



Donor Q001J

FINAL RESULTS



Carrier for genetic conditions in **multiple** genes.  
Genetic counseling is recommended.

TEST PERFORMED

**Custom Sonic Beacon Male  
Carrier Screening**

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

**Condition and Gene**

**Inheritance**

**Leber congenital amaurosis 8**  
*CRB1*

AR

⊕ Carrier N/A  
c.3862G>A (p.Gly1288Ser)

**Short branched chain acyl-CoA dehydrogenase  
(SBCAD) deficiency**  
*ACADSB*

AR

⊕ Carrier N/A  
c.655G>A (p.Val219Met)

INTERPRETATION:

**Notes and Recommendations:**

- Based on these results, this individual is positive for carrier mutations in 2 genes. The risk estimates below are quantified based on general population carrier frequencies. Carrier screening for the reproductive partner is recommended to accurately assess this risk:
  - There is a 1/416 chance of having a child affected with Leber congenital amaurosis 8, a *CRB1*-related condition.
  - There is a 1/1472 chance of having a child affected with Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency, an *ACADSB*-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 the 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.
- 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 8

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<b>Result</b>	<b>+</b> Carrier	N/A
<b>Variant Details</b>	<b>CRB1</b> (NM_201253.3) c.3862G>A (p.Gly1288Ser)	N/A

### *What is Leber congenital amaurosis 8?*

Leber congenital amaurosis (LCA) is an inherited retinal degenerative disease characterized by severe loss of vision at birth. Abnormal eye movements, light sensitivity, and deep-set eyes may also be present. The signs and symptoms may become apparent initially by a specific behavior called Franceschetti's oculo-digital sign consisting of poking, pressing, and rubbing of the eyes. In rare cases, developmental delay and intellectual disability have also been reported. There are many different types of LCA marked by their genetic cause, patterns of vision loss, and related eye abnormalities.

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

Leber congenital amaurosis 8 is inherited in an autosomal recessive manner. The risk for being a carrier for a CRB1-related condition is 1/104. 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 currently no treatments for most types of LCA. Low vision aids to maximize visual function can be very useful for patients with LCA. Educational programs and support agencies for the visually impaired are often helpful.

### *What mutation was detected?*

The detected heterozygous variant was NM\_201253.3:c.3862G>A (p.Gly1288Ser). This missense variant, p.Gly1288Ser, has been reported in the compound heterozygous state in at least four unrelated individuals with retinal degeneration or early onset severe retinal dystrophy (EOSRD) (PubMed: [33342761](#), [31630094](#)). This variant has been reported to segregate with disease in at least one family with retinal degeneration (PubMed: [33342761](#)). The laboratory classifies this variant as pathogenic.



## SHORT BRANCHED CHAIN ACYL-COA DEHYDROGENASE (SBCAD) DEFICIENCY

### Patient

<b>Result</b>	⊕ Carrier	N/A
<b>Variant Details</b>	<b>ACADSB</b> (NM_001609.4) c.655G>A (p.Val219Met)	N/A

### *What is Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency?*

Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency is a condition that prevents the body from converting certain amino acids to energy. Most people with SBCAD deficiency have no health problems related to the disorder and show normal development. Very few symptomatic patients have been reported. Signs and symptoms typically appear during infancy or early childhood and can include microcephaly, apneic episodes in infancy, low muscle tone, lethargy, seizures, hypoglycemia, and developmental delay.

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

Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency is inherited in an autosomal recessive manner. This means that when both parents are carriers for the same condition, there is a 25% (1 in 4) risk of having an affected child. The carrier frequency of SBCAD is estimated at 1 in 368 in the general population, and 1 in 6 in the Hmong population.

### *What kind of medical management is available?*

To date, most patients identified by newborn screening with short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency are of Hmong descent and remain asymptomatic. Signs and symptoms may be triggered by infections, prolonged periods without food (fasting), or an increased amount of protein-rich foods in the diet. Doctor might recommend special baby formulas and foods designed for children with SBCAD. L-carnitine supplements might be prescribed.

### *What mutation was detected?*

The detected heterozygous variant was NM\_001609.4:c.655G>A (p.Val219Met). This missense variant, p.Val219Met, has been reported in the compound heterozygous state in 3 unrelated individuals with short/Branched Chain Acyl-CoA Dehydrogenase Deficiency with elevated 2-methylbutyrylglycine levels (PubMed: [31555323](#)). The laboratory classifies this variant as likely pathogenic.



