Expanded Carrier Screen by Next Generation Sequencing with Fragile X

Patient: [redacted]
DOB: Age: 27 Sex: F
Patient Identifiers: [redacted]
Visit Number (FIN): [redacted]

Client: [redacted]
Physician: [redacted]

ARUP Test Code: 2014677

Collection Date: 12/08/2023
Received in lab: 12/11/2023
Completion Date: 12/21/2023

Test Information

Test performed at: Myriad Women's Health, Inc., 180 Kimball Way, South San Francisco, CA 94080

Patient Report

Patient's genetic report from Myriad Women's Health, Inc. continues on following page(s).

ARUP LABORATORIES | 800-522-2787 | aruplab.com
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Jonathan R. Genzen, MD, PhD, Laboratory Director

Chart continues on following page(s)

myriad WOMEN'S HEALTH

RESULTS RECIPIENT
ARUP LABORATORIES
Attn: [redacted]
500 S Chipeta Way
Salt Lake City, UT 84108
Phone: (801) 583-2787
Fax: (801) 584-5249
NPI: [redacted]
Report Date: 12/21/2023

FEMALE
[redacted]
DOB:
Ethnicity: Hispanic
Sample Type: EDTA Blood
Date of Collection: 12/08/2023
Date Received: 12/12/2023
Date Tested: 12/20/2023
Barcode: [redacted]
Accession ID: 23-342-136370
Indication: Screening for genetic disease carrier status

MALE
N/A

Foresight® Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

RESULTS SUMMARY

Risk Details	[redacted]	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel Fragile X Syndrome **(176 conditions tested)**	N/A
POSITIVE: CARRIER GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness **Reproductive Risk:** 1 in 130 **Inheritance:** Autosomal Recessive	**CARRIER*** NM_004004.5(GJB2):c.139G>T (E47*) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".
POSITIVE: CARRIER Alpha Thalassemia, HBA1/HBA2-related **Reproductive Risk:** Not Calculated **Inheritance:** Autosomal Recessive	**CARRIER*** 1 disease-causing mutation detected. Alpha globin status: -a/aa.	Reproductive risk can be more accurately assessed after carrier screening of the partner. Carrier testing should be considered. See "Next Steps".

*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 10.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

DISCLAIMERS

- The terms 'male', 'female', 'he', 'she', 'women', and 'men' refer to sex assigned at birth.

POSITIVE: CARRIER

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness

Reproductive risk: 1 in 130
Risk before testing: 1 in 4,100

Gene: GJB2 | **Inheritance Pattern:** Autosomal Recessive

Patient	[redacted]	**No partner tested**
Result	Carrier	N/A
Variant(s)	NM_004004.5(GJB2):c.139G>T(E47*) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v4.0)	N/A
Interpretation	This individual is a carrier of GJB2-related DFNB1 nonsyndromic hearing loss and deafness. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_004004:1-2.	N/A

What Is GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness?

DFNB1 nonsyndromic hearing loss and deafness is an inherited condition in which an individual has mild to severe hearing loss, usually, from birth. It is caused by mutations in *GJB2* (which encodes the protein connexin 26) and *GJB6* (which encodes connexin 30). The condition does not typically worsen over time, but in some cases may be slowly progressive. The word "nonsyndromic" refers to the fact that there are no other symptoms or systems of the body involved with the disease. Unlike some other forms of hearing loss, DFNB1 nonsyndromic hearing loss and deafness does not affect balance or movement. The degree of hearing loss is difficult to predict based on which genetic mutation one has. Even if members of the same family are affected by DFNB1 nonsyndromic hearing loss and deafness, the degree of hearing loss may vary among them.

How Common Is GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness?

In the United States, the United Kingdom, France, Australia, and New Zealand, approximately 14 in 100,000 individuals have DFNB1 nonsyndromic hearing loss and deafness. This may be an underestimate as individuals with a mild presentation may not be diagnosed. Roughly 1 in 33 Caucasian individuals are carriers a the mutation that causes the condition.

While this condition is most recognized in the Caucasian population, it has also been observed in other ethnicities.

How Is GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness Treated?

Individuals with DFNB1 nonsyndromic hearing loss and deafness may show improvement by using hearing aids. For those with profound deafness, cochlear implants may also be helpful. They may also want to consider enrolling in an educational program for the hearing impaired.

What is the Prognosis for an Individual with GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness?

While an individual with GJB2-related DFNB1 nonsyndromic hearing loss and deafness will have mild to severe hearing loss, it does not affect lifespan and does not affect any other part of the body.

myriad WOMEN'S HEALTH

RESULTS RECIPIENT
ARUP LABORATORIES
Attn: [redacted]
NPI: [redacted]
Report Date: 12/21/2023

FEMALE
[redacted]
DOB:
Ethnicity: Hispanic
Barcode: [redacted]

MALE
N/A

POSITIVE: CARRIER

Alpha Thalassemia, HBA1/HBA2-related

Genes: HBA1, HBA2 | **Inheritance Pattern:** Autosomal Recessive

Patient	[redacted]	**No partner tested**
Result	✚ Carrier	N/A
Variant(s)	-alpha3.7 [chr16:g.(?_226678)_(227520_?)del] heterozygote	N/A
Alpha globin status	-a/aa	N/A
Methodology	Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (v4.0)	N/A
Interpretation	This individual is a carrier of alpha thalassemia. Carriers do not experience symptoms, but may have hematologic abnormalities. -alpha3.7 is a pathogenic deletional alpha thalassemia variant. Based on this result, the patient's alpha globin status is -a/aa (alpha+ carrier), where "-" indicates a deleted or nonfunctional alpha globin gene.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000517:1-3; NM_000558:1-3. ##	N/A

In addition to the exons sequenced, the following targeted variants were also tested: -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40.

