



Patient Information:

DOB: [REDACTED]
Sex: M
MR#: BFA 0184
Patient#: [REDACTED]

Partner Information:

Not Tested

Physician:

Shaikly, Valerie
ATTN: Shaikly, Valerie
Fertility Genetics
1 Lanswood Park
Elmstead Market, Essex CO7 7FD GB
Phone: 7711197938

Laboratory:

Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: Apr 01, 2023

Accession:

Test#: [REDACTED]
Specimen Type: Saliva Swab
Collected: Mar 10, 2023

Accession:

N/A

FINAL RESULTS



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

TEST PERFORMED

176 Matched Fors Male with XL
(177 Gene Panel; gene sequencing
with deletion and duplication analysis)

Condition and Gene

Inheritance



Partner

Congenital disorder of glycosylation type 1a
PMM2

AR

Carrier
c.422G>A (p.Arg141His)

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 the risk for any autosomal recessive conditions:
 - There is a 1/252 chance of having a child affected with Congenital disorder of glycosylation type 1a, a *PMM2*-related condition.
- 33 CGG repeats were observed in this individual.
- 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.



CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1A

| | | |
|-----------------|---|---------|
| Patient |  | Partner |
| Result |  Carrier | N/A |
| Variant Details | PMM2 (NM_000303.3) c.422G>A (p.Arg141His) | N/A |

What is Congenital disorder of glycosylation type 1a?

Congenital disorder of glycosylation, type 1a (CDG1a) is caused by the production of abnormally-functioning proteins in many organs and tissues, which means that the disease affects many body systems. Infants with this disorder may have hypotonia, an abnormal distribution of fat, strabismus, developmental delay, and failure to thrive. Infants with CDG1a also frequently have an underdeveloped cerebellum that can lead to seizures and intellectual disability. About 20 percent of infants with CDG1a do not survive their first year of life due to complications of the disease. Occasionally the disease does not show signs until childhood or adulthood, in which case symptoms are often milder.

What is my risk of having an affected child?

CDG1a is inherited in an autosomal recessive manner. This means that if both parents are carriers of the disease, the risk of having an affected child is 1 in 4 (25%). The overall risk of being a carrier for CDG1a is very low, with a carrier frequency of less than 1 in 500. Individuals of Ashkenazi Jewish descent have an increased carrier risk of 1 in 57. Individuals of Caucasian/European descent have an increased carrier risk of 1 in 71.

What kind of medical management is available?

Many affected babies die before birth or are stillborn. Affected individuals who survive infancy may have moderate intellectual disability, and some are unable to walk independently. Affected individuals may also experience stroke-like episodes. There is no specific treatment for the disorder itself, but management is mostly symptom-based and can include anti-epileptic drugs to prevent seizures, occupational therapy and physical therapy for developmental delay, and the use of a nasogastric tube or gastrostomy tube to ensure appropriate caloric intake.

What mutation was detected?

The detected heterozygous variant was NM_000303.3:c.422G>A (p.Arg141His). This variant, p.Arg141His, has been identified in the compound heterozygous state in multiple unrelated individuals with CDG1a (Congenital Disorder Of Glycosylation, Type 1A) (PubMed: [9140401](#), [9781039](#), [10527672](#), [10922383](#), [11916319](#), [17166182](#), [9497260](#), [11517108](#), [25355454](#), [19357119](#)). It is one of the most common mutations found in patients with CDG1a, with a carrier frequency of approximately 1 in 70 individuals in the Dutch and Danish populations (PubMed: [10854097](#)). In vitro functional studies using protein expressed from bacteria showed that enzymatic activity was completely abolished in the presence of this variant (PubMed: [21541725](#)). Furthermore, a mouse model demonstrated that mouse embryos compound heterozygous or homozygous for this variant (p.Arg137His in the homologous mouse gene) either died as embryos or were unable to survive postnatally and that supplementation with mannose did not reverse this effect (PubMed: [27053713](#)). This variant is classified as "Pathogenic" in ClinVar, with multiple submitters in agreement (ClinVar: 7706). The laboratory classifies this variant as pathogenic.



