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Patient Information:
[redacted]

Partner Information:
Not Tested

Physician:
Strickland, Sophie
ATTN: Strickland, Sophie
Repromed
180 Fullarton Road
Dulwich, SA 5065 AU
Phone: 03 9420 8286

Laboratory:
Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Jan 03,2024**

Accession:
FT-6912635
Test#: FT-TS14725702
Specimen Type: Saliva Swab
Collected: Dec 06,2023

Accession:
N/A

FINAL RESULTS

Carrier for genetic conditions in **multiple** genes.
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	[redacted]	Partner
Neuronal ceroid lipofuscinosis, MFSD8-related *MFSD8*	AR	Carrier c.1436G>A (p.Trp479*)	N/A
Cystic Fibrosis *CFTR*	AR	Carrier c.1521_1523del (p.Phe508del)	N/A
Short-rib thoracic dysplasia 3 with or without polydactyly *DYNC2H1*	AR	Carrier c.10626+1G>T (p.?)	N/A
Congenital hypothyroidism, DUOX2-related *DUOX2*	AR	Carrier c.2895_2898del (p.Phe966Serfs*29)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for carrier mutations in 4 genes. The risk estimates below are quantified based on general population carrier frequencies. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions:
  - There is a 1/2000 chance of having a child affected with Neuronal ceroid lipofuscinosis, MFSD8-related, a *MFSD8*-related condition.
  - There is a 1/128 chance of having a child affected with Cystic Fibrosis, a *CFTR*-related condition.
  - There is a 1/272 chance of having a child affected with Short-rib thoracic dysplasia 3 with or without polydactyly, a *DYNC2H1*-related condition.
  - There is a 1/1464 chance of having a child affected with Congenital hypothyroidism, DUOX2-related, a *DUOX2*-related condition.
- Testing for copy number changes in the SMN1 gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family

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history.

- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- This report does not include variants of uncertain significance.
- Genetic counseling is recommended. Contact your physician about the available options for genetic counseling.

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NEURONAL CEROID LIPOFUSCINOSIS, MFSD8-RELATED

Patient	[redacted]	Partner
Result	⊕ Carrier	N/A
Variant Details	***MFSD8*** (NM_152778.3) c.1436G>A (p.Trp479*)	N/A

What is Neuronal ceroid lipofuscinosis, MFSD8-related?

Neuronal ceroid-lipofuscinoses (NCLs) are a group of inherited, neurodegenerative, lysosomal storage disorders characterized by progressive neurological and motor deterioration, seizures, and early death. Vision loss is also a common feature of the disease. The different types are characterized by their age, onset, and specific gene involvement. Symptoms typically first appear in childhood, ranging from late infancy to adolescence and in rare cases, adulthood.

What is my risk of having an affected child?

Neuronal Ceroid-Lipofuscinosis, MFSD8-Related is inherited in an autosomal recessive manner. The risk for being a carrier for MFSD8-related Neuronal Ceroid-Lipofuscinosis, MFSD8-Related is very low (carrier frequency less than 1/500). 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 is no cure for NCLs. Treatment is typically based on the management of symptoms and palliative care.

What mutation was detected?

The detected heterozygous variant was NM_152778.3:c.1436G>A (p.Trp479*). This variant is predicted to introduce a premature stop codon in the last exon or the last 50 nucleotides of the penultimate exon and result in a truncated protein. While this variant is not anticipated to cause nonsense-mediated mRNA decay (PubMed: 25741868, 30192042), it is expected to disrupt the last 40 (7%) amino acids of the original protein. The truncated or altered region is critical to protein function, as indicated by at least one pathogenic variant or at least three cases carrying truncating variants downstream of this position (PubMed: 19177532, 25976102, 28708303). The laboratory classifies this variant as likely pathogenic.

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CYSTIC FIBROSIS

Patient	[redacted]	**Partner**
Result	⊕ Carrier	N/A
Variant Details	***CFTR*** (NM_000492.4) c.1521_1523del (p.Phe508del)	N/A

What is Cystic Fibrosis?

Cystic fibrosis (CF) is a progressive lung disease caused by the body producing mucus that is abnormally thick and sticky. This results in a buildup of mucus in the lungs and the digestive system. This buildup can lead to chronic respiratory infections, lung damage, and malabsorption of nutrients, resulting in poor growth, diarrhea, and a form of diabetes known as cystic fibrosis-related diabetes mellitus. The symptoms are highly variable among individuals, from severe to mild, and may also include complications in pregnancy and male infertility. While CF used to be considered a fatal disease of childhood, many people with CF now live into adulthood.