REPRODUCTIVE RISK SUMMARY

Reproductive risk can be more accurately assessed after carrier screening of the partner. Genetic counseling is recommended to review results and risks in further detail.

What is Alpha Thalassemia, HBA1/HBA2-related?

Alpha thalassemia is an inherited blood disorder that affects hemoglobin. Hemoglobin is a protein found in red blood cells (RBCs) that makes it possible for RBCs to bind and carry oxygen throughout the body. Hemoglobin is made up of two different protein chains, which are referred to as alpha and beta chains (or alpha and beta globin). Alpha thalassemia is caused by harmful genetic changes (variants) in the *HBA1* and *HBA2* genes. These genes work together to make the alpha globin protein.

Most individuals inherit two normal copies of the *HBA1* gene (one from each parent) and two normal copies of the *HBA2* gene. This means that everyone has four gene copies that make up the alpha chain of their hemoglobin (two *HBA1* and two *HBA2*). Individuals can inherit a harmful change in one, two, three, or all four gene copies. There are also different types of changes within the *HBA1* and *HBA2* genes. Larger changes that remove most or all of the gene are called "deletional," while smaller changes are called "non-deletional."




The symptoms associated with alpha thalassemia can range from a reduced number of RBCs (anemia) to fetal death. The different forms of alpha thalassemia are described below. Because there are several forms of alpha thalassemia and the risk for disease depends on a variety of factors, individuals with variants in *HBA1* and *HBA2* should consult a genetics professional to determine both their personal risk for disease and their reproductive risk.

SILENT CARRIER

Silent carriers of alpha thalassemia have a change in just one of the four alpha globin genes. Individuals with this finding are known as silent carriers because they typically do not have any disease symptoms or visible abnormalities in their RBCs.

ALPHA THALASSEMIA-TRAIT (CARRIER)

Carriers of alpha thalassemia have a change in two of the four alpha globin genes. Individuals with this finding generally have RBCs that are pale (hypochromic) and small (microcytic) when visualized. They may also have a mild decrease in the amount of RBCs (mild anemia). Symptoms of anemia can include tiredness, shortness of breath, lightheadedness, or dizziness. Individuals with only two functional alpha globin genes normally do not require treatment, as they generally do not exhibit symptoms of disease. However, there are reports of individuals with two non-deletional changes who have a diagnosis of a more severe form of the disease called hemoglobin H (HbH) (see below). One example of this is when individuals have two copies of the hemoglobin Constant Spring variant, which is common in the Southeast Asian population.

HEMOGLOBIN H DISEASE

HbH disease is typically the result of changes in three of the four alpha globin genes. This form is highly variable, and symptoms depend on the type of changes present in an individual. Some individuals with HbH do not have any symptoms, while some may have mild to moderate anemia. Other symptoms of HbH can include yellowing of the skin or eyes (jaundice), enlargement of the spleen, and other rarer complications. Although the severity of symptoms can vary, individuals with HbH disease are still considered affected with alpha thalassemia and treatment is often required.

HEMOGLOBIN BART SYNDROME

Hemoglobin Bart (Hb Bart) syndrome is typically the result of changes in all four of the alpha globin genes. Hb Bart is generally associated with fetal death due to the buildup of excess fluid in the body and tissues (hydrops fetalis). Most babies with this condition are stillborn or die soon after birth. Signs and symptoms in the newborn period can include severe anemia, enlargement of the liver and spleen, and birth defects of the heart, urinary system, and genitals. When fetal blood transfusions are successful, survival is possible; however, there is a high risk for intellectual and physical disability in these survivors.

DELETIONAL VS. NON-DELETIONAL VARIANTS

Historically, the predicted severity of alpha thalassemia was based on how many *HBA1* and *HBA2* genes were impacted. In general, individuals with changes in more of their alpha globin genes typically have more severe symptoms (i.e. variants in three or four genes result in more clinical features than variants in only one or two genes). However, research has shown that both the number *and* the type of changes determine the severity of an individual's symptoms. Larger changes that remove most or all of the gene are called "deletional," while smaller changes are called "non-deletional." Some non-deletional changes are associated with a higher risk for severe symptoms than deletional changes. Thus, the severity of an individual's condition can vary based on the combination of deletional and non-deletional changes they have. Given the many different factors that can influence an individual's personal and reproductive risk for alpha thalassemia, a consult with a genetics professional may be recommended.

How common is Alpha Thalassemia, HBA1/HBA2-related?

The incidence of alpha thalassemia in the population is approximately 1 in 10,000 births. However, the incidence of Hb Bart and HbH is much higher among individuals of Southeast Asian, Mediterranean, and Middle Eastern descent. Southeast Asia has the highest documented incidence, with estimates around 1 in 400 affected births.

How is Alpha Thalassemia, HBA1/HBA2-related treated?

Treatment for HbH disease varies based on the severity of the symptoms. Many individuals will need a blood transfusion during times of severe illness (crises). This is usually a rare occurrence, and it can be caused by environmental stressors such as fever or exposure to specific medications. Individuals with more severe symptoms may require regular blood transfusions, folic acid supplementation, antibiotics during certain procedures, removal of excess iron from the body (iron chelation therapy), removal of the spleen, and possibly therapies to increase fetal hemoglobin levels.