GENES TESTED:

176 Matched Fors Male with XL - 177 Genes

This analysis was run using the 176 Matched Fors Male with XL gene list. 177 genes were tested with 99.20% of targets sequenced at >20x coverage. For more gene specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

| | | | | | |
|---------|---------|---------|---------|---------|---------|
| ABCC8 | ABCD1 | ACADM | ACADS | ACADVL | ADA |
| AGA | AGL | AGXT | AIRE | ALDH3A2 | ALDOB |
| ALG6 | ALMS1 | ALPL | AMT | ARG1 | ARSA |
| ASL | ASPA | ASS1 | ATM | ATP7A | ATP7B |
| BBS1 | BBS10 | BBS12 | BBS2 | BCKDHA | BCKDHB |
| BCS1L | BLM | BTD | CAPN3 | CBS | CFTR |
| CLN3 | CLN5 | CLN6 | CLN8 | CLRN1 | COL4A3 |
| COL4A4 | CPS1 | CPT1A | CPT2 | CRYL1 | CTNS |
| CTSK | CYP11B1 | CYP21A2 | CYP27A1 | DBT | DHCR7 |
| DLD | DMD | DYSF | ELP1 | ERCC6 | ERCC8 |
| EVC | EVC2 | FAH | FANCA | FANCC | FKRP |
| FKTN | FMR1 | G6PC | GAA | GALC | GALK1 |
| GALT | GBA | GCDH | GJB2 | GJB6 | GLA |
| GLB1 | GLDC | GNE | GNPTAB | GNPTG | GRHPR |
| HADHA | HBA1 | HBA2 | HBB | HEXA | HEXB |
| HGSNAT | HLCS | HMGCL | HOGA1 | HSD17B4 | HYLS1 |
| IDS | IDUA | IL2RG | IVD | KCNJ11 | LAMA2 |
| LAMA3 | LAMB3 | LAMC2 | LIPA | LRPPRC | MAN2B1 |
| MCOLN1 | MEFV | MESP2 | MKS1 | MLC1 | MMAA |
| MMAB | MMACHC | MPI | MUT | MYO7A | NAGLU |
| NBN | NEB | NPC1 | NPC2 | NPHS1 | NPHS2 |
| NR0B1 | OPA3 | OTC | PAH | PC | PCCA |
| PCCB | PCDH15 | PEX1 | PEX10 | PEX12 | PEX2 |
| PEX6 | PEX7 | PKHD1 | PMM2 | POMGNT1 | PPT1 |
| PROP1 | PTS | RMRP | RS1 | RTEL1 | SACS |
| SGCA | SGCB | SGCD | SGCG | SGSH | SLC12A6 |
| SLC17A5 | SLC22A5 | SLC26A2 | SLC26A4 | SLC37A4 | SMN1 |
| SMPD1 | STAR | TAT | TCIRG1 | TGM1 | TH |
| TMEM216 | TPP1 | TTPA | USH1C | USH2A | VPS13B |
| 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, 99.23% and 99.20% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.



LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

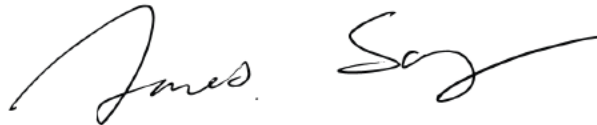
Gene Specific Notes and Limitations

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. **CFTR**: Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. **CRYL1**: As mutations in the CRYL1 gene are not known to be associated with any clinical condition, sequence variants in this gene are not analyzed. However, to increase copy number detection sensitivity for large deletions including this gene and a neighboring gene on the panel (GJB6, also known as connexin 30), this gene was evaluated for copy number variation. **CYP11B1**: The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **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. **DMD**: Single exon deletion/duplication analysis is limited to exons with >1 patient reported in the UMD database (http://www.umd.be/DMD/W_DMD/index.html), accessed Dec 29,2020 and all out-of-frame exons after exon 3. This includes deletion of exon 1, and duplication of exon 2, and del/dup for exons 3,6~8,11,12,17~22,43~46,48,50~56,58~63,65~70,75,76 and 78. Single-exon detection is limited to blood samples. **FMR1**: The exact size of alleles >200 CGG repeats cannot be determined; these alleles are pathogenic for X-Linked Fragile X Syndrome. Alleles with <10 repeats may fail to amplify; these alleles are benign. The repeat length for this gene may vary by +/- 1 repeat unit. Methylation is not analyzed. RP-PCR analysis of the FMR1 promoter is not routinely performed in males. Small degrees of size mosaicism, including gonadal mosaicism, may not be