**GENES TESTED:**

**Custom Sonic Beacon Male Carrier Screening - 351 Genes**

351 genes tested (99.52% at >20x). For more gene specific information and assistance with residual risk calculation, see SUPPLEMENTAL TABLE.

<i>ABCB11</i>	<i>ABCC8</i>	<i>ABCD4</i>	<i>ACAD9</i>	<i>ACADM</i>	<i>ACADS</i>
<i>ACADSB</i>	<i>ACADVL</i>	<i>ACAT1</i>	<i>ACOX1</i>	<i>ACSF3</i>	<i>ADA</i>
<i>ADAMTS2</i>	<i>ADGRG1</i>	<i>ADK</i>	<i>AGA</i>	<i>AGL</i>	<i>AGPS</i>
<i>AGXT</i>	<i>AHCY</i>	<i>AHI1</i>	<i>AIP1</i>	<i>AIRE</i>	<i>ALDH3A2</i>
<i>ALDH4A1</i>	<i>ALDOB</i>	<i>ALG6</i>	<i>ALMS1</i>	<i>ALPL</i>	<i>AMT</i>
<i>AQP2</i>	<i>ARG1</i>	<i>ARL13B</i>	<i>ARSA</i>	<i>ARSB</i>	<i>ASL</i>
<i>ASNS</i>	<i>ASPA</i>	<i>ASS1</i>	<i>ATM</i>	<i>ATP6V1B1</i>	<i>ATP7B</i>
<i>BBS1</i>	<i>BBS10</i>	<i>BBS12</i>	<i>BBS2</i>	<i>BCKDHA</i>	<i>BCKDHB</i>
<i>BCS1L</i>	<i>BLM</i>	<i>BSND</i>	<i>BTD</i>	<i>CAPN3</i>	<i>CASQ2</i>
<i>CBS</i>	<i>CCDC103</i>	<i>CCDC151</i>	<i>CCDC39</i>	<i>CDH23</i>	<i>CEP290</i>
<i>CERKL</i>	<i>CFTR</i>	<i>CHRNE</i>	<i>CHRNA3</i>	<i>CHST6</i>	<i>CIITA</i>
<i>CLN3</i>	<i>CLN5</i>	<i>CLN6</i>	<i>CLN8</i>	<i>CLRN1</i>	<i>CNGA1</i>
<i>CNGB1</i>	<i>CNGB3</i>	<i>COL27A1</i>	<i>COL4A3</i>	<i>COL4A4</i>	<i>COL7A1</i>
<i>CPS1</i>	<i>CPT1A</i>	<i>CPT2</i>	<i>CRB1</i>	<i>CRYL1</i>	<i>CTNS</i>
<i>CTSK</i>	<i>CYBA</i>	<i>CYP11B1</i>	<i>CYP11B2</i>	<i>CYP17A1</i>	<i>CYP19A1</i>
<i>CYP11B1</i>	<i>CYP21A2</i>	<i>CYP27A1</i>	<i>DBT</i>	<i>DCLRE1C</i>	<i>DHCR7</i>
<i>DHDDS</i>	<i>DLD</i>	<i>DNAH5</i>	<i>DNAI1</i>	<i>DNAI2</i>	<i>DNAL1</i>
<i>DPYD</i>	<i>DUOX2</i>	<i>DUOXA2</i>	<i>DYSF</i>	<i>EIF2AK3</i>	<i>EIF2B5</i>
<i>ELP1</i>	<i>ERCC6</i>	<i>ERCC8</i>	<i>ESCO2</i>	<i>ETFA</i>	<i>ETFB</i>
<i>ETFDH</i>	<i>ETHE1</i>	<i>EVC</i>	<i>EVC2</i>	<i>EXOSC3</i>	<i>EYS</i>
<i>F11</i>	<i>FAH</i>	<i>FAM161A</i>	<i>FANCA</i>	<i>FANCC</i>	<i>FANCG</i>
<i>FH</i>	<i>FKRP</i>	<i>FKTN</i>	<i>FTCD</i>	<i>G6PC</i>	<i>GAA</i>
