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Partner Information:
Not Tested

Physician:
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Laboratory:
Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Apr 08,2022**

Sex: M
[redacted]

Accession:
FT-6628010
Test#: FT-TS12793574
Specimen Type: Blood (EDTA)
Collected: Mar 16,2022

Accession:
N/A

FINAL RESULTS

Carrier for **ONE** genetic condition
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	[redacted]	Partner
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency *CYP21A2*	AR	Possible Carrier c.955C>T(;)*12C>T + CYP21A2 duplication p.(Gln319*)(;)(?)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for a carrier mutation in 1 gene. The risk estimates below are quantified based on general population carrier frequencies. Carrier screening for the reproductive partner is recommended to accurately assess this risk:
  - There is up to a 1/244 chance of having a child affected with Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, a *CYP21A2*-related condition.
- Testing for copy number changes in the *SMN1* gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- 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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CONGENITAL ADRENAL HYPERPLASIA DUE TO 21-HYDROXYLASE DEFICIENCY

Patient		Partner
Result	Possible Carrier	N/A
Variant Details	***CYP21A2*** (NM_000500.9) c.955C>T(;)*12C>T + CYP21A2 duplication p.(Gln319*)(;)(?)	N/A

What is Congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency is an inherited disorder that affects the adrenal glands and hormone production. Approximately 75 percent of individuals with classic 21-hydroxylase deficiency have the salt-wasting type, whereby the body excretes too much salt in urine. Affected infants present with poor feeding, weight loss, dehydration, and vomiting, all of which can be life-threatening. Females with this condition typically have ambiguous genitalia, while males usually have normal genitalia, but with small testes. Individuals with the simple virilizing form and the non-classic form of the disease do not experience salt loss. Males and females with either the classic or non-classic forms of 21-hydroxylase deficiency tend to have an early growth spurt, but their final adult height is usually shorter than others in their family, and affected individuals may have reduced fertility. Additionally, individuals may have excessive body hair growth, hair loss, and irregular menstruation. Some individuals (male or female) with the non-classic form of the disease may have mild, non-life-threatening symptoms, while others may never develop symptoms of the disorder at all.

What is my risk of having an affected child?

CAH due to 21-hydroxylase deficiency is inherited in an autosomal recessive manner. The risk for being a carrier for CYP21A2-related CAH is 1/61. Individuals of Inuit descent have an increased carrier risk of 1/9. Individuals of Middle-Eastern descent have an increased carrier risk of 1/35. 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?

Treatment consists of early initiation of hormone replacement therapy and/or surgery for females. Prognosis is good for patients with appropriate medical management and psychological support.

What mutation was detected?

The heterozygous variants c.955C>T (p.Gln319*) and a whole gene duplication of CYP21A2 were detected in this sample. In addition, the benign polymorphism c.*12C>T was also detected. The phase of these variants is unknown but could be determined through parental testing.

The nonsense variant, p.Gln319*, introduces a premature stop codon and is expected to result in the loss of function of the protein product of the CYP21A2 gene, either as the result of protein truncation or of nonsense-mediated mRNA decay. This variant, also reported as Q318*, is a classic 21-hydroxylase-deficient congenital adrenal hyperplasia mutation and has been reported in multiple affected individuals (PubMed: 3267225, 12220458, 12915679). The variant, p.Gln319*, and the polymorphism c.*12C>T are known to frequently occur in a duplicated copy of the CYP21A2 gene coexisting with a normal copy of CYP21A2 on the same chromosome. This haplotype was identified in approximately 2% of the general population and in ~80% of carriers of p.Gln319*, and such a configuration may represent a benign allele (PubMed: 28401898, 19773403). Nonetheless, there is a possibility that p.Gln319* occurs on a chromosome with only a single copy of CYP21A2, in which case it results in a pathogenic allele. If multiple copies of CYP21A2 are present, we cannot be certain if this p.Gln319* variant occurs on a chromosome with one (i.e. pathogenic state) or two (i.e. benign state) copies of CYP21A2. While this combination of variants may represent a benign allele, the laboratory classifies the variant p.Gln319* as likely pathogenic.

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GENES TESTED:

Custom Sonic Beacon Male Carrier Screening - 351 Genes

351 genes tested (99.47% at >20x). For more gene specific information and assistance with residual risk calculation, see SUPPLEMENTAL TABLE.