detected. GALT: In general, the D2 "Duarte" allele is not reported if detected, but can be reported upon request. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state (PubMed: 25473725, 30593450). GBA: The current testing method may not be able to reliably detect certain pathogenic variants in the GBA gene due to homologous recombination between the pseudogene and the functional gene. HBA1: The phase of heterozygous alterations in the HBA1 gene cannot be determined, but can be confirmed through parental testing. HBA2: The phase of heterozygous alterations in the HBA2 gene cannot be determined, but can be confirmed through parental testing. NEB: This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis. NPHS2: If detected, the variant NM_014625.3:c.686G>A (p.Arg229Gln) will not be reported as this variant is not significantly associated with disease when homozygous or in the compound heterozygous state with variants in exons 1-6 of NPHS2. 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.

SIGNATURE:



Jianbo Song, Ph.D., ABMGG, CGMB, CCS, FACMG on 4/1/2023 03:58 PM PDT
Electronically signed

DISCLAIMER:

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



Supplemental Table

| Gene | Condition | Inheritance | Ethnicity | Carrier Rate | Detection Rate | Post-test Carrier Probability* | Residual Risk* |
|---------|---|-------------|-------------------------------------|--------------|----------------|--------------------------------|------------------|
| 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 |
| ABCD1 | Adrenoleukodystrophy, X-linked | XL | General Population | 1 in 21,000 | 99% | 1 in 2,099,901 | 1 in 8,399,804 |
| 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 |
| 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 |
| ADA | Adenosine deaminase deficiency | AR | General Population | 1 in 224 | 93% | 1 in 3,187 | 1 in 2,855,552 |
| 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 |
| 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 |
| 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 |
| 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 |
| ARG1 | Arginase deficiency | AR | General Population | 1 in 296 | 98% | 1 in 14,751 | <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 |
| ASL | Argininosuccinate lyase deficiency | AR | General Population | 1 in 132 | 90% | 1 in 1,311 | 1 in 692,208 |
| 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 |
| ATP7A | Menkes disease | XL | General Population | 1 in 50,000 | 99% | 1 in 4,999,901 | <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 | Bardet-Biedl syndrome 2 | AR | General Population | 1 in 621 | 99% | 1 in 62,001 | <1 in 10 million |
| | | | Ashkenazi Jewish Population | 1 in 107 | 99% | 1 in 10,601 | 1 in 4,537,228 |
| BBS2 | Retinitis Pigmentosa 74 | AR | General Population | 1 in 621 | 99% | 1 in 62,001 | <1 in 10 million |
| | | | Ashkenazi Jewish Population | 1 in 107 | 99% | 1 in 10,601 | 1 in 4,537,228 |

Patient: [REDACTED] Sex: M;
 DOB: [REDACTED] MR#: BFA 0184

Accession#: [REDACTED] FD Patient#: [REDACTED]
 DocID: [REDACTED] PAGE 6 of 13



Supplemental Table

| Gene | Condition | Inheritance | Ethnicity | Carrier Rate | Detection Rate | Post-test Carrier Probability* | Residual Risk* |
|---------|--|-------------|-------------------------------------|--------------|----------------|--------------------------------|------------------|
| 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 | 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 | 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 |
| BTD | Biotinidase deficiency | AR | General Population | 1 in 124 | 99% | 1 in 12,301 | 1 in 6,101,296 |
| | | | Caucasian / European Population | 1 in 71 | 99% | 1 in 7,001 | 1 in 1,988,284 |
| | | | Latino Population | 1 in 136 | 99% | 1 in 13,501 | 1 in 7,344,544 |
| | | | Middle-Eastern Population | 1 in 55 | 99% | 1 in 5,401 | 1 in 1,188,220 |
| 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 |
| 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 |
| 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 |
| | | | General Population | 1 in 230 | 98% | 1 in 11,451 | <1 in 10 million |
| CLN3 | Neuronal ceroid lipofuscinosis | AR | Finnish Population | 1 in 72 | 98% | 1 in 3,551 | 1 in 1,022,688 |
| | | | General Population | <1 in 500 | 95% | 1 in 9,981 | <1 in 10 million |
| CLN5 | Neuronal ceroid lipofuscinosis 5 | AR | Finnish Population | 1 in 115 | 95% | 1 in 2,281 | 1 in 1,049,260 |
| | | | General Population | <1 in 500 | 92% | 1 in 6,239 | <1 in 10 million |
| CLN6 | Neuronal ceroid lipofuscinosis, CLN6-related | AR | General Population | <1 in 500 | 95% | 1 in 9,981 | <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 |
| CLRN1 | Usher syndrome, type 3A | AR | Finnish Population | 1 in 135 | 95% | 1 in 2,681 | 1 in 1,447,740 |
| | | | 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 |
| COL4A3 | Alport syndrome, COL4A3-related | AR | Finnish Population | 1 in 70 | 98% | 1 in 3,451 | 1 in 966,280 |
| | | | 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 |
| 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 |
| 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 |
| 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 |
| | | | Moroccan Jewish Population | 1 in 35 | 98% | 1 in 1,701 | 1 in 238,140 |
| 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 |
| | | | Moroccan 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 |
| 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 |
| DLSD | 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 |
| DMD | Duchenne Muscular Dystrophy | XL | General Population | 1 in 2,350 | 93% | 1 in 33,558 | 1 in 134,260 |
| DMD | Becker Muscular Dystrophy | XL | General Population | 1 in 2,350 | 93% | 1 in 33,558 | 1 in 134,260 |