<i>GALC</i>	<i>GALE</i>	<i>GALK1</i>	<i>GALNS</i>	<i>GALT</i>	<i>GAMT</i>
<i>GBA</i>	<i>GBE1</i>	<i>GCDH</i>	<i>GDAP1</i>	<i>GFM1</i>	<i>GJB2</i>
<i>GJB6</i>	<i>GLB1</i>	<i>GLDC</i>	<i>GLE1</i>	<i>GNE</i>	<i>GNPTAB</i>
<i>GNPTG</i>	<i>GNRHR</i>	<i>GNS</i>	<i>GP1BA</i>	<i>GP9</i>	<i>GRHRP</i>
<i>GUSB</i>	<i>HADHA</i>	<i>HAX1</i>	<i>HBA1</i>	<i>HBA2</i>	<i>HBB</i>
<i>HEXA</i>	<i>HEXB</i>	<i>HGD</i>	<i>HGSNAT</i>	<i>HJV</i>	<i>HLCS</i>
<i>HMGCL</i>	<i>HOGA1</i>	<i>HPS1</i>	<i>HPS3</i>	<i>HSD17B4</i>	<i>HSD3B2</i>
<i>HYAL1</i>	<i>HYLS1</i>	<i>IDH3B</i>	<i>IDUA</i>	<i>IVD</i>	<i>IYD</i>
<i>JAK3</i>	<i>KCNJ11</i>	<i>LAMA2</i>	<i>LAMA3</i>	<i>LAMB3</i>	<i>LAMC2</i>
<i>LCA5</i>	<i>LHX3</i>	<i>LIFR</i>	<i>LIPA</i>	<i>LMBRD1</i>	<i>LOXHD1</i>
<i>LPL</i>	<i>LRPPRC</i>	<i>LYST</i>	<i>MAN2B1</i>	<i>MCCC1</i>	<i>MCCC2</i>
<i>MCEE</i>	<i>MCOLN1</i>	<i>MED17</i>	<i>MESP2</i>	<i>MFSD8</i>	<i>MKS1</i>
<i>MLC1</i>	<i>MMAA</i>	<i>MMAB</i>	<i>MMACHC</i>	<i>MMADHC</i>	<i>MPI</i>
<i>MPL</i>	<i>MPV17</i>	<i>MTMR2</i>	<i>MTRR</i>	<i>MTTP</i>	<i>MUT</i>
<i>MVK</i>	<i>MYO7A</i>	<i>NAGLU</i>	<i>NAGS</i>	<i>NBN</i>	<i>NDRG1</i>
<i>NDUFAF5</i>	<i>NDUFS6</i>	<i>NEB</i>	<i>NPC1</i>	<i>NPC2</i>	<i>NPHP1</i>
<i>NPHS1</i>	<i>NPHS2</i>	<i>NR2E3</i>	<i>NTRK1</i>	<i>OAT</i>	<i>OPA3</i>
<i>OTOF</i>	<i>P3H1</i>	<i>PAH</i>	<i>PANK2</i>	<i>PC</i>	<i>PCBD1</i>
<i>PCCA</i>	<i>PCCB</i>	<i>PCDH15</i>	<i>PDE6A</i>	<i>PDHB</i>	<i>PEX1</i>
<i>PEX10</i>	<i>PEX12</i>	<i>PEX2</i>	<i>PEX6</i>	<i>PEX7</i>	<i>PFKM</i>
<i>PGK1</i>	<i>PHGDH</i>	<i>PKHD1</i>	<i>PLA2G6</i>	<i>PLOD1</i>	<i>PMM2</i>
<i>POLG</i>	<i>POLR1C</i>	<i>POMGNT1</i>	<i>POMT1</i>	<i>POMT2</i>	<i>PPT1</i>
<i>PQBP1</i>	<i>PROP1</i>	<i>PSAP</i>	<i>PTS</i>	<i>PUS1</i>	<i>PYGM</i>
<i>QDPR</i>	<i>RAB23</i>	<i>RAG1</i>	<i>RAG2</i>	<i>RAPSN</i>	<i>RARS2</i>
<i>RAX</i>	<i>RDH12</i>	<i>RMRP</i>	<i>RPE65</i>	<i>RPGRIP1L</i>	<i>RTEL1</i>
<i>SACS</i>	<i>SAMHD1</i>	<i>SEPSECS</i>	<i>SERPINA1</i>	<i>SGCA</i>	<i>SGCB</i>