ABCB11	*ABCC8*	*ABCD4*	*ACAD9*	*ACADM*	*ACADS*
ACADSB	*ACADVL*	*ACAT1*	*ACOX1*	*ACSF3*	*ADA*
ADAMTS2	*ADGRG1*	*ADK*	*AGA*	*AGL*	*AGPS*
AGXT	*AHCY*	*AHI1*	*AIPL1*	*AIRE*	*ALDH3A2*
ALDH4A1	*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*	*BTD*	*CAPN3*	*CASQ2*
CBS	*CCDC103*	*CCDC151*	*CCDC39*	*CDH23*	*CEP290*
CERKL	*CFTR*	*CHRNE*	*CHRNG*	*CHST6*	*CIITA*
CLN3	*CLN5*	*CLN6*	*CLN8*	*CLRN1*	*CNGA1*
CNGB1	*CNGB3*	*COL27A1*	*COL4A3*	*COL4A4*	*COL7A1*
CPS1	*CPT1A*	*CPT2*	*CRB1*	*CRYL1*	*CTNS*
CTSK	*CYBA*	*CYP11B1*	*CYP11B2*	*CYP17A1*	*CYP19A1*
CYP1B1	*CYP21A2*	*CYP27A1*	*DBT*	*DCLRE1C*	*DHCR7*
DHDDS	*DLD*	*DNAH5*	*DNAI1*	*DNAI2*	*DNAL1*
DPYD	*DUOX2*	*DUOXA2*	*DYSF*	*EIF2AK3*	*EIF2B5*
ELP1	*ERCC6*	*ERCC8*	*ESCO2*	*ETFA*	*ETFB*
ETFDH	*ETHE1*	*EVC*	*EVC2*	*EXOSC3*	*EYS*
F11	*FAH*	*FAM161A*	*FANCA*	*FANCC*	*FANCG*
FH	*FKRP*	*FKTN*	*FTCD*	*G6PC*	*GAA*
GALC	*GALE*	*GALK1*	*GALNS*	*GALT*	*GAMT*
GBA	*GBE1*	*GCDH*	*GDAP1*	*GFM1*	*GJB2*
GJB6	*GLB1*	*GLDC*	*GLE1*	*GNE*	*GNPTAB*
GNPTG	*GNRHR*	*GNS*	*GP1BA*	*GP9*	*GRHPR*
GUSB	*HADHA*	*HAX1*	*HBA1*	*HBA2*	*HBB*
HEXA	*HEXB*	*HGD*	*HGSNAT*	*HJV*	*HLCS*
HMGCL	*HOGA1*	*HPS1*	*HPS3*	*HSD17B4*	*HSD3B2*
HYAL1	*HYLS1*	*IDH3B*	*IDUA*	*IVD*	*IYD*
JAK3	*KCNJ11*	*LAMA2*	*LAMA3*	*LAMB3*	*LAMC2*
LCA5	*LHX3*	*LIFR*	*LIPA*	*LMBRD1*	*LOXHD1*
LPL	*LRPPRC*	*LYST*	*MAN2B1*	*MCCC1*	*MCCC2*
MCEE	*MCOLN1*	*MED17*	*MESP2*	*MFSD8*	*MKS1*
MLC1	*MMAA*	*MMAB*	*MMACHC*	*MMADHC*	*MPI*
MPL	*MPV17*	*MTMR2*	*MTRR*	*MTTP*	*MUT*
MVK	*MYO7A*	*NAGLU*	*NAGS*	*NBN*	*NDRG1*
NDUFAF5	*NDUFS6*	*NEB*	*NPC1*	*NPC2*	*NPHP1*
NPHS1	*NPHS2*	*NR2E3*	*NTRK1*	*OAT*	*OPA3*
OTOF	*P3H1*	*PAH*	*PANK2*	*PC*	*PCBD1*
PCCA	*PCCB*	*PCDH15*	*PDE6A*	*PDHB*	*PEX1*
PEX10	*PEX12*	*PEX2*	*PEX6*	*PEX7*	*PFKM*
PGK1	*PHGDH*	*PKHD1*	*PLA2G6*	*PLOD1*	*PMM2*
POLG	*POLR1C*	*POMGNT1*	*POMT1*	*POMT2*	*PPT1*
PQBP1	*PROP1*	*PSAP*	*PTS*	*PUS1*	*PYGM*
QDPR	*RAB23*	*RAG1*	*RAG2*	*RAPSN*	*RARS2*
RAX	*RDH12*	*RMRP*	*RPE65*	*RPGRIP1L*	*RTEL1*
SACS	*SAMHD1*	*SEPSECS*	*SERPINA1*	*SGCA*	*SGCB*

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SGCD	*SGCG*	*SGSH*	*SH3TC2*	*SLC12A3*	*SLC12A6*
SLC16A2	*SLC17A5*	*SLC22A5*	*SLC25A13*	*SLC25A15*	*SLC25A20*
SLC26A2	*SLC26A3*	*SLC26A4*	*SLC35A3*	*SLC37A4*	*SLC39A4*
SLC46A1	*SLC4A11*	*SLC5A5*	*SLC6A19*	*SLC7A7*	*SMARCAL1*
SMN1	*SMPD1*	*SPG11*	*SPG7*	*STAR*	*SUMF1*
SURF1	*TAT*	*TCIRG1*	*TECPR2*	*TFR2*	*TG*
TGM1	*TH*	*TMEM216*	*TPO*	*TPP1*	*TRDN*
TRIM32	*TRMU*	*TSFM*	*TSHB*	*TTC37*	*TTPA*
TYMP	*UGT1A1*	*USH1C*	*USH1G*	*USH2A*	*VPS13A*
VPS13B	*VPS45*	*VRK1*	*VSX2*	*WHRN*	*WNT10A*
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, 99.52% and 99.47% 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 identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. 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 >=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

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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

Biotinidase deficiency: BTD
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.