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Supplemental Table

| Gene | Condition | Inheritance | Ethnicity | Carrier Rate | Detection Rate | Post-test Carrier Probability* | Residual Risk* |
|-------|---|-------------|-------------------------------------|--------------|----------------|--------------------------------|------------------|
| 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 |
| 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 |
| ERCC6 | De Sanctis-Cacchione syndrome | AR | General Population | 1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| | | | Japanese Population | 1 in 74 | 99% | 1 in 7,301 | 1 in 2,161,096 |
| ERCC6 | Cockayne syndrome type B | AR | General Population | 1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| | | | Japanese Population | 1 in 74 | 99% | 1 in 7,301 | 1 in 2,161,096 |
| ERCC8 | Cockayne syndrome type A | AR | General Population | 1 in 822 | 98% | 1 in 41,051 | <1 in 10 million |
| EVC | Weyers acrofacial dysostosis, EVC-related | AR | General Population | 1 in 142 | 98% | 1 in 7,051 | 1 in 4,004,968 |
| | | | Amish Population | 1 in 7 | 98% | 1 in 301 | 1 in 8,428 |
| EVC | Ellis-van Creveld syndrome, EVC-related | AR | General Population | 1 in 142 | 98% | 1 in 7,051 | 1 in 4,004,968 |
| | | | Amish Population | 1 in 7 | 98% | 1 in 301 | 1 in 8,428 |
| EVC2 | Weyers acrofacial dysostosis, EVC2-related | AR | General Population | 1 in 240 | 98% | 1 in 11,951 | <1 in 10 million |
| | | | Amish Population | 1 in 7 | 98% | 1 in 301 | 1 in 8,428 |
| EVC2 | Ellis-van Creveld syndrome, EVC2-related | AR | General Population | 1 in 240 | 98% | 1 in 11,951 | <1 in 10 million |
| | | | Amish Population | 1 in 7 | 98% | 1 in 301 | 1 in 8,428 |
| 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 |
| 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 |
| 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 | <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 |
| 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 |
| FMR1 | Fragile X Syndrome Intermediate Allele | XL | General Population | 1 in 259 | 99% | 1 in 25,801 | 1 in 103,204 |
| | | | Ashkenazi Jewish Population | 1 in 115 | 99% | 1 in 11,401 | 1 in 45,604 |
| FMR1 | Fragile X Syndrome Premutation | XL | General Population | 1 in 259 | 99% | 1 in 25,801 | 1 in 103,204 |
| | | | Ashkenazi Jewish Population | 1 in 115 | 99% | 1 in 11,401 | 1 in 45,604 |
| 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 |
| 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 |
| 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 |
| 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 |
| 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 |