Rare cases of survivors with Hb Bart syndrome have been reported when fetal blood transfusions were given, followed by regular treatments similar to those given to individuals with HbH disease. Treatment or surgical correction of birth defects may also be possible. There is a high risk for intellectual and physical disability in these survivors. These individuals may be candidates for hematopoietic stem cell transplantation.

What is the prognosis for an individual with Alpha Thalassemia, HBA1/HBA2-related?

The long-term outcome of HbH ultimately depends on the severity of the disease. Mild disease may be manageable with little effect on daily life. However, more severe disease will require frequent and regular therapy and may be associated with a shortened lifespan. When treated, individuals with HbH disease can have a near-normal lifespan.

Hb Bart syndrome is the most severe clinical condition related to alpha thalassemia, and death may occur during pregnancy (*in utero*) or in the newborn period. There may also be maternal complications during pregnancy if the fetus has Hb Bart syndrome. These complications include high blood pressure with fluid buildup and protein in the urine (preeclampsia); excessive amniotic fluid (polyhydramnios) or reduced amniotic fluid (oligohydramnios); hemorrhage; and premature delivery. When fetal blood transfusions are successful, survival is possible. However, there is a high risk for intellectual and physical disability in survivors.

Methods and Limitations

[redacted] **[Foresight Carrier Screen]**: Sequencing with copy number analysis, triplet repeat detection, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): fragile x, DTS v4.0).

Sequencing with copy number analysis

Hybridization capture-based target enrichment, high-throughput sequencing, and read-depth-based copy number analysis are used to analyze the genes listed in the Conditions Tested section of the report. Except where otherwise noted, the region of interest (ROI) comprises the indicated coding regions and 20 non-coding bases flanking each region. In rare instances where genomic features (e.g., homopolymers) or other variables compromise calling fidelity, the affected regions are excluded from the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. Select genes or regions for which pseudogenes or other types of homology impede reliable variant detection may be assayed using alternate technology or may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with a sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel with a sensitivity of ≥75%. Selected founder deletions may be detected at higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from tiled regions and, when available, using junction reads. Only exons included in the region affected by the copy number variant (CNV) are included in the variant nomenclature. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed.

Triplet repeat detection

Polymerase chain reaction (PCR) with fluorescently labeled primers is used to amplify the CGG repeat region in the 5' UTR of *FMR1* (NM_002024.4: c.1-131CGG[1_n]), and PCR products are sized using capillary electrophoresis. Reported sizes are accurate to ±1 repeat for normal or intermediate alleles and ± two repeats for premutation alleles. Alleles above 200 CGG repeats (full mutations), while identified, are not specifically sized and will be reported as ">200" CGG repeats. In an unknown number of cases, other genetic variation may interfere with CGG repeat analysis. Other *FMR1* pathogenic variation will not be detected. *FMR1* promoter methylation is not analyzed. Allele size mosaicism may not be detected, as the test has been calibrated to yield results that are equivalent to the results from Southern blot. Opt-in testing of *FMR1* AGG interruptions is available for results showing between 50 and 54 CGG repeats. Automatic reflex testing of AGG interruptions is performed for results showing between 55 and 90 CGG repeats. AGG interruption analysis is performed by a reference laboratory, and methods are provided in the appended report, when applicable. This assay is designed to detect germline (constitutional) variation of the CGG repeat in the 5' UTR of *FMR1*; gonadal mosaicism will not be detected. Results assume a normal karyotype. Sex chromosome variations and aneuploidies may affect the accuracy of this assay.

Spinal muscular atrophy

Targeted copy number analysis via hybridization capture-based target enrichment and high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. In an unknown number of cases, other genetic variation may interfere with this analysis. Some individuals with two copies of *SMN1* are "silent" carriers with both *SMN1* genes on one chromosome and no copies of the gene on the other chromosome. This is more likely in individuals who have two copies of the *SMN1* gene and are positive for the g.27134T>G single-nucleotide polymorphism (SNP) (PMID: 9199562, 23788250, and 28676062), which affects the reported residual risk. Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have two apparent copies of *SMN1*. Further, individuals who are negative for the g.27134T>G SNP, and who are reported as having two copies of the *SMN1* gene may have additional *SMN1* gene copies. If additional unreported *SMN1* copies are present, the reported residual risk for these individuals may be overestimated. Other rare carrier states, where complex exchanges exist between gene copies or chromosomes, may not be detected by the assay.

Analysis of homologous regions

Hybridization capture-based target enrichment, high-throughput sequencing, targeted genotyping, and read-depth-based copy number analysis are used to determine the number of functional gene copies and/or the presence of selected variants in genes that have significant homology to other genomic regions. The precise breakpoints of large deletions in these genes cannot be determined but are instead estimated from copy number analysis. Pseudogenes may interfere with this analysis, especially when many pseudogene copies are present.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a pathogenic variant may or may not be a carrier of 21-hydroxylase deficient congenital adrenal hyperplasia (CAH), depending on the chromosomal location of the variants (i.e., phase). Benign *CYP21A2* gene duplications and/or triplications will only be reported in this context. Some individuals with two functional *CYP21A2* gene copies may be "silent" carriers, with two gene copies resulting from a duplication on one chromosome and a gene deletion on the other chromosome. This and other rare carrier states, where complementary changes exist between gene copies or chromosomes, may not be detected by the assay. Given that the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk estimates on the report are based on the published incidence for classic CAH. However, the published prevalence of non-classic CAH is highest in individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent. Therefore, the residual and reproductive risks are likely an underestimate for CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis

Hybridization capture-based target enrichment, high-throughput sequencing, and copy number analysis are used to identify sequence variation and functional gene copies within the region of interest (ROI) of *HBA1* and *HBA2*, which includes the exons listed in the assay specifications plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g., homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. For large deletions or duplications in these genes, the precise breakpoints cannot be determined but are instead estimated from copy number analysis. This assay has been validated to detect up to two additional copies of each alpha globin gene. In rare instances where assay results suggest greater than two additional copies are present, this will be noted but the specific number of gene copies observed will not be provided. Extensive sequence homology exists between *HBA1* and *HBA2*. This sequence homology can prevent certain variants from being localized to one gene over the other. In these instances, variant nomenclature will be provided for both genes. If follow-up testing is indicated for patients with the nomenclature provided for both genes, both *HBA1* and *HBA2* should be tested. Some individuals with four functional alpha globin gene copies may be "silent" carriers, with three gene copies resulting from triplication on one chromosome and a single gene deletion on the other chromosome. This and other rare carrier states, where complementary changes exist between gene copies or chromosomes, may not be detected by the assay.

Interpretation of reported variants

The interpretation and classification of variants reflect the current state of Myriad's scientific understanding based on information available at the time of variant assessment. Variants are classified according to internally defined criteria, which are compatible with the ACMG Standards and Guidelines for the Interpretation of Sequence Variants (PMID: 25741868). Variants that have been determined by Myriad to be disease-causing or likely disease-causing (i.e., pathogenic or likely pathogenic) are reported. Benign variants, likely benign variants, variants of uncertain clinical significance (VUS), and variants not directly associated with the specified disease phenotype(s) are not reported. Variant classification and interpretation may change over time for a variety of reasons, including but not limited to, improvements to classification techniques, availability of additional scientific information, and observation of a variant in additional individuals. If the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If a report is updated or re-issued for other reasons, the variants reported may change based on their classification at the time of re-issue. This can include changes to the variants displayed in gene-specific 'variants tested' sections. Healthcare providers may contact Myriad directly to request updated variant classification information specific to this test result.

Limitations

The Foresight® Carrier Screen is designed to detect and report germline (constitutional) alterations. Mosaic (somatic) variation may not be detected, and if it is detected, it may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants reside on the same chromosome or different chromosomes (i.e., phase). This assay is not validated to detect sex chromosome copy number variations; however, limited sex chromosome analysis is performed for quality control purposes. If present, sex chromosome variations including copy number variations, aneuploidies, aneusomies, rearrangements, or other structural changes may significantly reduce test sensitivity and accuracy of risk estimates. Variant interpretation and residual and reproductive risk estimates assume a normal karyotype. The Foresight™ Carrier Screen reports carrier status for only genes/phenotypes specified by the ordering healthcare provider. Other heritable and non-heritable conditions and defects exist that are not addressed by this test. Furthermore, not all forms of genetic variation are detected by this assay (e.g., duplications [except in specified genes], chromosomal rearrangements, structural abnormalities, etc.). Additional testing may be appropriate for some individuals. Pseudogenes and other regions of homology may interfere with this analysis. In an unknown number of cases, other genetic variation may interfere with variant detection. Rare carrier states where complementary changes exist between genes or chromosomes may not be detected by the assay. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions, and technical or analytical errors.

Detection rates are determined using published scientific literature and/or reputable databases, when available, to estimate the fraction of disease alleles, weighted by frequency, that the methodology is predicted to be able or unable to detect. Detection rates are approximate and only account for analytical sensitivity. Certain variants that have been previously described in the literature may not be reported if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease-specific rates of *de novo* variation.

This test was developed, and its performance characteristics determined, by Myriad Women's Health, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician's evaluation.

Incidental Findings

Unless otherwise indicated, these results and interpretations are limited to the specific disease panel(s) requested by the ordering healthcare provider. In some cases, standard data analyses may identify genetic findings beyond the region(s) of interest specified by the test, and such findings may not be reported. These findings may include genomic abnormalities with major, minor, or no clinical significance.

If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

Resources

GENOME CONNECT | http://www.genomeconnect.org

Patients can share their reports using research registries such as Genome Connect, an online research registry building a genetics and health knowledge base. Genome Connect provides patients, physicians, and researchers an opportunity to share genetic information to support the study of the impact of genetic variation on health conditions.

SENIOR LABORATORY DIRECTOR

Karla R. Bowles

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Matthew Meredith, PhD, FACMG, CGMBS on Dec 21, 2023

Genetic testing was completed by CLIA accredited laboratories in the United States located at: 180 Kimball Way, South San Francisco, CA 94080. CLIA IDs: 05D1102604.




ARUP LABORATORIES | 800-522-2787 | aruplab.com
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Jonathan R. Genzen, MD, PhD, Laboratory Director

Chart continues on following page(s)

Conditions Tested

6-pyruvoyl-tetrahydropterin Synthase Deficiency - **Gene:** PTS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000317:1-6. **Detection Rate:** Hispanic >99%.

Adenosine Deaminase Deficiency - **Gene:** ADA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000022:1-12. **Detection Rate:** Hispanic 98%.