Cystic Fibrosis: 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.

GJB6-CRYL1 related nonsyndromic hearing loss, GJB6-CRYL1 related nonsyndromic hearing loss: 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.

Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency: 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.

Corticosterone methyloxidase deficiency: 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.

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: 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.

Congenital hypothyroidism, DUOX2-related: 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.

Galactosemia: 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).

Gaucher disease: 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.

Nonsyndromic hearing loss, GJB2-related: GJB2
If detected, the variant NM_004004.6:c.109G>A (p.Val37Ile) will not be reported as this variant is associated with low disease penetrance and is primarily associated with late onset and/or mild hearing loss when homozygous.

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Alpha thalassemia, Alpha thalassemia: HBA1
The phase of heterozygous alterations in the *HBA1* gene cannot be determined, but can be confirmed through parental testing.

Alpha thalassemia, Alpha thalassemia: HBA2
The phase of heterozygous alterations in the *HBA2* gene cannot be determined, but can be confirmed through parental testing.

Nemaline myopathy: NEB
This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis.

Congenital nephrotic syndrome, type 2: 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.

Alpha-1 antitrypsin deficiency: 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.

Spinal muscular atrophy: 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.

Catecholaminergic polymorphic ventricular tachycardia, TRDN-related: 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.

Crigler-Najjar syndrome: UGT1A1
Common variants in the *UGT1A1 gene* (population allele frequency >5%) are typically not reported as they do not cause a Mendelian condition.

Odontoonychodermal dysplasia, Schopf-Schulz-Passarge syndrome: 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:

Yan Meng, Ph.D., CGMB, FACMG on **4/8/2022 05:20 PM PDT**
Electronically signed

DISCLAIMER:

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

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of the test result, including its residual risks, uncertainties and reproductive or medical options.

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
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
ACADS	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
ACADSB	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
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
AHCY	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR	General Population	<1 in 500	99%	1 in 49,901	<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
AIRE	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
ALDH3A2	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
ALDH4A1	Hyperprolinemia type II	AR	General Population	<1 in 500	99%	1 in 49,901	<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

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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 Caucasian / European Population Yemenite Jewish Population	1 in 100 1 in 78 1 in 75	99% 99% 99%	1 in 9,901 1 in 7,701 1 in 7,401	1 in 3,960,400 1 in 2,402,712 1 in 2,220,300
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR	General Population Western Australian Population	1 in 250 1 in 283	98% 98%	1 in 12,451 1 in 14,101	<1 in 10 million <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 Iranian Jewish Population	<1 in 500 1 in 80	99% 99%	1 in 49,901 1 in 7,901	<1 in 10 million 1 in 2,528,320
ASPA	Canavan disease	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 55	97% 96%	1 in 9,968 1 in 1,351	<1 in 10 million 1 in 297,220
ASS1	Citrullinemia	AR	General Population East Asian Population	1 in 119 1 in 132	96% 96%	1 in 2,951 1 in 3,276	1 in 1,404,676 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 Caucasian / European Population Ashkenazi Jewish Population	1 in 87 1 in 42 1 in 70	98% 98% 98%	1 in 4,301 1 in 2,051 1 in 3,451	1 in 1,496,748 1 in 344,568 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	Bardet-Biedl syndrome 2	AR	General Population Ashkenazi Jewish Population	1 in 621 1 in 107	99% 99%	1 in 62,001 1 in 10,601	<1 in 10 million 1 in 4,537,228
BBS2	Retinitis Pigmentosa 74	AR	General Population Ashkenazi Jewish Population	1 in 621 1 in 107	99% 99%	1 in 62,001 1 in 10,601	<1 in 10 million 1 in 4,537,228
BCKDHA	Maple syrup urine disease type Ia	AR	General Population Mennonite Population	1 in 321 1 in 10	98% 98%	1 in 16,001 1 in 451	<1 in 10 million 1 in 18,040
BCKDHB	Maple syrup urine disease type Ib	AR	General Population Ashkenazi Jewish Population	1 in 364 1 in 97	98% 98%	1 in 18,151 1 in 4,801	<1 in 10 million 1 in 1,862,788
BCS1L	Björnstad syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	GRACILE syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
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 Ashkenazi Jewish Population	1 in 800 1 in 134	87% 99%	1 in 6,147 1 in 13,301	<1 in 10 million 1 in 7,129,336
BSND	Bartter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
BTD	Biotinidase deficiency	AR	General Population Caucasian / European Population Latino Population Middle-Eastern Population	1 in 124 1 in 71 1 in 136 1 in 55	99% 99% 99% 99%	1 in 12,301 1 in 7,001 1 in 13,501 1 in 5,401	1 in 6,101,296 1 in 1,988,284 1 in 7,344,544 1 in 1,188,220
CAPN3	Limb-girdle muscular dystrophy type 2A	AR	General Population Caucasian / European Population	<1 in 500 1 in 103	98% 98%	1 in 24,951 1 in 5,101	<1 in 10 million 1 in 2,101,612
CASQ2	Catecholaminergic polymorphic ventricular tachycardia, CASQ2-related	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 Caucasian / European Population Middle-Eastern Population	1 in 224 1 in 86 1 in 21	99% 99% 99%	1 in 22,301 1 in 8,501 1 in 2,001	<1 in 10 million 1 in 2,924,344 1 in 168,084
CCDC103	Primary ciliary dyskinesia, type 17	AR	General Population	1 in 316	98%	1 in 15,751	<1 in 10 million
CCDC151	Primary ciliary dyskinesia, type 30	AR	General Population	1 in 365	98%	1 in 18,201	<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
CDH23	Usher syndrome, type 1D	AR	General Population	1 in 285	90%	1 in 2,841	1 in 11,364
CEP290	Joubert syndrome 5	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Leber congenital amaurosis 10	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Bardet-Biedl syndrome 14	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	CEP290-related disorders	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Senior-Løken syndrome 6	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Meckel syndrome 4	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CERKL	Retinitis pigmentosa 26	AR	General Population	1 in 148	98%	1 in 7,351	1 in 4,351,792