What is my risk of having an affected child?

Cystic fibrosis is inherited in an autosomal recessive manner. This means that if both parents are carriers, their risk of having an affected child is 1 in 4 (25%). The overall risk of being a carrier for *CFTR*-related CF in the general population is 1 in 32. Individuals of Caucasian/European descent have an increased carrier risk of 1 in 25.

What kind of medical management is available?

Medical advancements have significantly improved the longevity of patients with CF with the median predicted survival age now close to 40 years. Treatments vary depending on severity but may include nebulizers (machines that deliver liquid medicine to the lungs in the form of a fine mist), inhalers, antibiotics, and enzymatic supplementation. Men with congenital absence of the vas deferens (CAVD) may require fertility treatments to father children.

What mutation was detected?

The detected heterozygous variant was NM_000492.4:c.1521_1523del (p.Phe508del). This variant, c.1521_1523del (p.Phe508del), also known as deltaF508, results in the deletion of 3 base pairs in exon 11, leading to an in-frame deletion of phenylalanine at codon 508 of CFTR. This variant is the most common mutation found in patients with CF (PubMed: 20301428, 2475911, 9725922, 23974870, 19774621, 18507830, 36703223). Individuals who are homozygous for the variant demonstrate the classic features of CF, whereas individuals compound heterozygous for the variant may have a modified disease phenotype (PubMed: 19880712, 2570460). Heterozygous carriers of this variant are usually asymptomatic, however, may be at increased risk for developing a CFTR-related disorder (PubMed: 15379964, 1658649). This variant is classified as "Pathogenic" in ClinVar, with a practice guideline assertion (ClinVar: 7105). Functional studies have demonstrated that the ΔF508 mutation impairs the biosynthetic maturation of the CFTR protein, limiting its expression as a phosphorylation-dependent channel on the cell surface and impairing adaptive immune response (PubMed: 28968805, 23436935, 25330774). The laboratory classifies this variant as pathogenic.

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SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY

Patient	[redacted]	Partner
Result	Carrier	N/A
Variant Details	***DYNC2H1*** (NM_001080463.2) c.10626+1G>T (p.?)	N/A

What is Short-rib thoracic dysplasia 3 with or without polydactyly?

Short-rib thoracic dysplasia 3 with or without polydactyly is a disorder that primarily affects the growth of the skeletal system. The effects are a shortening of long bones in the arms and legs, a narrow chest, and short ribs, and some individuals may have extra fingers and/or toes. The features associated with this condition can vary in severity. People with this disorder typically have severe respiratory issues due to having a narrow chest, and as a result, may only live through infancy or early childhood. However, if the difficulty breathing is closely monitored patients with this condition may have some improvements as they age.

What is my risk of having an affected child?

The risk for being a carrier for Short-rib thoracic dysplasia 3 with or without polydactyly is 1/68. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

There are no approved treatments for Short-rib thoracic dysplasia 3 with or without polydactyly, although this is an area of active research and clinical trials. Lifelong monitoring by a medical geneticist is recommended.

What mutation was detected?

The detected heterozygous variant was NM_001080463.2:c.10626+1G>T (p.?). This intronic variant, c.10626+1G>T, alters the highly conserved splice donor site for exon 70 of this transcript and is predicted by all four splice site prediction tools queried to abolish canonical splice donor activity. This variant is expected to result in altered function of the DYNC2H1 gene product as a result of aberrant splicing. While this canonical splice site variant has not, to our knowledge, been previously reported, other splice-disrupting variants in this gene have been established as pathogenic (PubMed: 23339108, 29068549, 29068549, 33452237). The laboratory classifies this variant as likely pathogenic.

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CONGENITAL HYPOTHYROIDISM, DUOX2-RELATED

Patient	[redacted]	Partner
Result	Carrier	N/A
Variant Details	***DUOX2*** (NM_014080.4) c.2895_2898del (p.Phe966Serfs*29)	N/A

What is Congenital hypothyroidism, DUOX2-related?

Congenital hypothyroidism, DUOX2-related is characterized by partial or complete loss of function of the thyroid gland at birth. Signs and symptoms of the condition usually manifest first in infancy or early childhood, and may include feeding difficulty, constipation, low muscle tone, puffy face and a hoarse-sounding cry. If left untreated, the condition can lead to intellectual disability, developmental delay, and poor growth.