Alpha Thalassemia, HBA1/HBA2-related - **Genes:** HBA1, HBA2. Autosomal Recessive. Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis. **Exons:** NM_000517:1-3; NM_000558:1-3. **Variants (16):** -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40. **Detection Rate:** Hispanic >99%.

Alpha-mannosidosis - **Gene:** MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000528:1-23. **Detection Rate:** Hispanic >99%.

Alpha-sarcoglycanopathy - **Gene:** SGCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000023:1-9. **Detection Rate:** Hispanic >99%.

Alport Syndrome, COL4A3-related - **Gene:** COL4A3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000091:1-52. **Detection Rate:** Hispanic 94%.

Alport Syndrome, COL4A4-related - **Gene:** COL4A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000092:2-48. **Detection Rate:** Hispanic >99%.

Alstrom Syndrome - **Gene:** ALMS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015120:1-23. **Detection Rate:** Hispanic >99%.

Andermann Syndrome - **Gene:** SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133647:1-25. **Detection Rate:** Hispanic >99%.

Argininemia - **Gene:** ARG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000045:1-8. **Detection Rate:** Hispanic 97%.

Argininosuccinic Aciduria - **Gene:** ASL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001024943:1-16. **Detection Rate:** Hispanic >99%.

Aspartylglucosaminuria - **Gene:** AGA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000027:1-9. **Detection Rate:** Hispanic >99%.

Ataxia with Vitamin E Deficiency - **Gene:** TTPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000370:1-5. **Detection Rate:** Hispanic >99%.

Ataxia-telangiectasia - **Gene:** ATM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000051:2-63. **Detection Rate:** Hispanic 96%.

ATP7A-related Disorders - **Gene:** ATP7A. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000052:2-23. **Detection Rate:** Hispanic 90%.

Autoimmune Polyglandular Syndrome Type 1 - **Gene:** AIRE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000383:1-14. **Detection Rate:** Hispanic >99%.

Autosomal Recessive Osteopetrosis Type 1 - **Gene:** TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006019:2-20. **Detection Rate:** Hispanic 96%.

Autosomal Recessive Polycystic Kidney Disease, PKHD1-related - **Gene:** PKHD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138694:2-67. **Detection Rate:** Hispanic >99%.

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - **Gene:** SACS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014363:2-10. **Detection Rate:** Hispanic 99%.

Bardet-Biedl Syndrome, BBS1-related - **Gene:** BBS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024649:1-17. **Detection Rate:** Hispanic >99%.

Bardet-Biedl Syndrome, BBS10-related - **Gene:** BBS10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024685:1-2. **Detection Rate:** Hispanic >99%.

Bardet-Biedl Syndrome, BBS12-related - **Gene:** BBS12. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_152618:2. **Detection Rate:** Hispanic >99%.

Bardet-Biedl Syndrome, BBS2-related - **Gene:** BBS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_031885:1-17. **Detection Rate:** Hispanic >99%.

BCS1L-related Disorders - **Gene:** BCS1L. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004328:3-9. **Detection Rate:** Hispanic >99%.

Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - **Gene:** HBB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000518:1-3. **Detection Rate:** Hispanic >99%.

Beta-sarcoglycanopathy - **Gene:** SGCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000232:1-6. **Detection Rate:** Hispanic >99%.

Biotinidase Deficiency - **Gene:** BTD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000060:1-4. **Detection Rate:** Hispanic >99%.

Bloom Syndrome - **Gene:** BLM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000057:2-22. **Detection Rate:** Hispanic >99%.

Calpainopathy - **Gene:** CAPN3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000070:1-24. **Detection Rate:** Hispanic 99%.

Canavan Disease - **Gene:** ASPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000049:1-6. **Detection Rate:** Hispanic 98%.

Carbamoylphosphate Synthetase I Deficiency - **Gene:** CPS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001875:1-38. **Detection Rate:** Hispanic >99%.

Carnitine Palmitoyltransferase IA Deficiency - **Gene:** CPT1A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001876:2-19. **Detection Rate:** Hispanic >99%.

Carnitine Palmitoyltransferase II Deficiency - **Gene:** CPT2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000098:1-5. **Detection Rate:** Hispanic >99%.

Cartilage-hair Hypoplasia - **Gene:** RMRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NR_003051:1. **Detection Rate:** Hispanic >99%.

Cerebrotendinous Xanthomatosis - **Gene:** CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000784:1-9. **Detection Rate:** Hispanic >99%.

Citrullinemia Type 1 - **Gene:** ASS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000050:3-16. **Detection Rate:** Hispanic >99%.

CLN3-related Disorders - **Gene:** CLN3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001042432:2-16. **Detection Rate:** Hispanic >99%.

CLN5-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006493:1-4. **Detection Rate:** Hispanic >99%.

CLN8-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_018941:2-3. **Detection Rate:** Hispanic >99%.

Cohen Syndrome - **Gene:** VPS13B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017890:2-62. **Detection Rate:** Hispanic 97%.

Combined Pituitary Hormone Deficiency, PROP1-related - **Gene:** PROP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006261:1-3. **Detection Rate:** Hispanic >99%.

Congenital Adrenal Hyperplasia, CYP11B1-related - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** Hispanic 97%.

Congenital Adrenal Hyperplasia, CYP21A2-related - **Gene:** CYP21A2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Hispanic 95%.

Congenital Disorder of Glycosylation Type Ic - **Gene:** ALG6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_013339:2-15. **Detection Rate:** Hispanic >99%.

