 in 50	98% 98%	1 in 14,401 1 in 2,451	<1 in 10 million 1 in 490,200
<i>NPHS2</i>	Congenital nephrotic syndrome, type 2	AR	General Population Finnish Population	1 in 289 1 in 50	98% 98%	1 in 14,401 1 in 2,451	<1 in 10 million 1 in 490,200
<i>NTRK1</i>	Congenital insensitivity to pain with anhidrosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>OAT</i>	Gyrate atrophy of choroid and retina	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>OCA2</i>	Oculocutaneous albinism type II	AR	General Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>OPA3</i>	Costeff syndrome	AR	General Population Iraqi Jewish Population	<1 in 500 1 in 50	98% 98%	1 in 24,951 1 in 2,451	<1 in 10 million 1 in 490,200



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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	PCDH15-related sensory loss	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
PEX1	Zellweger syndrome, PEX1-related	AR	General Population	1 in 147	95%	1 in 2,921	1 in 1,717,548
PEX10	Zellweger syndrome, PEX10-related	AR	General Population	1 in 500	95%	1 in 9,981	<1 in 10 million
			Japanese Population	1 in 354	95%	1 in 7,061	1 in 9,998,376
PEX12	Zellweger syndrome, PEX12-related	AR	General Population	1 in 373	95%	1 in 7,441	<1 in 10 million
PEX2	Zellweger syndrome, PEX2-related	AR	General Population	1 in 500	95%	1 in 9,981	<1 in 10 million
			Ashkenazi Jewish Population	1 in 123	95%	1 in 2,441	1 in 1,200,972
PEX26	Zellweger syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PEX6	Zellweger syndrome, PEX6-related	AR	General Population	1 in 280	99%	1 in 27,901	<1 in 10 million
			Yemenite Jewish Population	1 in 18	99%	1 in 1,701	1 in 122,472
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
PFKM	Glycogen storage disease VII	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 120	99%	1 in 11,901	1 in 5,712,480
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 280	98%	1 in 13,951	<1 in 10 million
PHYH	Refsum disease	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PKHD1	Polycystic kidney disease, PKHD1-related	AR	General Population	1 in 70	98%	1 in 3,451	1 in 966,280
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 2,268,828
PLA2G6	Infantile neuroaxonal dystrophy	AR	General Population	1 in 500	97%	1 in 16,634	<1 in 10 million
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
PMM2	PMM2-glycosylation disorders	AR	General Population	1 in 63	99%	1 in 6,201	1 in 1,562,652
			Ashkenazi Jewish Population	1 in 57	99%	1 in 5,601	1 in 1,277,028
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 1,988,284
POLG	POLG-related disorders	AR	General Population	1 in 113	99%	1 in 11,201	1 in 5,062,852
POLR1C	POLR1C-related disorders	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR	General Population	1 in 462	98%	1 in 23,051	<1 in 10 million
			Finnish Population	1 in 111	98%	1 in 5,501	1 in 2,442,444
POMT1	POMT1 Alpha-dystroglycanopathies	AR	General Population	1 in 290	99%	1 in 28,901	<1 in 10 million
POMT2	POMT2 Alpha-dystroglycanopathies	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
POR	Antley-Bixler syndrome	AR	General Population	1 in 159	98%	1 in 7,901	1 in 5,025,036
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General Population	1 in 368	98%	1 in 18,351	<1 in 10 million
			Caucasian / European Population	1 in 488	98%	1 in 24,351	<1 in 10 million
			Finnish Population	1 in 75	98%	1 in 3,701	1 in 1,110,300
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR	General Population	1 in 149	99%	1 in 14,801	1 in 8,821,396
PROP1	Combined pituitary hormone deficiency 2	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PTS	Tetrahydrobiopterin deficiency	AR	General Population	1 in 354	96%	1 in 8,826	<1 in 10 million
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAB23	Carpenter syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAG1	Omenn syndrome, RAG1-related	AR	General Population	1 in 290	98%	1 in 14,451	1 in 16,763,160



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>	RAPSN-associated acetylcholine receptor deficiency	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	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 456	98%	1 in 22,751	<1 in 10 million
<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 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 21,601	1 in 18,749,668
<i>RPE65</i>	RPE65-related retinopathy	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
<i>RPGRIP1L</i>	RPGRIP1L-related ciliopathies	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	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
<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
			Ashkenazi Jewish Population	1 in 106	99%	1 in 10,501	1 in 4,452,424
<i>SLC22A5</i>	Systemic primary carnitine deficiency	AR	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
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 1,020,204
<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>	SLC26A2-related disorders	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>SLC26A3</i>	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
<i>SLC35A3</i>	Arthrogyposis, 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
<i>SLC37A4</i>	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
<i>SLC39A4</i>	Acrodermatitis enteropathica	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*
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 immunoosseous 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 22,301	<1 in 10 million
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	TCTN2-related ciliopathies	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
TECP2	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 500	98%	1 in 24,951	<1 in 10 million
TMEM216	TMEM216-related ciliopathies	AR	General Population	1 in 141	98%	1 in 7,001	1 in 3,948,564
TPO	Thyroid dysmorphogenesis, TPO-related	AR	Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
			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	TRIM32-related disorders	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
TRMU	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
TSEN54	Pontocerebellar hypoplasia type 2A	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TSFM	Combined oxidative phosphorylation deficiency, TSFM-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
TSHB	Congenital hypothyroidism, TSHB-related	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
TTC37	Trichohepatoenteric syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
TTPA	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
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TYR	Oculocutaneous albinism types 1A and 1B	AR	General Population	1 in 20	99%	1 in 1,901	1 in 152,080
TYRP1	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
UGT1A1	Crigler-Najjar syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
USH1C	USH1C-related disorders	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
USH1G	Usher syndrome type IG	AR	General Population	1 in 434	99%	1 in 43,301	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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