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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, 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
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
CNGA1	Retinitis Pigmentosa, CNGA1-related	AR	General Population	1 in 210	99%	1 in 20,901	<1 in 10 million
CNGB1	Retinitis Pigmentosa, CNGB1-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
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
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	Leber congenital amaurosis 8	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRB1	Retinitis pigmentosa 12	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
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
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
CYP19A1	Aromatase 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
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

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DHDDS	Retinitis pigmentosa 59	AR	General Population Ashkenazi Jewish Population	1 in 296 1 in 118	98% 98%	1 in 14,751 1 in 5,851	<1 in 10 million 1 in 2,761,672
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 107	98% 98%	1 in 24,951 1 in 5,301	<1 in 10 million 1 in 2,268,828
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR	General Population Ashkenazi Jewish Population	1 in 142 1 in 113	98% 99%	1 in 7,051 1 in 11,201	1 in 4,004,968 1 in 5,062,852
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
DNAL1	Primary ciliary dyskinesia, DNAL1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<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
DYSF	Limb-girdle muscular dystrophy type 2B	AR	General Population Japanese Population Libyan Jewish Population	<1 in 500 1 in 332 1 in 18	95% 95% 95%	1 in 9,981 1 in 6,621 1 in 341	<1 in 10 million 1 in 8,792,688 1 in 24,552
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	98%	1 in 24,951	<1 in 10 million
ELP1	Familial Dysautonomia	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 31	99% 99%	1 in 29,901 1 in 3,001	<1 in 10 million 1 in 372,124
ERCC6	De Sanctis-Cacchione syndrome	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
ERCC6	Cockayne syndrome type B	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
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
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 East Asian Population	1 in 250 1 in 74	98% 98%	1 in 12,451 1 in 3,651	<1 in 10 million 1 in 1,080,696
ETHE1	Ethylmalonic encephalopathy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EVC	Weyers acrofacial dysostosis, EVC-related	AR	General Population Amish Population	1 in 142 1 in 7	98% 98%	1 in 7,051 1 in 301	1 in 4,004,968 1 in 8,428
EVC	Ellis-van Creveld syndrome, EVC-related	AR	General Population Amish Population	1 in 142 1 in 7	98% 98%	1 in 7,051 1 in 301	1 in 4,004,968 1 in 8,428
EVC2	Weyers acrodental dysostosis, EVC2-related	AR	General Population Amish Population	1 in 240 1 in 7	98% 98%	1 in 11,951 1 in 301	<1 in 10 million 1 in 8,428
EVC2	Ellis-van Creveld syndrome, EVC2-related	AR	General Population Amish Population	1 in 240 1 in 7	98% 98%	1 in 11,951 1 in 301	<1 in 10 million 1 in 8,428
EXOSC3	Pontocerebellar hypoplasia type 1B	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
EYS	Retinitis pigmentosa 25	AR	General Population	1 in 66	98%	1 in 3,251	1 in 858,264
F11	Factor XI deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 11	98% 98%	1 in 24,951 1 in 501	<1 in 10 million 1 in 22,044
FAH	Tyrosinemia, type 1	AR	General Population Ashkenazi Jewish Population Finnish Population French Canadian Population South Asian/Indian Population	1 in 99 1 in 150 1 in 122 1 in 66 1 in 172	95% 95% 95% 95% 95%	1 in 1,961 1 in 2,981 1 in 2,421 1 in 1,301 1 in 3,421	1 in 776,556 1 in 1,788,600 1 in 1,181,448 1 in 343,464 1 in 2,353,648
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 Moroccan Jewish Indian Jewish Population	1 in 239 1 in 100 1 in 27	99% 99% 99%	1 in 23,801 1 in 9,901 1 in 2,601	<1 in 10 million 1 in 3,960,400 1 in 280,908
FANCC	Fanconi anemia group C	AR	General Population Ashkenazi Jewish Population	1 in 535 1 in 99	99% 99%	1 in 53,401 1 in 9,801	<1 in 10 million 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 Ashkenazi Jewish Population	<1 in 500 1 in 99	99% 99%	1 in 49,901 1 in 9,801	<1 in 10 million 1 in 3,881,196
FKRP	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
FKTN	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	General Population Ashkenazi Jewish Population Japanese Population	<1 in 500 1 in 150 1 in 82	99% 99% 99%	1 in 49,901 1 in 14,901 1 in 8,101	<1 in 10 million 1 in 8,940,600 1 in 2,657,128