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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 |
| GLA | Fabry disease | XL | General Population | 1 in 25,000 | 99% | 1 in 2,499,901 | 1 in 9,999,804 |
| 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 |
| GNE | Inclusion body myopathy type 2 (Nonaka myopathy) | AR | General Population | <1 in 500 | 99% | 1 in 49,901 | 1 in 99,802,000 |
| | | | Iranian Jewish Population | 1 in 11 | 99% | 1 in 1,001 | 1 in 44,044 |
| GNPTAB | Mucopolipidosis II alpha/beta | AR | General Population | <1 in 500 | 95% | 1 in 9,981 | <1 in 10 million |
| GNPTAB | Mucopolipidosis III alpha/beta | AR | General Population | <1 in 500 | 95% | 1 in 9,981 | <1 in 10 million |
| GNPTG | Mucopolipidosis III gamma | AR | General Population | <1 in 500 | 95% | 1 in 9,981 | <1 in 10 million |
| GRHPR | Primary hyperoxaluria type II | AR | General Population | <1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| HADHA | Trifunctional protein deficiency | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| | | | Finnish Population | 1 in 124 | 98% | 1 in 6,151 | 1 in 3,050,896 |
| HADHA | Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| | | | Finnish Population | 1 in 124 | 98% | 1 in 6,151 | 1 in 3,050,896 |
| 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 |
| 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 |
| | | | 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 |
| 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* |
|----------------|--|-------------|---|--|--------------------------|--|--|
| <i>HGSNAT</i> | Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C) | AR | General Population Caucasian / European Population | 1 in 434 1 in 345 | 98% 98% | 1 in 21,651 1 in 17,201 | <1 in 10 million <1 in 10 million |
| <i>HLCS</i> | Holocarboxylase synthetase deficiency | AR | General Population | 1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| <i>HMGCL</i> | 3-hydroxy-3-methylglutaryl-CoA lyase deficiency | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| <i>HOGA1</i> | Primary hyperoxaluria type III | AR | General Population | 1 in 184 | 99% | 1 in 18,301 | <1 in 10 million |
| <i>HSD17B4</i> | D-bifunctional protein deficiency | AR | General Population | 1 in 158 | 98% | 1 in 7,851 | 1 in 4,961,832 |
| <i>HYLS1</i> | Hydrolethalus syndrome | AR | General Population Finnish Population | <1 in 500 1 in 50 | 98% 98% | 1 in 24,951 1 in 2,451 | <1 in 10 million 1 in 490,200 |
| <i>IDS</i> | Mucopolysaccharidosis type II (Hunter syndrome) | XL | General Population | 1 in 50,000 | 91% | 1 in 555,545 | 1 in 2,222,204 |
| <i>IDUA</i> | Mucopolysaccharidosis, type I (Hurler syndrome) | AR | General Population Caucasian / European Population | <1 in 500 1 in 153 | 95% 95% | 1 in 9,981 1 in 3,041 | <1 in 10 million 1 in 1,861,092 |
| <i>IL2RG</i> | Severe combined immunodeficiency, X-linked | XL | General Population | 1 in 25,000 | 99% | 1 in 2,499,901 | 1 in 9,999,804 |
| <i>IVD</i> | Isovaleric Acidemia | AR | General Population African/African American Population Caucasian / European Population East Asian Population | 1 in 167 1 in 100 1 in 115 1 in 407 | 90% 90% 90% 90% | 1 in 1,661 1 in 991 1 in 1,141 1 in 4,061 | 1 in 1,109,548 1 in 396,400 1 in 524,860 1 in 6,611,308 |
| <i>KCNJ11</i> | 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 |