Chart continues on following page(s)

Congenital Disorder of Glycosylation, MPI-related - Gene: MPI. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002435:1-8. Detection Rate: Hispanic >99%.
Congenital Disorder of Glycosylation, PMM2-related - Gene: PMM2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000303:1-8. Detection Rate: Hispanic >99%.
Costeff Optic Atrophy Syndrome - Gene: OPA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_025136:1-2. Detection Rate: Hispanic >99%.
Cystic Fibrosis - Gene: CFTR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. Detection Rate: Hispanic >99%.
Cystinosis - Gene: CTNS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004937:3-12. Detection Rate: Hispanic >99%.
D-bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000414:1-24. Detection Rate: Hispanic 98%.
Delta-sarcoglycanopathy - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000337:2-9. Detection Rate: Hispanic 96%.
Dihydrolipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000108:1-14. Detection Rate: Hispanic >99%.
Dysferlinopathy - Gene: DYSF. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003494:1-55. Detection Rate: Hispanic 98%.
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - Gene: DMD. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_004006:1-79. Detection Rate: Hispanic 99%.
ERCC6-related Disorders - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000124:2-21. Detection Rate: Hispanic 96%.
ERCC8-related Disorders - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000082:1-12. Detection Rate: Hispanic 97%.
EVC-related Ellis-van Creveld Syndrome - Gene: EVC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_153717:1-21. Detection Rate: Hispanic 96%.
EVC2-related Ellis-van Creveld Syndrome - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_147127:1-22. Detection Rate: Hispanic 98%.
Fabry Disease - Gene: GLA. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000169:1-7. Detection Rate: Hispanic 98%.
Familial Dysautonomia - Gene: ELP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003640:2-37. Detection Rate: Hispanic >99%.
Familial Hyperinsulinism, ABCC8-related - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000352:1-39. Detection Rate: Hispanic >99%.
Familial Hyperinsulinism, KCNJ11-related - Gene: KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_000525:1. Detection Rate: Hispanic >99%.
Familial Mediterranean Fever - Gene: MEFV. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000243:1-10. Detection Rate: Hispanic >99%.
Fanconi Anemia Complementation Group A - Gene: FANCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000135:1-43. Detection Rate: Hispanic 92%.
Fanconi Anemia, FANCC-related - Gene: FANCC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000136:2-15. Detection Rate: Hispanic >99%.
FKRP-related Disorders - Gene: FKRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_024301:4. Detection Rate: Hispanic >99%.
FKTN-related Disorders - Gene: FKTN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001079802:3-11. Detection Rate: Hispanic >99%.
Fragile X Syndrome - Gene: FMR1. X-linked Dominant. Triplet repeat detection. Variant (1): FMR1 CGG repeat number. Detection Rate: Hispanic >99%.
Free Sialic Acid Storage Disorders - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012434:1-11. Detection Rate: Hispanic 98%.
Galactokinase Deficiency - Gene: GALK1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000154:1-8. Detection Rate: Hispanic >99%.
Galactosemia - Gene: GALT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000155:1-11. Detection Rate: Hispanic >99%.
Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. Detection Rate: Hispanic 87%.
Gaucher Disease - Gene: GBA1. Autosomal Recessive. Analysis of homologous regions. Variants (10): D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. Detection Rate: Hispanic 60%.
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - Gene: GJB2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004004:1-2. Detection Rate: Hispanic >99%.
GLB1-related Disorders - Gene: GLB1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000404:1-16. Detection Rate: Hispanic >99%.
Glutaric Acidemia, GCDH-related - Gene: GCDH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000159:2-12. Detection Rate: Hispanic >99%.
Glycine Encephalopathy, AMT-related - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000481:1-9. Detection Rate: Hispanic >99%.
Glycine Encephalopathy, GLDC-related - Gene: GLDC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000170:1-25. Detection Rate: Hispanic 94%.
Glycogen Storage Disease Type Ia - Gene: G6PC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000151:1-5. Detection Rate: Hispanic 98%.
Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001164277:3-11. Detection Rate: Hispanic >99%.
Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000642:2-34. Detection Rate: Hispanic >99%.
GNE Myopathy - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001128227:1-12. Detection Rate: Hispanic >99%.
GNPTAB-related Disorders - Gene: GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024312:1-21. Detection Rate: Hispanic >99%.
HADHA-related Disorders - Gene: HADHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000182:1-20. Detection Rate: Hispanic >99%.
Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000035:2-9. Detection Rate: Hispanic >99%.
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000520:1-14. Detection Rate: Hispanic >99%.
HMG-CoA Lyase Deficiency - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000191:1-9. Detection Rate: Hispanic >99%.
Holocarboxylase Synthetase Deficiency - Gene: HLCS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000411:4-12. Detection Rate: Hispanic >99%.
Homocystinuria, CBS-related - Gene: CBS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000071:3-17. Detection Rate: Hispanic >99%.
Hydrolethalus Syndrome - Gene: HYLS1. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_145014:4. Detection Rate: Hispanic >99%.
Hypophosphatasia - Gene: ALPL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000478:2-12. Detection Rate: Hispanic >99%.
Isovaleric Acidemia - Gene: IVD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002225:1-12. Detection Rate: Hispanic >99%.
Joubert Syndrome 2 - Gene: TMEM216. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001173990:1-5. Detection Rate: Hispanic >99%.