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FKTN	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
FTCD	Glutamate formiminotransferase deficiency	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
GALE	Galactose epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GALK1	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
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
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
GJB2	Nonsyndromic hearing loss, 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
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
GLB1	GM1-gangliosidosis	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
GLB1	Mucopolysaccharidosis type IVB (Morquio syndrome B)	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
GLDC	Glycine encephalopathy, GLDC-related	AR	General Population	1 in 193	98%	1 in 9,601	1 in 7,411,972
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 5,429,268
GLE1	Lethal congenital contracture syndrome 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	<1 in 500	99%	1 in 49,901	1 in 99,802,000
			Iranian Jewish Population	1 in 11	99%	1 in 1,001	1 in 44,044
GNPTAB	Mucolipidosis II alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTAB	Mucolipidosis III alpha/beta	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
GNRHR	Hypogonadotropic hypogonadism, GNRHR-related	AR	General Population	1 in 347	99%	1 in 34,601	<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
GP1BA	Bernard-Soulier syndrome type A1	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GP9	Bernard-Soulier syndrome type C	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
GRHPR	Primary hyperoxaluria type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
GUSB	Mucopolysaccharidosis type VII	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
HADHA	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
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 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
HBA2	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
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	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
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
HGD	Alkaptonuria	AR	General Population	1 in 250	90%	1 in 2,491	1 in 2,491,000
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
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
HYAL1	Mucopolysaccharidosis type IX	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
IDH3B	Retinitis pigmentosa, IDH3B-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
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

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Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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	Congenital hyperinsulinism	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
KCNJ11	Permanent neonatal diabetes mellitus	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
LAMA2	Muscular dystrophy, LAMA2-related	AR	General Population Caucasian / European Population	<1 in 500 1 in 125	99% 99%	1 in 49,901 1 in 12,401	<1 in 10 million 1 in 6,200,500
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMA3	Laryngo-onycho-cutaneous syndrome	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
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 Caucasian / European Population Iranian Jewish Population	<1 in 500 1 in 112 1 in 26	99% 99% 99%	1 in 49,901 1 in 11,101 1 in 2,501	<1 in 10 million 1 in 4,973,248 1 in 260,104
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, LOXHD1-related	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
LPL	Familial lipoprotein lipase deficiency	AR	General Population French Canadian Population	1 in 500 1 in 46	99% 99%	1 in 49,901 1 in 4,501	<1 in 10 million 1 in 828,184
LRPPRC	Leigh syndrome with Complex IV deficiency	AR	General Population Faroese Population French Canadian Population	1 in 447 1 in 21 1 in 22	98% 98% 98%	1 in 22,301 1 in 1,001 1 in 1,051	<1 in 10 million 1 in 84,084 1 in 92,488
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 Caucasian / European Population	1 in 354 1 in 274	99% 99%	1 in 35,301 1 in 27,301	<1 in 10 million <1 in 10 million
MCCC1	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
MCCC2	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
MCEE	Methylmalonyl-CoA epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
MCOLN1	Mucolipidosis IV	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 100	99% 99%	1 in 29,901 1 in 9,901	<1 in 10 million 1 in 3,960,400
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR	General Population Bukharan/Kurdish Jewish Population	<1 in 500 1 in 20	99% 99%	1 in 49,901 1 in 1,901	<1 in 10 million 1 in 152,080
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	Bardet-Biedl syndrome 13	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MKS1	Joubert syndrome 28	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MKS1	Meckel syndrome 1	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
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
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

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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
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 acidemia, MUT-related	AR	General Population East Asian Population Middle-Eastern Population	1 in 195 1 in 53 1 in 52	96% 96% 96%	1 in 4,851 1 in 1,301 1 in 1,276	1 in 3,783,780 1 in 275,812 1 in 265,408
MUT	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR	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 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
MYO7A	Non-syndromic hearing loss, MYO7A-related	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
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
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome), NDUFAF5-related	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
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome), NDUFS6-related	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
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
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 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
NPHP1	Nephronophthisis	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
NPHP1	NPHP1-related disorders	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
NPHP1	Senior-Løken syndrome 1	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
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 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
P3H1	Osteogenesis imperfecta, type VIII	AR	General Population West African Population African American Population	<1 in 500 1 in 67 1 in 250	99% 99% 99%	1 in 49,901 1 in 6,601 1 in 24,901	<1 in 10 million 1 in 1,769,068 <1 in 10,000,000
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	General Population Caucasian / European Population Middle-Eastern Population South East Asian	1 in 93 1 in 63 1 in 74 1 in 59	99% 99% 99% 99%	1 in 9,201 1 in 6,201 1 in 7,301 1 in 5,801	1 in 3,422,772 1 in 1,562,652 1 in 2,161,096 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