| <i>KCNJ11</i> | 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 |
| <i>LAMA2</i> | Muscular dystrophy, LAMA2-related | AR | General Population Caucasian / European Population | <1 in 500 1 in 125 | 99% 99% | 1 in 49,901 1 in 12,401 | <1 in 10 million 1 in 6,200,500 |
| <i>LAMA3</i> | Junctional epidermolysis bullosa, LAMA3-related | AR | General Population | 1 in 781 | 98% | 1 in 39,001 | <1 in 10 million |
| <i>LAMA3</i> | Laryngo-onycho-cutaneous syndrome | AR | General Population | 1 in 781 | 98% | 1 in 39,001 | <1 in 10 million |
| <i>LAMB3</i> | Junctional epidermolysis bullosa, LAMB3-related | AR | General Population | 1 in 781 | 98% | 1 in 39,001 | <1 in 10 million |
| <i>LAMC2</i> | Junctional epidermolysis bullosa, LAMC2-related | AR | General Population | 1 in 781 | 98% | 1 in 39,001 | <1 in 10 million |
| <i>LIPA</i> | 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 |
| <i>LRPPRC</i> | Leigh syndrome with Complex IV deficiency | AR | General Population Faroese Population French Canadian Population | 1 in 447 1 in 21 1 in 22 | 98% 98% 98% | 1 in 22,301 1 in 1,001 1 in 1,051 | <1 in 10 million 1 in 84,084 1 in 92,488 |
| <i>MAN2B1</i> | Alpha-Mannosidosis | AR | General Population Caucasian / European Population | 1 in 354 1 in 274 | 99% 99% | 1 in 35,301 1 in 27,301 | <1 in 10 million <1 in 10 million |
| <i>MCOLN1</i> | Mucopolipidosis IV | AR | General Population Ashkenazi Jewish Population | 1 in 300 1 in 100 | 99% 99% | 1 in 29,901 1 in 9,901 | <1 in 10 million 1 in 3,960,400 |
| <i>MEFV</i> | Familial Mediterranean fever | AR | General Population Mediterranean Population | 1 in 20 1 in 7 | 99% 90% | 1 in 1,901 1 in 61 | 1 in 152,080 1 in 1,708 |
| <i>MESP2</i> | Spondylocostal dysostosis | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| <i>MKS1</i> | 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 |
| <i>MKS1</i> | 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 |
| <i>MKS1</i> | 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 |
| <i>MLC1</i> | Megalencephalic leukoencephalopathy with subcortical cysts | AR | General Population Libyan Jewish Population | <1 in 500 1 in 40 | 99% 99% | 1 in 49,901 1 in 3,901 | <1 in 10 million 1 in 624,160 |
| <i>MMAA</i> | Methylmalonic aciduria, cblA type | AR | General Population | 1 in 301 | 97% | 1 in 10,001 | <1 in 10 million |
| <i>MMAB</i> | Methylmalonic aciduria, cblB type | AR | General Population | 1 in 435 | 98% | 1 in 21,701 | <1 in 10 million |
| <i>MMACHC</i> | Methylmalonic aciduria and homocystinuria, cblC type | AR | General Population | 1 in 134 | 90% | 1 in 1,331 | 1 in 713,416 |
| <i>MPI</i> | Congenital disorder of glycosylation type Ib | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| <i>MUT</i> | Methylmalonic aciduria-methylmalonyl-CoA mutase deficiency | AR | General Population | 1 in 100 | 99% | 1 in 9,901 | 1 in 3,960,400 |
| <i>MYO7A</i> | 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 |
| <i>MYO7A</i> | 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 |
| <i>NAGLU</i> | Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B) | AR | General Population Caucasian / European Population East Asian Population | <1 in 500 1 in 346 1 in 298 | 99% 99% 99% | 1 in 49,901 1 in 34,501 1 in 29,701 | <1 in 10 million <1 in 10 million <1 in 10 million |
| <i>NBN</i> | Nijmegen breakage syndrome | AR | General Population | 1 in 158 | 99% | 1 in 15,701 | 1 in 9,923,032 |