Junctional Epidermolysis Bullosa, LAMA3-related - **Gene:** LAMA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000227:1-38. **Detection Rate:** Hispanic >99%.
Junctional Epidermolysis Bullosa, LAMB3-related - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228:2-23. **Detection Rate:** Hispanic >99%.
Junctional Epidermolysis Bullosa, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** Hispanic >99%.
Krabbe Disease - **Gene:** GALC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000153:1-17. **Detection Rate:** Hispanic >99%.
Leigh Syndrome, French-Canadian Type - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133259:1-38. **Detection Rate:** Hispanic >99%.
Lipoid Congenital Adrenal Hyperplasia - **Gene:** STAR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000349:1-7. **Detection Rate:** Hispanic >99%.
Lysosomal Acid Lipase Deficiency - **Gene:** LIPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000235:2-10. **Detection Rate:** Hispanic 98%.
Maple Syrup Urine Disease Type Ia - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000709:1-9. **Detection Rate:** Hispanic >99%.
Maple Syrup Urine Disease Type Ib - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_183050:1-10. **Detection Rate:** Hispanic >99%.
Maple Syrup Urine Disease Type II - **Gene:** DBT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001918:1-11. **Detection Rate:** Hispanic 97%.
Medium-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000016:1-12. **Detection Rate:** Hispanic >99%.
Megalencephalic Leukoencephalopathy with Subcortical Cysts - **Gene:** MLC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015166:2-12. **Detection Rate:** Hispanic >99%.
Metachromatic Leukodystrophy - **Gene:** ARSA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000487:1-8. **Detection Rate:** Hispanic >99%.
Methylmalonic Acidemia, cblA Type - **Gene:** MMAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_172250:2-7. **Detection Rate:** Hispanic >99%.
Methylmalonic Acidemia, cblB Type - **Gene:** MMAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_052845:1-9. **Detection Rate:** Hispanic >99%.
Methylmalonic Acidemia, MMUT-related - **Gene:** MMUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** Hispanic >99%.
Methylmalonic Aciduria and Homocystinuria, cblC Type - **Gene:** MMACHC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015506:1-4. **Detection Rate:** Hispanic >99%.
MKS1-related Disorders - **Gene:** MKS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017777:1-18. **Detection Rate:** Hispanic >99%.
Mucolipidosis III Gamma - **Gene:** GNPTG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032520:1-11. **Detection Rate:** Hispanic 98%.
Mucolipidosis IV - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_020533:1-14. **Detection Rate:** Hispanic >99%.
Mucopolysaccharidosis Type I - **Gene:** IDUA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000203:1-14. **Detection Rate:** Hispanic >99%.
Mucopolysaccharidosis Type II - **Gene:** IDS. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000202:1-9. **Detection Rate:** Hispanic 89%.
Mucopolysaccharidosis Type IIIA - **Gene:** SGSH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000199:1-8. **Detection Rate:** Hispanic >99%.
Mucopolysaccharidosis Type IIIB - **Gene:** NAGLU. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000263:1-6. **Detection Rate:** Hispanic >99%.
Mucopolysaccharidosis Type IIIC - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_152419:1-18. **Detection Rate:** Hispanic >99%.
Muscular Dystrophy, LAMA2-related - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-43,45-65. **Detection Rate:** Hispanic 98%.
MYO7A-related Disorders - **Gene:** MYO7A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000260:2-49. **Detection Rate:** Hispanic >99%.
NEB-related Nemaline Myopathy - **Gene:** NEB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** Hispanic 92%.
Nephrotic Syndrome, NPHS1-related - **Gene:** NPHS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004646:1-29. **Detection Rate:** Hispanic >99%.
Nephrotic Syndrome, NPHS2-related - **Gene:** NPHS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014625:1-8. **Detection Rate:** Hispanic >99%.
Neuronal Ceroid Lipofuscinosis, CLN6-related - **Gene:** CLN6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017882:1-7. **Detection Rate:** Hispanic >99%.
Niemann-Pick Disease Type C1 - **Gene:** NPC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000271:1-25. **Detection Rate:** Hispanic >99%.
Niemann-Pick Disease Type C2 - **Gene:** NPC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006432:1-5. **Detection Rate:** Hispanic >99%.
Niemann-Pick Disease, SMPD1-related - **Gene:** SMPD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000543:1-6. **Detection Rate:** Hispanic >99%.
Nijmegen Breakage Syndrome - **Gene:** NBN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002485:1-16. **Detection Rate:** Hispanic >99%.
Ornithine Transcarbamylase Deficiency - **Gene:** OTC. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000531:1-10. **Detection Rate:** Hispanic 97%.
PCCA-related Propionic Acidemia - **Gene:** PCCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000282:1-24. **Detection Rate:** Hispanic 95%.
PCCB-related Propionic Acidemia - **Gene:** PCCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000532:1-15. **Detection Rate:** Hispanic >99%.
PCDH15-related Disorders - **Gene:** PCDH15. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_033056:2-33. **Detection Rate:** Hispanic 93%.
Pendred Syndrome - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000441:2-21. **Detection Rate:** Hispanic >99%.
Peroxisome Biogenesis Disorder Type 1 - **Gene:** PEX1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000466:1-24. **Detection Rate:** Hispanic >99%.
Peroxisome Biogenesis Disorder Type 3 - **Gene:** PEX12. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000286:1-3. **Detection Rate:** Hispanic >99%.
Peroxisome Biogenesis Disorder Type 4 - **Gene:** PEX6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000287:1-17. **Detection Rate:** Hispanic 97%.
Peroxisome Biogenesis Disorder Type 5 - **Gene:** PEX2. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000318:4. **Detection Rate:** Hispanic >99%.
Peroxisome Biogenesis Disorder Type 6 - **Gene:** PEX10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153818:1-6. **Detection Rate:** Hispanic >99%.
Phenylalanine Hydroxylase Deficiency - **Gene:** PAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000277:1-13. **Detection Rate:** Hispanic >99%.