<i>SGCD</i>	<i>SGCG</i>	<i>SGSH</i>	<i>SH3TC2</i>	<i>SLC12A3</i>	<i>SLC12A6</i>
<i>SLC16A2</i>	<i>SLC17A5</i>	<i>SLC22A5</i>	<i>SLC25A13</i>	<i>SLC25A15</i>	<i>SLC25A20</i>
<i>SLC26A2</i>	<i>SLC26A3</i>	<i>SLC26A4</i>	<i>SLC35A3</i>	<i>SLC37A4</i>	<i>SLC39A4</i>
<i>SLC46A1</i>	<i>SLC4A11</i>	<i>SLC5A5</i>	<i>SLC6A19</i>	<i>SLC7A7</i>	<i>SMARCA1</i>
<i>SMN1</i>	<i>SMPD1</i>	<i>SPG11</i>	<i>SPG7</i>	<i>STAR</i>	<i>SUMF1</i>
<i>SURF1</i>	<i>TAT</i>	<i>TCIRG1</i>	<i>TECPR2</i>	<i>TFR2</i>	<i>TG</i>
<i>TGM1</i>	<i>TH</i>	<i>TMEM216</i>	<i>TPO</i>	<i>TPP1</i>	<i>TRDN</i>
<i>TRIM32</i>	<i>TRMU</i>	<i>TSFM</i>	<i>TSHB</i>	<i>TTC37</i>	<i>TTPA</i>
<i>TYMP</i>	<i>UGT1A1</i>	<i>USH1C</i>	<i>USH1G</i>	<i>USH2A</i>	<i>VPS13A</i>
<i>VPS13B</i>	<i>VPS45</i>	<i>VRK1</i>	<i>VSX2</i>	<i>WHRN</i>	<i>WNT10A</i>
<i>XPA</i>	<i>XPC</i>	<i>ZFYVE26</i>			

## 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, 99.61% and 99.52% 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. New York patients: diagnostic findings are confirmed by Sanger, MLPA, or qPCR; exception SNV variants in genes for which confirmation of NGS results has been performed  $\geq 10$  times may not be confirmed if identified with high quality by NGS. 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

**BTB**: If detected, the variant NM\_001370658.1:c.1270G>C (p.Asp424His) will not be reported as this variant is associated with low disease penetrance and is primarily associated with reduced enzyme activity when homozygous. **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 (CJB6, 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. The variants c.188A>T (p.His63Leu), c.844G>T (p.Val282Leu), c.1174G>A (p.Ala392Thr), and c.1360C>T (p.Pro454Ser) in CYP21A2 will not be routinely reported as these variants are primarily associated with non-classic congenital adrenal hyperplasia and low disease penetrance. Additionally, the variant c.955C>T (p.Gln319Ter) is in the region with pseudogene interference, and the probability of this variant occurring in the real gene is greater than 50%. When observed, this variant will be reported as a possible carrier without LR-PCR. The confirmation test is recommended if the second reproductive partner is tests positive for variants in CYP21A2. **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. **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. **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 variants NM\_004004.6:c.109G>A (p.Val37Ile) and c.1096G>A (p.Glu366Lys) will not be reported as these variants are associated with low disease penetrance and are 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 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. **WNT10A**: If detected, certain common variants which are associated with autosomal dominant selective tooth agenesis are not reported. These variants are associated with low penetrance for autosomal recessive disease and are commonly found as homozygous in healthy controls.

SIGNATURE:

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A handwritten signature in black ink, appearing to read "Zhenbin Chen".