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Supplemental Table

| Gene | Condition | Inheritance | Ethnicity | Carrier Rate | Detection Rate | Post-test Carrier Probability* | Residual Risk* |
|---------|---|-------------|---------------------------------|--------------|----------------|--------------------------------|------------------|
| NEB | Nemaline myopathy | AR | General Population | 1 in 112 | 98% | 1 in 5,551 | 1 in 2,486,848 |
| | | | Amish Population | 1 in 11 | 98% | 1 in 501 | 1 in 22,044 |
| | | | Ashkenazi Jewish Population | 1 in 108 | 98% | 1 in 5,351 | 1 in 2,311,632 |
| | | | Finnish Population | 1 in 112 | 98% | 1 in 5,551 | 1 in 2,486,848 |
| NPC1 | Niemann-Pick disease, type C1 | AR | General Population | 1 in 194 | 90% | 1 in 1,931 | 1 in 1,498,456 |
| NPC2 | Niemann-Pick disease, type C2 | AR | General Population | 1 in 194 | 99% | 1 in 19,301 | <1 in 10 million |
| NPHS1 | Congenital nephrotic syndrome, type 1 | AR | General Population | 1 in 289 | 98% | 1 in 14,401 | <1 in 10 million |
| | | | Finnish Population | 1 in 50 | 98% | 1 in 2,451 | 1 in 490,200 |
| NPHS2 | Congenital nephrotic syndrome, type 2 | AR | General Population | 1 in 289 | 98% | 1 in 14,401 | <1 in 10 million |
| | | | Finnish Population | 1 in 50 | 98% | 1 in 2,451 | 1 in 490,200 |
| NR0B1 | Congenital adrenal hypoplasia, X-linked | XL | General Population | 1 in 6,250 | 99% | 1 in 624,901 | 1 in 2,499,804 |
| OPA3 | Costeff syndrome | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| | | | Iraqi Jewish Population | 1 in 50 | 98% | 1 in 2,451 | 1 in 490,200 |
| OTC | Ornithine transcarbamylase deficiency | XL | General Population | 1 in 7,000 | 90% | 1 in 69,991 | 1 in 279,984 |
| 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 |
| PC | Pyruvate carboxylase deficiency | AR | General Population | 1 in 250 | 95% | 1 in 4,981 | 1 in 4,981,000 |
| PCCA | Propionic acidemia, PCCA-related | AR | General Population | 1 in 224 | 96% | 1 in 5,576 | 1 in 4,996,096 |
| | | | Native American Population | 1 in 85 | 96% | 1 in 2,101 | 1 in 714,340 |
| PCCB | Propionic acidemia, PCCB-related | AR | General Population | 1 in 224 | 99% | 1 in 22,301 | <1 in 10 million |
| | | | Native American Population | 1 in 85 | 99% | 1 in 8,401 | 1 in 2,856,340 |
| PCDH15 | Non-syndromic hearing loss, PCDH15-related | AR | General Population | 1 in 395 | 98% | 1 in 19,701 | 1 in 78,804 |
| | | | Ashkenazi Jewish Population | 1 in 72 | 98% | 1 in 3,551 | 1 in 14,204 |
| PCDH15 | Usher syndrome, type 1F | AR | General Population | 1 in 395 | 98% | 1 in 19,701 | 1 in 78,804 |
| | | | Ashkenazi Jewish Population | 1 in 72 | 98% | 1 in 3,551 | 1 in 14,204 |
| 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 |
| 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 |
| 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 |
| PMM2 | Congenital disorder of glycosylation type 1a | 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 |
| POMGNT1 | Muscular dystrophy-dystroglycanopathy | AR | General Population | 1 in 462 | 98% | 1 in 23,051 | <1 in 10 million |
| | | | Finnish Population | 1 in 111 | 98% | 1 in 5,501 | 1 in 2,442,444 |
| POMGNT1 | Retinitis pigmentosa 76 | AR | General Population | 1 in 462 | 98% | 1 in 23,051 | <1 in 10 million |
| | | | Finnish Population | 1 in 111 | 98% | 1 in 5,501 | 1 in 2,442,444 |
| 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 |
| PROP1 | Combined pituitary hormone deficiency 2 | AR | General Population | 1 in 45 | 98% | 1 in 2,201 | 1 in 396,180 |
| PTS | Tetrahydrobiopterin deficiency | AR | General Population | 1 in 354 | 96% | 1 in 8,826 | <1 in 10 million |
| RMRP | Metaphyseal dysplasia without hypotrichosis | AR | General Population | <1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| | | | Amish Population | 1 in 16 | 99% | 1 in 1,501 | 1 in 96,064 |
| | | | Finnish Population | 1 in 76 | 99% | 1 in 7,501 | 1 in 2,280,304 |
| 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 500 | 99% | 1 in 49,901 | <1 in 10 million |
| | | | Finnish Population | <1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| RMRP | Anauxetic dysplasia | AR | General Population | <1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| | | | Amish Population | 1 in 16 | 99% | 1 in 1,501 | 1 in 96,064 |
| | | | Finnish Population | 1 in 76 | 99% | 1 in 7,501 | 1 in 2,280,304 |
| RMRP | Cartilage-hair hypoplasia | AR | General Population | <1 in 500 | 99% | 1 in 49,901 | <1 in 10 million |
| | | | Amish Population | 1 in 16 | 99% | 1 in 1,501 | 1 in 96,064 |
| | | | Finnish Population | 1 in 76 | 99% | 1 in 7,501 | 1 in 2,280,304 |
| RS1 | Juvenile retinoschisis, X-linked | XL | General Population | 1 in 2,500 | 96% | 1 in 62,476 | 1 in 249,956 |