What is my risk of having an affected child?

Congenital hypothyroidism, DUOX2-related 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 DUOX2-related congenital hypothyroidism is estimated to be 1 in 366 in the general population.

What kind of medical management is available?

Most states offer congenital hypothyroidism as part of the newborn screening panel. If treatment starts soon after birth, children with primary congenital hypothyroidism (CH) can have healthy growth and development. The most common treatment for primary congenital hypothyroidism (CH) is thyroid hormone replacement therapy. If untreated, congenital hypothyroidism may result in developmental delay or intellectual disability and poor growth.

What mutation was detected?

The detected heterozygous variant was NM_014080.4:c.2895_2898del (p.Phe966Serfs*29). This frameshift variant is the result of a 4-bp deletion which leads to an out of frame transcript, and the introduction of a premature stop codon. This variant is predicted to result in loss of function of the protein product of the DUOX2 gene either as the result of protein truncation, or of nonsense mediated mRNA decay. There's sufficient evidence that loss of function in this gene is a known disease mechanism for thyroid dyshormonogenesis 6 (PubMed: 17684392, 31172499, 33310921, 20187165). This frameshift variant has been reported in the compound heterozygous state in several individuals with partial or full congenital hypothyroidism (PubMed: 12110737, 21565790, 24423310, 31030636). However, the frequency of the variant in control populations is not indicative of it being a highly penetrant variant. Functional studies have demonstrated that this variant leads to complete inhibition of the H2O2-generating activity normal to this protein (PubMed: 21565790, 24423310). This variant is classified as "Pathogenic" or "Likely Pathogenic" in ClinVar, with multiple submitters in agreement (ClinVar:189229). The laboratory classifies this variant as pathogenic.

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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, DUOXA2, 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, MFSD8, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MPI, MPL, MPV17, MTHFR, MTMR2, MTRR, MTTP, 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, RTEL1, SACS, SAMD9, SAMHD1, SCO2, SEPSECS, SERPINA1, SGCA, SGCB, SGCD, SGCG, SGSH, SH3TC2, SLC12A6, SLC17A5, SLC19A3, SLC1A4, SLC22A5, SLC25A13, SLC25A15, SLC26A2, SLC26A3, SLC35A3, SLC37A4, SLC39A4, SLC45A2, SLC46A1, SLC5A5, SLC7A7, SMARCAL1, SMN1, SMPD1, SPG11, SPINK5, STAR, SUMF1, SURF1, TCIRG1, TCTN2, TECPR2, TF, TG, TGM1, TH, TMEM216, TPO, TPP1, TRDN, TRIM32, TRMU, TSEN54, TSFM, TSHB, TTC37, TTPA, TYMP, TYR, TYRP1, UGT1A1, USH1C, USH1G, USH2A, VPS13A, VPS13B, VPS45, VPS53, VRK1, VSX2, WHRN, WRN, XPA, XPC, ZFYVE26

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 99.57% 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

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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:* 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. *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

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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:

Jianbo Song, Ph.D., ABMGG, CGMB, CCS, FACMG on **1/3/2024 04:45 PM PST**
Electronically signed

DISCLAIMER:

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ABCA12	Congenital ichthyosis, ABCA12-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
ABCA4	Stargardt disease	AR	General Population	1 in 51	98%	1 in 2,501	1 in 510,204
ABCB11	Progressive familial intrahepatic cholestasis	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
ABCC8	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
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACADM	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
ACADVL	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
ACAT1	3-ketothiolase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACSF3	Combined malonic and methylmalonic aciduria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADA	Adenosine deaminase deficiency	AR	General Population	1 in 224	93%	1 in 3,187	1 in 2,855,552
ADAMTS2	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
ADGRG1	Bilateral frontoparietal polymicrogyria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
AGA	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
AGL	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
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
AGXT	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
AHI1	Joubert syndrome, AHI1-related	AR	General Population	1 in 448	99%	1 in 44,701	<1 in 10 million
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR	General Population	1 in 409	99%	1 in 40,801	<1 in 10 million
ALDH3A2	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
ALDOB	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
ALG6	Congenital disorder of glycosylation type Ic	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ALMS1	Alstrom syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
ALPL	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
AMT	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
AQP2	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
ARG1	Arginase deficiency	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
ARL13B	Joubert syndrome, ARL13B-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ARSA	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
ARSB	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
ASL	Argininosuccinate lyase deficiency	AR	General Population	1 in 132	90%	1 in 1,311	1 in 692,208
ASNS	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