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PC	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
PCBD1	Tetrahydrobiopterin deficiency, PCBD1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PCCA	Propionic acidemia, PCCA-related	AR	General Population Native American Population	1 in 224 1 in 85	96% 96%	1 in 5,576 1 in 2,101	1 in 4,996,096 1 in 714,340
PCCB	Propionic acidemia, PCCB-related	AR	General Population Native American Population	1 in 224 1 in 85	99% 99%	1 in 22,301 1 in 8,401	<1 in 10 million 1 in 2,856,340
PCDH15	Non-syndromic hearing loss, PCDH15-related	AR	General Population Ashkenazi Jewish Population	1 in 395 1 in 72	98% 98%	1 in 19,701 1 in 3,551	1 in 78,804 1 in 14,204
PCDH15	Usher syndrome, type 1F	AR	General Population Ashkenazi Jewish Population	1 in 395 1 in 72	98% 98%	1 in 19,701 1 in 3,551	1 in 78,804 1 in 14,204
PDE6A	Retinitis pigmentosa, PDE6A-related	AR	General Population	1 in 133	99%	1 in 13,201	1 in 7,022,932
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 Japanese Population	1 in 500 1 in 354	95% 95%	1 in 9,981 1 in 7,061	<1 in 10 million 1 in 9,998,376
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 Ashkenazi Jewish Population	1 in 500 1 in 123	95% 95%	1 in 9,981 1 in 2,441	<1 in 10 million 1 in 1,200,972
PEX6	Zellweger syndrome, PEX6-related	AR	General Population Yemenite Jewish Population	1 in 280 1 in 18	99% 99%	1 in 27,901 1 in 1,701	<1 in 10 million 1 in 122,472
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 Ashkenazi Jewish Population	<1 in 500 1 in 120	99% 99%	1 in 49,901 1 in 11,901	<1 in 10 million 1 in 5,712,480
PGK1	Phosphoglycerate kinase 1 deficiency	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 280	98% 98%	1 in 24,951 1 in 13,951	<1 in 10 million <1 in 10 million
PKHD1	Polycystic kidney disease, PKHD1-related	AR	General Population Ashkenazi Jewish Population	1 in 70 1 in 107	98% 98%	1 in 3,451 1 in 5,301	1 in 966,280 1 in 2,268,828
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	Congenital disorder of glycosylation type 1a	AR	General Population Ashkenazi Jewish Population Caucasian / European Population	<1 in 500 1 in 57 1 in 71	99% 99% 99%	1 in 49,901 1 in 5,601 1 in 7,001	<1 in 10 million 1 in 1,277,028 1 in 1,988,284
POLG	Ataxia neuropathy spectrum	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Progressive external ophthalmoplegia	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Myocerebrohepatopathy syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	POLG-related disorders	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Alpers-Huttenlocher syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLR1C	Hypomyelinating Leukodystrophy, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POLR1C	Treacher Collins syndrome, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMGNT1	Muscular dystrophy-dystroglycanopathy	AR	General Population Finnish Population	1 in 462 1 in 111	98% 98%	1 in 23,051 1 in 5,501	<1 in 10 million 1 in 2,442,444
POMGNT1	Retinitis pigmentosa 76	AR	General Population Finnish Population	1 in 462 1 in 111	98% 98%	1 in 23,051 1 in 5,501	<1 in 10 million 1 in 2,442,444
POMT1	Dystroglycanopathy, POMT1-related	AR	General Population	1 in 290	99%	1 in 28,901	<1 in 10 million
POMT2	Muscular dystrophy-dystroglycanopathy, POMT2-related	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General Population Caucasian / European Population Finnish Population	1 in 368 1 in 488 1 in 75	98% 98% 98%	1 in 18,351 1 in 24,351 1 in 3,701	<1 in 10 million <1 in 10 million 1 in 1,110,300
PQBP1	Renpenning syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
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
PYGM	Glycogen storage disease type V	AR	General Population Caucasian / European Population	<1 in 500 1 in 206	99% 99%	1 in 49,901 1 in 20,501	<1 in 10 million <1 in 10 million
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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 137	98%	1 in 6,801	1 in 3,726,948
RAG2	Omenn syndrome, RAG2-related	AR	General Population	1 in 137	98%	1 in 6,801	1 in 3,726,948
RAPSN	Congenital myasthenic syndrome, RAPSN-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAPSN	Fetal akinesia deformation sequence	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
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 Caucasian / European Population	<1 in 500 1 in 456	98% 98%	1 in 24,951 1 in 22,751	<1 in 10 million <1 in 10 million
RMRP	Metaphyseal dysplasia without hypotrichosis	AR	General Population Amish Population Finnish Population	<1 in 500 1 in 16 1 in 76	99% 99% 99%	1 in 49,901 1 in 1,501 1 in 7,501	<1 in 10 million 1 in 96,064 1 in 2,280,304
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	General Population Amish Population Finnish Population	<1 in 500 <1 in 500 <1 in 500	99% 99% 99%	1 in 49,901 1 in 49,901 1 in 49,901	<1 in 10 million <1 in 10 million <1 in 10 million
RMRP	Anauxetic dysplasia	AR	General Population Amish Population Finnish Population	<1 in 500 1 in 16 1 in 76	99% 99% 99%	1 in 49,901 1 in 1,501 1 in 7,501	<1 in 10 million 1 in 96,064 1 in 2,280,304
RMRP	Cartilage-hair hypoplasia	AR	General Population Amish Population Finnish Population	<1 in 500 1 in 16 1 in 76	99% 99% 99%	1 in 49,901 1 in 1,501 1 in 7,501	<1 in 10 million 1 in 96,064 1 in 2,280,304
RPE65	Retinitis pigmentosa 20	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
RPE65	Leber congenital amaurosis 2	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
RPGRIP1L	COACH syndrome	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RPGRIP1L	Joubert syndrome 7	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RPGRIP1L	Meckel syndrome 5	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
RTEL1	Dyskeratosis congenita type 5	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 203	99% 99%	1 in 49,901 1 in 20,201	<1 in 10 million <1 in 10 million
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	General Population French Canadian Population	<1 in 500 1 in 19	95% 95%	1 in 9,981 1 in 361	<1 in 10 million 1 in 27,436
SAMHD1	Aicardi-Goutieres syndrome	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
SEPSECS	Pontocerebellar hypoplasia, type 2D	AR	General Population Moroccan/Iraqi Jewish Population	<1 in 500 1 in 44	99% 99%	1 in 49,901 1 in 4,301	<1 in 10 million 1 in 756,976
SERPINA1	Alpha-1 antitrypsin deficiency	AR	General Population Caucasian / European Population	1 in 33 1 in 19	95% 95%	1 in 641 1 in 361	1 in 84,612 1 in 27,436
SGCA	Limb-girdle muscular dystrophy, type 2D	AR	General Population Caucasian / European Population Finnish Population	<1 in 500 1 in 288 1 in 150	98% 98% 98%	1 in 24,951 1 in 14,351 1 in 7,451	<1 in 10 million <1 in 10 million 1 in 4,470,600
SGCB	Limb-girdle muscular dystrophy, type 2E	AR	General Population Caucasian / European Population	1 in 500 1 in 406	98% 98%	1 in 24,951 1 in 20,251	<1 in 10 million <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 Moroccan Population Roma / Gypsy Population	1 in 381 1 in 250 1 in 96	98% 98% 98%	1 in 19,001 1 in 12,451 1 in 4,751	<1 in 10 million <1 in 10 million 1 in 1,824,384
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR	General Population Caucasian / European Population	1 in 454 1 in 253	98% 98%	1 in 22,651 1 in 12,601	<1 in 10 million <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
SLC12A3	Gitelman syndrome	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
SLC12A6	Andermann syndrome	AR	General Population French Canadian Population	<1 in 500 1 in 23	98% 99%	1 in 24,951 1 in 2,201	<1 in 10 million 1 in 202,492
SLC16A2	Allan-Herndon-Dudley syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC17A5	Sialic acid storage disorder	AR	General Population Finnish Population	<1 in 500 1 in 100	91% 91%	1 in 5,545 1 in 1,101	<1 in 10 million 1 in 440,400
SLC22A5	Systemic primary carnitine deficiency	AR	General Population African/African American Population East Asian Population Faroese Population Pacific Islander Population South Asian/Indian Population	1 in 129 1 in 86 1 in 77 1 in 9 1 in 37 1 in 51	76% 76% 76% 76% 76% 76%	1 in 534 1 in 355 1 in 318 1 in 34 1 in 151 1 in 209	1 in 275,544 1 in 122,120 1 in 97,944 1 in 1,224 1 in 22,348 1 in 42,636