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Supplemental Table

| Gene | Condition | Inheritance | Ethnicity | Carrier Rate | Detection Rate | Post-test Carrier Probability* | Residual Risk* |
|---------|---|-------------|-------------------------------------|--------------|----------------|--------------------------------|------------------|
| 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 |
| 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 |
| SLC12A6 | Andermann syndrome | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| SLC17A5 | Sialic acid storage disorder | AR | General Population | <1 in 500 | 91% | 1 in 5,545 | <1 in 10 million |
| | | | Finnish Population | 1 in 100 | 91% | 1 in 1,101 | 1 in 440,400 |
| 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 |
| SLC26A2 | Diastrophic dysplasia | AR | General Population | 1 in 158 | 90% | 1 in 1,571 | 1 in 992,872 |
| | | | Finnish Population | 1 in 50 | 90% | 1 in 491 | 1 in 98,200 |
| SLC26A2 | Achondrogenesis, type IB | AR | General Population | 1 in 158 | 90% | 1 in 1,571 | 1 in 992,872 |
| | | | Finnish Population | 1 in 50 | 90% | 1 in 491 | 1 in 98,200 |
| SLC26A2 | Multiple epiphyseal dysplasia | AR | General Population | 1 in 158 | 90% | 1 in 1,571 | 1 in 992,872 |
| | | | Finnish Population | 1 in 50 | 90% | 1 in 491 | 1 in 98,200 |
| SLC26A2 | Atelosteogenesis II | AR | General Population | 1 in 158 | 90% | 1 in 1,571 | 1 in 992,872 |
| | | | Finnish Population | 1 in 50 | 90% | 1 in 491 | 1 in 98,200 |
| SLC26A4 | Pendred syndrome | AR | General Population | 1 in 80 | 98% | 1 in 3,951 | 1 in 1,264,320 |
| | | | African/African American Population | 1 in 76 | 98% | 1 in 3,751 | 1 in 1,140,304 |
| | | | Caucasian / European Population | 1 in 88 | 98% | 1 in 4,351 | 1 in 1,531,552 |
| | | | East Asian Population | 1 in 74 | 98% | 1 in 3,651 | 1 in 1,080,696 |
| 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 |
| 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 |
| | | | 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 |
| STAR | Lipoid congenital adrenal hyperplasia | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <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 1 | AR | General Population | 1 in 250 | 98% | 1 in 12,451 | <1 in 10 million |
| TGM1 | Congenital ichthyosis | AR | General Population | 1 in 224 | 95% | 1 in 4,461 | 1 in 3,997,056 |
| TH | Segawa syndrome | AR | General Population | 1 in 224 | 98% | 1 in 11,151 | 1 in 9,991,296 |
| TMEM216 | Joubert syndrome 2 | AR | General Population | 1 in 141 | 98% | 1 in 7,001 | 1 in 3,948,564 |
| | | | Ashkenazi Jewish Population | 1 in 92 | 98% | 1 in 4,551 | 1 in 1,674,768 |
| TMEM216 | Meckel syndrome 2 | AR | General Population | 1 in 141 | 98% | 1 in 7,001 | 1 in 3,948,564 |
| | | | Ashkenazi Jewish Population | 1 in 92 | 98% | 1 in 4,551 | 1 in 1,674,768 |
| 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 |
| TPPA | 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 |

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 DOB: [REDACTED] MR#: BFA 0184

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| Supplemental Table | | | | | | | |
|--------------------|---|-------------|---------------------------------|--------------|----------------|--------------------------------|------------------|
| Gene | Condition | Inheritance | Ethnicity | Carrier Rate | Detection Rate | Post-test Carrier Probability* | Residual Risk* |
| USH1C | Usher syndrome, type 1C | 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 |
| USH1C | Non-syndromic hearing loss, USH1C-related | AR | General Population | 1 in 353 | 90% | 1 in 3,521 | 1 in 4,971,652 |
| | | | French Canadian Population | 1 in 227 | 90% | 1 in 2,261 | 1 in 2,052,988 |
| 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 |
| VPS13B | Cohen syndrome | AR | General Population | <1 in 500 | 98% | 1 in 24,951 | <1 in 10 million |
| 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 ($\alpha\alpha/\alpha-$) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis ($\alpha\alpha/-$) is 1 in 1000.

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