**Zhenbin Chen, Ph.D., CGMBS, FACMG** on 8/12/2022 10:34 PM PDT  
Electronically signed

#### DISCLAIMER:

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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>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>ABCD4</i>	Methylmalonic aciduria and homocystinuria, cblJ type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<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>ACADS</i>	Short-chain acyl-coA dehydrogenase (SCAD) deficiency	AR	General Population	1 in 85	99%	1 in 8,401	1 in 2,856,340
			African/African American Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			Caucasian / European Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
			Middle-Eastern Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 1,020,204
<i>ACADSB</i>	Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR	General Population	1 in 368	99%	1 in 36,701	<1 in 10 million
			Hmong Population	1 in 6	99%	1 in 501	<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
			General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<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
			General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<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>AHCY</i>	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR	General Population	<1 in 500	99%	1 in 49,901	<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>AIRE</i>	Autoimmune polyendocrinopathy syndrome type I	AR	General Population	1 in 150	98%	1 in 7,451	1 in 4,470,600
			Finnish Population	1 in 79	98%	1 in 3,901	1 in 1,232,716
<i>ALDH3A2</i>	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>ALDH4A1</i>	Hyperprolinemia type II	AR	General Population	<1 in 500	99%	1 in 49,901	<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
			Menonite Population	1 in 25	95%	1 in 481	1 in 48,100
			General Population	1 in 373	98%	1 in 18,601	<1 in 10 million
<i>AMT</i>	Glycine encephalopathy	AR	Finnish Population	1 in 117	98%	1 in 5,801	1 in 2,714,868
			General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<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





Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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
<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 18,040
<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>BTD</i>	Biotinidase deficiency	AR	General Population	1 in 124	99%	1 in 12,301	1 in 6,101,296
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 1,988,284
			Latino Population	1 in 136	99%	1 in 13,501	1 in 7,344,544
			Middle-Eastern Population	1 in 55	99%	1 in 5,401	1 in 1,188,220
<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, CASQ2-related	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>CCDC103</i>	Primary ciliary dyskinesia, type 17	AR	General Population	1 in 316	98%	1 in 15,751	<1 in 10 million
<i>CCDC151</i>	Primary ciliary dyskinesia, type 30	AR	General Population	1 in 365	98%	1 in 18,201	<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>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>CERKL</i>	Retinitis pigmentosa 26	AR	General Population	1 in 148	98%	1 in 7,351	1 in 4,351,792



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>CHRNA3</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, CLN5-related	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>CNGA1</i>	Retinitis Pigmentosa, CNGA1-related	AR	General Population	1 in 210	99%	1 in 20,901	<1 in 10 million
<i>CNGB1</i>	Retinitis Pigmentosa, CNGB1-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
<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
<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	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
<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>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	1 in 354	90%	1 in 3,531	1 in 4,999,896
			Hutterite Population	1 in 16	90%	1 in 151	1 in 9,664
<i>CPT2</i>	Carnitine palmitoyltransferase II deficiency	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Ashkenazi Jewish Population	1 in 51	95%	1 in 1,001	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	1 in 158	99%	1 in 15,701	1 in 9,923,032
			British Population	1 in 81	99%	1 in 8,001	1 in 2,592,324
			Moroccan Jewish Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<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>CYP11B1</i>	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
			Moroccan Jewish Population	1 in 35	98%	1 in 1,701	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>CYP19A1</i>	Aromatase 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	1 in 61	99%	1 in 6,001	1 in 1,464,244
			Inuit Population	1 in 9	99%	1 in 801	1 in 28,836
			Middle-Eastern Population	1 in 35	99%	1 in 3,401	1 in 476,140
<i>CYP27A1</i>	Cerebrotendinous xanthomatosis	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Moroccan Jewish Population	1 in 5	98%	1 in 201	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>DHCR7</i>	Smith-Lemli-Opitz syndrome	AR	General Population	1 in 30	96%	1 in 726	1 in 87,120
			African/African American Population	1 in 138	96%	1 in 3,426	1 in 1,891,152
			Ashkenazi Jewish Population	1 in 36	96%	1 in 876	1 in 126,144



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>DHDDS</i>	Retinitis pigmentosa 59	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
			Ashkenazi Jewish Population	1 in 118	98%	1 in 5,851	1 in 2,761,672
<i>DLD</i>	Dihydroipoamide dehydrogenase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 2,268,828
<i>DNAH5</i>	Primary ciliary dyskinesia, DNAH5-related	AR	General Population	1 in 142	98%	1 in 7,051	1 in 4,004,968
			Ashkenazi Jewish Population	1 in 113	99%	1 in 11,201	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>DNAL1</i>	Primary ciliary dyskinesia, DNAL1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>DPYD</i>	Dihydropyrimidine dehydrogenase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<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>DUOXA2</i>	Congenital hypothyroidism, DUOXA2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>DYSF</i>	Limb-girdle muscular dystrophy type 2B	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Japanese Population	1 in 332	95%	1 in 6,621	1 in 8,792,688
			Libyan Jewish Population	1 in 18	95%	1 in 341	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	98%	1 in 24,951	<1 in 10 million
<i>ELP1</i>	Familial Dysautonomia	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 31	99%	1 in 3,001	1 in 372,124
<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>EYS</i>	Retinitis pigmentosa 25	AR	General Population	1 in 66	98%	1 in 3,251	1 in 858,264
<i>F11</i>	Factor XI deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 11	98%	1 in 501	1 in 22,044
<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>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>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





Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>FTCD</i>	Glutamate formiminotransferase deficiency	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>GALE</i>	Galactose epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>GALK1</i>	Galactokinase deficiency	AR	General Population	1 in 110	95%	1 in 2,181	1 in 959,640
			Irish Population	1 in 64	95%	1 in 1,261	1 in 322,816
<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
			General Population	1 in 387	99%	1 in 38,601	<1 in 10 million
<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>GFM1</i>	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GJB2</i>	Nonsyndromic hearing loss, GJB2-related	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>	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
<i>GLB1</i>	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
<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
<i>GNPTAB</i>	Mucopolysaccharidosis II alpha/beta	AR	General Population	1 in 11	99%	1 in 1,001	1 in 44,044
			Iranian Jewish Population	1 in 11	99%	1 in 1,001	1 in 44,044
<i>GNPTAB</i>	Mucopolysaccharidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNPTAB</i>	Mucopolysaccharidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNPTG</i>	Mucopolysaccharidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNRHR</i>	Hypogonadotropic hypogonadism, GNRHR-related	AR	General Population	1 in 347	99%	1 in 34,601	<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>GP1BA</i>	Bernard-Soulier syndrome type A1	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GP9</i>	Bernard-Soulier syndrome type C	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GRHPR</i>	Primary hyperoxaluria type II	AR	General Population	<1 in 500	99%	1 in 49,901	<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



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>HADHA</i>	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
<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 18	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 1000	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 18	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 1000	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	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>HEXA</i>	Tay-Sachs disease	AR	General Population	1 in 300
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
General Population	1 in 600	98%				1 in 29,951	<1 in 10 million
<i>HEXB</i>	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million
<i>HGD</i>	Alkaptonuria	AR	General Population	1 in 250	90%	1 in 2,491	1 in 2,491,000
<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>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>HYAL1</i>	Mucopolysaccharidosis type IX	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HYLS1</i>	Hydrolethalus 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>IDH3B</i>	Retinitis pigmentosa, IDH3B-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
<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



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>IYD</i>	Thyroid dys-hormonogenesis, 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
<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, LOXHD1-related	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>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>MCCC1</i>	3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	AR	General Population	1 in 95	98%	1 in 4,701	1 in 1,786,380
<i>MCCC2</i>	3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	AR	General Population	1 in 95	98%	1 in 4,701	1 in 1,786,380
<i>MCEE</i>	Methylmalonyl-CoA epimerase deficiency	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>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>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



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
MPV17	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
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MTTP	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
MUT	Methylmalonic acidemia, MUT-related	AR	General Population	1 in 195	96%	1 in 4,851	1 in 3,783,780
			East Asian Population	1 in 53	96%	1 in 1,301	1 in 275,812
			Middle-Eastern Population	1 in 52	96%	1 in 1,276	1 in 265,408
			General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
MVK	Hyperimmunoglobulinemia D syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MVK	Mevalonate kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MYO7A	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
MYO7A	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
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
NDUFA5	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
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
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
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
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
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
NR2E3	Retinitis pigmentosa 37	AR	General Population	1 in 209	98%	1 in 10,401	1 in 8,695,236
NR2E3	Enhanced S-cone syndrome	AR	General Population	1 in 209	98%	1 in 10,401	1 in 8,695,236
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
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





Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>PC</i>	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
<i>PCBD1</i>	Tetrahydrobiopterin deficiency, PCBD1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>PCCA</i>	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
<i>PCCB</i>	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
<i>PCDH15</i>	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
<i>PCDH15</i>	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
<i>PDE6A</i>	Retinitis pigmentosa, PDE6A-related	AR	General Population	1 in 133	99%	1 in 13,201	1 in 7,022,932
<i>PDHB</i>	Pyruvate dehydrogenase E1-beta deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<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	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
<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	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
<i>PEX6</i>	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
<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	<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
<i>PGK1</i>	Phosphoglycerate kinase 1 deficiency	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>PHGDH</i>	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
<i>PKHD1</i>	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
<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	<1 in 500	99%	1 in 49,901	<1 in 10 million
			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
<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	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
<i>POMGNT1</i>	Retinitis pigmentosa 76	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
<i>POMT1</i>	Dystroglycanopathy, POMT1-related	AR	General Population	1 in 290	99%	1 in 28,901	<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>PPT1</i>	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
<i>PQBP1</i>	Renpenning syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<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>PYGM</i>	Glycogen storage disease type V	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 206	99%	1 in 20,501	<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