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
SLC25A13	Citrin deficiency	AR	General Population East Asian Population	<1 in 500 1 in 65	95% 95%	1 in 9,981 1 in 1,281	<1 in 10 million 1 in 333,060
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR	General Population French Canadian Population	<1 in 500 1 in 37	99% 99%	1 in 49,901 1 in 3,601	<1 in 10 million 1 in 532,948
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC26A2	Diastrophic dysplasia	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A2	Achondrogenesis, type IB	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A2	Multiple epiphyseal dysplasia	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A2	Atelosteogenesis II	AR	General Population Finnish Population	1 in 158 1 in 50	90% 90%	1 in 1,571 1 in 491	1 in 992,872 1 in 98,200
SLC26A3	Congenital secretory chloride diarrhea	AR	General Population Middle-Eastern Population	<1 in 500 1 in 57	98% 98%	1 in 24,951 1 in 2,801	<1 in 10 million 1 in 638,628
SLC26A4	Pendred syndrome	AR	General Population African/African American Population Caucasian / European Population East Asian Population	1 in 80 1 in 76 1 in 88 1 in 74	98% 98% 98% 98%	1 in 3,951 1 in 3,751 1 in 4,351 1 in 3,651	1 in 1,264,320 1 in 1,140,304 1 in 1,531,552 1 in 1,080,696
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 453	98% 98%	1 in 24,951 1 in 22,601	<1 in 10 million <1 in 10 million
SLC37A4	Glycogen storage disease, type Ib	AR	General Population Ashkenazi Jewish Population	1 in 158 1 in 71	95% 95%	1 in 3,141 1 in 1,401	1 in 1,985,112 1 in 397,884
SLC39A4	Acrodermatitis enteropathica	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC46A1	Hereditary folate malabsorption	AR	General Population Puerto Rican Population	<1 in 500 1 in 500	99% 99%	1 in 49,901 1 in 49,901	<1 in 10 million <1 in 10 million
SLC4A11	Corneal endothelial dystrophy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC6A19	Hartnup disorder	AR	General Population	1 in 87	99%	1 in 8,601	1 in 2,993,148
SLC7A7	Lysinuric protein intolerance	AR	General Population Finnish Population Japanese Population	<1 in 500 1 in 122 1 in 119	95% 95% 95%	1 in 9,981 1 in 2,421 1 in 2,361	<1 in 10 million 1 in 1,181,448 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 African/African American Population Ashkenazi Jewish Population Caucasian / European Population East Asian Population Latino Population Sephardic Jewish Population	1 in 54 1 in 72 1 in 67 1 in 47 1 in 59 1 in 68 1 in 34	91% 71% 91% 95% 93% 90% 96%	1 in 590 1 in 246 1 in 734 1 in 921 1 in 830 1 in 671 1 in 826	1 in 127,440 1 in 70,848 1 in 196,712 1 in 173,148 1 in 195,880 1 in 182,512 1 in 112,336
SMPD1	Niemann-Pick disease, type A/B	AR	General Population Ashkenazi Jewish Population Latino Population	1 in 250 1 in 115 1 in 106	95% 95% 95%	1 in 4,981 1 in 2,281 1 in 2,101	1 in 4,981,000 1 in 1,049,260 1 in 890,824
SPG11	SPG11-related Neuromuscular Disorders	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
SPG7	Spastic paraplegia type 7	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SUMF1	Multiple sulfatase deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 320	98% 98%	1 in 24,951 1 in 15,951	<1 in 10 million <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
TAT	Tyrosinemia, type II	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TCIRG1	Osteopetrosis, TCIRG1-related	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TECPR2	Spastic paraplegia 49	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TFR2	Hemochromatosis, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
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 224	98%	1 in 11,151	1 in 9,991,296
TMEM216	Joubert syndrome 2	AR	General Population Ashkenazi Jewish Population	1 in 141 1 in 92	98% 98%	1 in 7,001 1 in 4,551	1 in 3,948,564 1 in 1,674,768
TMEM216	Meckel syndrome 2	AR	General Population Ashkenazi Jewish Population	1 in 141 1 in 92	98% 98%	1 in 7,001 1 in 4,551	1 in 3,948,564 1 in 1,674,768