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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	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
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
CHRNG	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

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CPS1	Carbamoylphosphate synthetase I deficiency	AR	General Population	1 in 570	98%	1 in 28,451	<1 in 10 million
CPT1A	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
CPT2	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
CRB1	CRB1-related retinopathy	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	UK	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
CTNS	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
CTSA	Galactosialidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CTSC	Papillon-Lefevre syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CTSD	Neuronal ceroid lipofuscinosis, CTSD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
CTSK	Pycnodysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYBA	Chronic granulomatous disease	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
CYP11A1	Congenital adrenal insufficiency	AR	General Population	1 in 114	99%	1 in 11,301	1 in 5,153,256
CYP11B1	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
			Morrocan Jewish Population	1 in 35	98%	1 in 1,701	1 in 238,140
CYP11B2	Corticosterone methyloxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
CYP1B1	Primary congenital glaucoma	AR	General Population	1 in 50	99%	1 in 4,901	1 in 980,200
CYP21A2	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
CYP27A1	Cerebrotendinous xanthomatosis	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Morrocan Jewish Population	1 in 5	98%	1 in 201	1 in 4,020
DBT	Maple syrup urine disease, type II	AR	General Population	1 in 481	98%	1 in 24,001	<1 in 10 million
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
DDX11	Warsaw breakage syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 68	99%	1 in 6,701	1 in 1,822,672
DHCR7	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
DHDDS	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
DLD	Dihydrolipoamide 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
DNAH5	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
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
DUOX2	Congenital hypothyroidism, DUOX2-related	AR	General Population	1 in 366	91%	1 in 4,057	1 in 5,938,797
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	General Population	1 in 68	98%	1 in 3,351	1 in 924,876
DYSF	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
EIF2AK3	Wolcott-Rallison Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EIF2B5	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ELP1	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
ERCC2	ERCC2-related disorders	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
ERCC5	Xeroderma Pigmentosa, group G	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC6	ERCC6-related disorders	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
ERCC8	Cockayne syndrome type A	AR	General Population	1 in 822	98%	1 in 41,051	<1 in 10 million
ESCO2	Roberts syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ETFA	Glutaric aciduria IIA	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million