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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 137	98%	1 in 6,801	1 in 3,726,948
<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	<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>	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>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>SAMHD1</i>	Aicardi-Goutieres syndrome	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<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>SLC12A3</i>	Gitelman syndrome	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
<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>SLC16A2</i>	Allan-Herndon-Dudley syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<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>SLC22A5</i>	Systemic primary carnitine deficiency	AR	General Population	1 in 129	76%	1 in 534	1 in 275,544
			African/African American Population	1 in 86	76%	1 in 355	1 in 122,120
			East Asian Population	1 in 77	76%	1 in 318	1 in 97,944
			Faroese Population	1 in 9	76%	1 in 34	1 in 1,224
			Pacific Islander Population	1 in 37	76%	1 in 151	1 in 22,348
			South Asian/Indian Population	1 in 51	76%	1 in 209	1 in 42,636



Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>SLC25A20</i>	Carnitine-acylcarnitine translocase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<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
<i>SLC26A2</i>	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
<i>SLC26A2</i>	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
<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>SLC26A4</i>	Pendred syndrome	AR	General Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
			African/African American Population	1 in 76	98%	1 in 3,751	1 in 1,140,304
			Caucasian / European Population	1 in 88	98%	1 in 4,351	1 in 1,531,552
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 1,080,696
<i>SLC35A3</i>	Arthrogyroposis, intellectual disability, and seizures	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SLC37A4</i>	Glycogen storage disease, type Ib	AR	Ashkenazi Jewish Population	1 in 453	98%	1 in 22,601	<1 in 10 million
			General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
<i>SLC39A4</i>	Acrodermatitis enteropathica	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SLC46A1</i>	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
<i>SLC4A11</i>	Corneal endothelial dystrophy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SLC5A5</i>	Thyroid dysmorphogenesis, SLC5A5-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>SLC6A19</i>	Hartnup disorder	AR	General Population	1 in 87	99%	1 in 8,601	1 in 2,993,148
<i>SLC7A7</i>	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
<i>SMARCAL1</i>	Schimke immunosseous dysplasia	AR	General Population	1 in 500	90%	1 in 4,991	1 in 9,982,000
<i>SMN1</i>	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
<i>SMPD1</i>	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
<i>SPG11</i>	SPG11-related Neuromuscular Disorders	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
<i>SPG7</i>	Spastic paraplegia type 7	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
<i>STAR</i>	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SUMF1</i>	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
<i>SURF1</i>	Charcot-Marie-Tooth disease, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>SURF1</i>	Leigh syndrome, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>TAT</i>	Tyrosinemia, type II	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>TCIRG1</i>	Osteopetrosis, TCIRG1-related	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>TECPR2</i>	Spastic paraplegia 49	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>TFR2</i>	Hemochromatosis, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>TG</i>	Thyroid dysmorphogenesis, TG-related	AR	General Population	1 in 241	99%	1 in 24,001	<1 in 10 million
<i>TGM1</i>	Congenital ichthyosis	AR	General Population	1 in 224	95%	1 in 4,461	1 in 3,997,056
<i>TH</i>	Segawa syndrome	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>TMEM216</i>	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
<i>TMEM216</i>	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





Supplemental Table							
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>TPO</i>	Thyroid dysmorphogenesis, TPO-related	AR	General Population	1 in 373	99%	1 in 37,201	<1 in 10 million
<i>TPP1</i>	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
<i>TRDN</i>	Catecholaminergic polymorphic ventricular tachycardia, TRDN-related	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
<i>TRIM32</i>	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
<i>TRIM32</i>	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
<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>TSFM</i>	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
<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>TPPA</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>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>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>WNT10A</i>	Schopf-Schulz-Passarge syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>WNT10A</i>	Odontoonychodermal dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<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 alpha thalassemia trait cis is described in rows marked with a dagger symbol.

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