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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 French Canadian Population	1 in 252 1 in 53	97% 97%	1 in 8,368 1 in 1,734	1 in 8,434,944 1 in 367,608
TRDN	Catecholaminergic polymorphic ventricular tachycardia, TRDN-related	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
TRIM32	Limb-girdle muscular dystrophy, type 2H	AR	General Population Hutterite Population	<1 in 500 1 in 12	98% 98%	1 in 24,951 1 in 551	<1 in 10 million 1 in 26,448
TRIM32	Bardet-Biedl syndrome 11	AR	General Population Hutterite Population	<1 in 500 1 in 12	98% 98%	1 in 24,951 1 in 551	<1 in 10 million 1 in 26,448
TRMU	Liver failure, acute infantile	AR	General Population Yemeni Jewish Population	<1 in 500 1 in 34	98% 98%	1 in 24,951 1 in 1,651	<1 in 10 million 1 in 224,536
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR	General Population Finnish Population	<1 in 500 1 in 80	98% 98%	1 in 24,951 1 in 3,951	<1 in 10 million 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 Caucasian / European Population	<1 in 500 1 in 267	98% 90%	1 in 24,951 1 in 2,661	<1 in 10 million 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
UGT1A1	Crigler-Najjar syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
USH1C	Usher syndrome, type IC	AR	General Population French Canadian Population	1 in 353 1 in 227	90% 90%	1 in 3,521 1 in 2,261	1 in 4,971,652 1 in 2,052,988
USH1C	Non-syndromic hearing loss, USH1C-related	AR	General Population French Canadian Population	1 in 353 1 in 227	90% 90%	1 in 3,521 1 in 2,261	1 in 4,971,652 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
USH2A	Usher syndrome, type 2A	AR	General Population Caucasian / European Population Ashkenazi Jewish Population Iranian Jewish Population	1 in 126 1 in 73 1 in 35 1 in 60	96% 96% 99% 99%	1 in 3,126 1 in 1,801 1 in 3,401 1 in 5,901	1 in 1,575,504 1 in 525,892 1 in 476,140 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
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
WNT10A	Schopf-Schulz-Passarge syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
WNT10A	Odontoonychodermal dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
XPA	Xeroderma pigmentosum, group A	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
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 alpha thalassemia trait cis is described in rows marked with a dagger symbol.
Abbreviations: AR, autosomal recessive; XL, X-linked