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ETFB	Glutaric aciduria IIB	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
ETFDH	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
ETHE1	Ethylmalonic encephalopathy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EVC	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
EVC2	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
EXOSC3	Pontocerebellar hypoplasia type 1B	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
F2	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
F5	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
FAH	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
FAM126A	Hypomyelinating leukodystropy type 5	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FAM161A	Retinitis pigmentosa 28	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
FANCA	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
FANCC	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
FANCG	Fanconi anemia group G	AR	General Population	1 in 632	90%	1 in 6,311	<1 in 10 million
FH	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
FKRP	FKRP Alpha-dystroglycanopathies	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
FKTN	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
FOXRED1	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FTCD	Glutamate formiminotransferase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FUCA1	Fucosidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
G6PC	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
GAA	Pompe disease	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
			African/African American Population	1 in 60	98%	1 in 2,951	1 in 708,240
			East Asian Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
GALC	Krabbe disease	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			Israeli Druze Population	1 in 6	99%	1 in 501	1 in 12,024
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR	General Population	1 in 224	97%	1 in 7,434	1 in 6,660,864
GALT	Galactosemia	AR	General Population	1 in 110	99%	1 in 10,901	1 in 4,796,440
			African/African American Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Ashkenazi Jewish Population	1 in 127	99%	1 in 12,601	1 in 6,401,308
GAMT	Guanidinoacetate methyltransferase deficiency	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
GBA	Gaucher disease	AR	General Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			African/African American Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Ashkenazi Jewish Population	1 in 15	99%	1 in 1,401	1 in 84,060
GBE1	Glycogen storage disease IV	AR	General Population	1 in 387	99%	1 in 38,601	<1 in 10 million
GCDH	Glutaric aciduria, type I	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Amish Population	1 in 9	98%	1 in 401	1 in 14,436
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR	General Population	1 in 152	99%	1 in 15,101	1 in 9,181,408
GDF5	Du Pan Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
GJB2	Nonsyndromic hearing loss 1A	AR	General Population	1 in 42	99%	1 in 4,101	1 in 688,968
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 240,100
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 168,084
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Latino Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 8,702,992
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
GLB1	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 980,200
GLDC	Glycine encephalopathy, GLDC-related	AR	General Population	1 in 193	98%	1 in 9,601	1 in 7,411,972
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 5,429,268
GLE1	Lethal congenital contracture syndrome 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	<1 in 500	99%	1 in 49,901	1 in 99,802,000
			Iranian Jewish Population	1 in 11	99%	1 in 1,001	1 in 44,044
GNPTAB	Mucolipidosis II & III	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTG	Mucolipidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GSS	Glutathione synthetase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GUCY2D	Leber congenital amaurosis 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GUSB	Mucopolysaccharidosis type VII	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
HADHA	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHB	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HAX1	Severe congenital neutropenia, HAX1-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
HBA1	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
HBA2	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
HBB	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
HBB	Hemoglobin C disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
HBB	Beta thalassemia	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			African/African American Population	1 in 10	99%	1 in 901	1 in 36,040
			East Asian Population	1 in 50	99%	1 in 4,901	1 in 980,200
			Latino Population	1 in 128	99%	1 in 12,701	1 in 6,502,912
			Mediterranean Population	1 in 3	99%	1 in 201	1 in 2,412
			South Asian/Indian Population	1 in 25	99%	1 in 2,401	1 in 240,100
HEXA	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
HEXB	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
HGSNAT	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
HJV	Hemochromatosis, type 2A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
HLCS	Holocarboxylase synthetase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HOGA1	Primary hyperoxaluria type III	AR	General Population	1 in 184	99%	1 in 18,301	<1 in 10 million
HPS1	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
HPS3	Hermansky-Pudlak syndrome 3	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
HPS4	Hermansky-Pudlak syndrome 4	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HSD17B4	D-bifunctional protein deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
HSD3B2	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
HYLS1	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
IDUA	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
IVD	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
IYD	Thyroid dyshormonogenesis, IYD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
JAK3	Severe combined immunodeficiency, JAK3-related	AR	General Population	1 in 299	99%	1 in 29,801	<1 in 10 million
KCNJ11	KCNJ11-related 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
LAMA2	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
LAMA3	Junctional epidermolysis bullosa 2	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LCA5	Leber congenital amaurosis 5	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
LDLRAP1	Familial Hypercholesterolemia	AR	General Population	1 in 8	99%	1 in 701	1 in 22,432
			Amish Population	1 in 2	99%	1 in 101	1 in 808
			Caucasian / European Population	1 in 7	99%	1 in 601	1 in 16,828
			French Canadian Population	1 in 8	99%	1 in 701	1 in 22,432
LHX3	Combined pituitary hormone deficiency 3	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
LIFR	Stuve-Wiedemann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
LIPA	Lysosomal acid lipase deficiency	AR	General Population	1 in 211	99%	1 in 21,001	<1 in 10 million
			Caucasian / European Population	1 in 161	99%	1 in 16,001	1 in 4,973,248
			Iranian Jewish Population	1 in 32	99%	1 in 3,101	1 in 396,928
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
LOXHD1	Nonsyndromic hearing loss 77	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 180	98%	1 in 8,951	1 in 6,444,720
LPL	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
LRP2	Donnai–Barrow syndrome	AR	General Population	1 in 214	99%	1 in 10,651	1 in 9,117,256
LRPPRC	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
LYST	Chediak-Higashi syndrome	AR	General Population	<1 in 500	90%	1 in 4,991	1 in 9,982,000
MAN2B1	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
MANBA	Beta-Mannosidosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MCOLN1	Mucolipidosis 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
MCPH1	Primary microcephaly 1, recessive	AR	General Population	1 in 147	99%	1 in 14,601	1 in 8,585,388
MED17	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
MESP2	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
MKS1	MKS1-related ciliopathies	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

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MLC1	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
MLYCD	Malonyl-CoA decarboxylase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MMAA	Methylmalonic aciduria, cblA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
MMAB	Methylmalonic aciduria, cblB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR	General Population	1 in 134	90%	1 in 1,331	1 in 713,416
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MPI	Congenital disorder of glycosylation type Ib	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MPL	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
MPV17	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
MTHFR	Homocystinuria, MTHFR-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
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 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
MUT	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
MVK	Mevalonate kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MYO7A	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
NAGA	Schindler disease types 1 and 3	AR	General Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
NAGLU	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
NAGS	N-acetylglutamate synthase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
NBN	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR	General Population	1 in 22	98%	1 in 1,051	1 in 92,488
NDUFAF2	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population 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
NDUFS4	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFS4	Mitochondrial complex I deficiency	AR	General Population 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
NDUFS6	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
NDUFS7	Mitochondrial complex I deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NEB	Nemaline myopathy	AR	General Population 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
NEU1	Sialidosis, type I and II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
NPC1	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
NPC2	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
NPHP1	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
NPHS1	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
NPHS2	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
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
OAT	Gyrate atrophy of choroid and retina	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
OCA2	Oculocutaneous albinism type II	AR	General Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
OPA3	Costeff syndrome	AR	General Population 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
OTOF	Nonsyndromic hearing loss, OTOF-related	AR	General Population Spanish Population	<1 in 500 1 in 106	99% 99%	1 in 49,901 1 in 10,501	<1 in 10 million 1 in 4,452,424

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Supplemental Table

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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
RAG2	Omenn syndrome, RAG2-related	AR	General Population	1 in 137	98%	1 in 6,801	1 in 3,726,948
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

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Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
RARS2	Pontocerebellar hypoplasia type 6	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAX	Microphthalmia, isolated 3	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million
RDH12	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
RMRP	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
RNASEH2B	Aicardi Goutieres syndrome 2	AR	General Population	1 in 217	99%	1 in 21,601	1 in 18,749,668
RPE65	RPE65-related retinopathy	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
RPGRIP1L	RPGRIP1L-related ciliopathies	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RTEL1	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
SACS	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
SAMD9	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
SAMHD1	Aicardi-Goutieres syndrome	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
SCO2	Mitochondrial complex IV deficiency	AR	General Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
SEPSECS	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
SERPINA1	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
SGCA	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
SGCB	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
SGCD	Limb-girdle muscular dystrophy, type 2F	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SGCG	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
SGSH	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
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2-related	AR	General Population	1 in 69	99%	1 in 6,801	1 in 1,877,076
SLC12A6	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
SLC17A5	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
SLC19A3	Biotin-responsive basal ganglia disease	AR	General Population	1 in 109	99%	1 in 5,401	1 in 2,354,836
SLC1A4	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
SLC22A5	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
SLC25A13	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
SLC25A15	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
SLC26A2	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
SLC26A3	Congenital secretory chloride diarrhea	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Middle-Eastern Population	1 in 57	98%	1 in 2,801	1 in 638,628
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 453	98%	1 in 22,601	<1 in 10 million
SLC37A4	Glycogen storage disease, type Ib	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Ashkenazi Jewish Population	1 in 71	95%	1 in 1,401	1 in 397,884
SLC39A4	Acrodermatitis enteropathica	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC45A2	Oculocutaneous albinism, type IV	AR	General Population	1 in 159	98%	1 in 7,901	1 in 5,025,036
			Japanese Population	1 in 146	98%	1 in 7,251	1 in 4,234,584

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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 dyshormonogenesis, 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 23,301	<1 in 10 million
			Ashkenazi Jewish Population	1 in 17	99%	1 in 1,601	1 in 108,868
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SUMF1	Multiple sulfatase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 320	98%	1 in 15,951	<1 in 10 million
SURF1	Charcot-Marie-Tooth disease, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SURF1	Leigh syndrome, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
TCIRG1	Osteopetrosis 1	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TCTN2	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
TECPR2	Spastic paraplegia 49	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TF	Atransferrinemia	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
TG	Thyroid dyshormonogenesis, 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
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
TPO	Thyroid dyshormonogenesis, TPO-related	AR	General Population	1 in 373	99%	1 in 37,201	<1 in 10 million
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	General Population	1 in 252	97%	1 in 8,368	1 in 8,434,944
			French Canadian Population	1 in 53	97%	1 in 1,734	1 in 367,608
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
TRIM32	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

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
USH2A	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
VPS13A	Choreoacanthocytosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS13B	Cohen syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS45	Severe congenital neutropenia, VPS45-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
VPS53	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
VRK1	Pontocerebellar hypoplasia type 1A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VSX2	Microphthalmia with or without coloboma	AR	General Population	1 in 91	98%	1 in 4,501	1 in 1,638,364
WHRN	Usher syndrome type 2D	AR	General Population	1 in 282	99%	1 in 28,101	<1 in 10 million
WRN	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
XPA	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
XPC	Xeroderma pigmentosum, group C	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
ZFYVE26	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 (αα/α-) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis (αα/- -) is 1 in 1000.
Abbreviations: AR, autosomal recessive; XL, X-linked