POMGNT-related Disorders - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017739:2-22. **Detection Rate:** Hispanic 96%.
Pompe Disease - **Gene:** GAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000152:2-20. **Detection Rate:** Hispanic 98%.
PPT1-related Neuronal Ceroid Lipofuscinosis - **Gene:** PPT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000310:1-9. **Detection Rate:** Hispanic >99%.
Primary Carnitine Deficiency - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003060:1-10. **Detection Rate:** Hispanic >99%.
Primary Hyperoxaluria Type 1 - **Gene:** AGXT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000030:1-11. **Detection Rate:** Hispanic >99%.
Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012203:1-9. **Detection Rate:** Hispanic >99%.
Primary Hyperoxaluria Type 3 - **Gene:** HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** Hispanic >99%.
Pycnodysostosis - **Gene:** CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000396:2-8. **Detection Rate:** Hispanic >99%.
Pyruvate Carboxylase Deficiency - **Gene:** PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000920:3-22. **Detection Rate:** Hispanic >99%.
Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000288:1-10. **Detection Rate:** Hispanic >99%.
RTEL1-related Disorders - **Gene:** RTEL1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032957:2-35. **Detection Rate:** Hispanic >99%.
Sandhoff Disease - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** Hispanic 98%.
Short-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000017:1-10. **Detection Rate:** Hispanic >99%.
Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** Hispanic 96%.
SLC26A2-related Disorders - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000112:2-3. **Detection Rate:** Hispanic >99%.
Smith-Lemli-Opitz Syndrome - **Gene:** DHCR7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001360:3-9. **Detection Rate:** Hispanic >99%.
Spastic Paraplegia Type 15 - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015346:2-42. **Detection Rate:** Hispanic >99%.
Spinal Muscular Atrophy - **Gene:** SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** Hispanic 91%.
Spondylothoracic Dysostosis - **Gene:** MESP2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** Hispanic >99%.
TGM1-related Autosomal Recessive Congenital Ichthyosis - **Gene:** TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000359:2-15. **Detection Rate:** Hispanic >99%.
TPP1-related Neuronal Ceroid Lipofuscinosis - **Gene:** TPP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000391:1-13. **Detection Rate:** Hispanic >99%.
Tyrosine Hydroxylase Deficiency - **Gene:** TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_199292:1-14. **Detection Rate:** Hispanic >99%.
Tyrosinemia Type I - **Gene:** FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** Hispanic >99%.
Tyrosinemia Type II - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** Hispanic >99%.
USH1C-related Disorders - **Gene:** USH1C. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005709:1-21. **Detection Rate:** Hispanic >99%.
USH2A-related Disorders - **Gene:** USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_206933:2-72. **Detection Rate:** Hispanic 98%.
Usher Syndrome Type 3 - **Gene:** CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** Hispanic >99%.
Very-long-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADVL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000018:1-20. **Detection Rate:** Hispanic >99%.
Wilson Disease - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** Hispanic >99%.
X-linked Adrenal Hypoplasia Congenita - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** Hispanic 97%.
X-linked Adrenoleukodystrophy - **Gene:** ABCD1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000033:1-6. **Detection Rate:** Hispanic 77%.
X-linked Alport Syndrome - **Gene:** COL4A5. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000495:1-51. **Detection Rate:** Hispanic 96%.
X-linked Juvenile Retinoschisis - **Gene:** RS1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000330:1-6. **Detection Rate:** Hispanic 98%.
X-linked Myotubular Myopathy - **Gene:** MTM1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000252:2-15. **Detection Rate:** Hispanic 96%.
X-linked Severe Combined Immunodeficiency - **Gene:** IL2RG. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000206:1-8. **Detection Rate:** Hispanic >99%.
Xeroderma Pigmentosum Group A - **Gene:** XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** Hispanic >99%.
Xeroderma Pigmentosum Group C - **Gene:** XPC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004628:1-16. **Detection Rate:** Hispanic 97%.

Risk Calculations

Below are the risk calculations for all conditions tested. Negative results do not rule out the possibility of being a carrier. Residual risk is an estimate of each patient's post-test likelihood of being a carrier, while the reproductive risk represents an estimated likelihood that the patients' future children could inherit each disease. These risks are inherent to all carrier-screening tests, may vary by ethnicity, are predicated on a negative family history, and are present even given a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. In addition, average carrier rates are estimated using incidence or prevalence data from published scientific literature and/or reputable databases, where available, and are incorporated into residual risk calculations for each population/ethnicity. When population-specific data is not available for a condition, average worldwide incidence or prevalence is used. Further, incidence and prevalence data are only collected for the specified phenotypes (which include primarily the classic or severe forms of disease) and may not include alternate or milder disease manifestations associated with the gene. Actual incidence rates, prevalence rates, and carrier rates, and therefore actual residual risks, may be higher or lower than the estimates provided. Carrier rates, incidence/prevalence, and/or residual risks are not provided for some genes with biological or heritable properties that would make these estimates inaccurate. A '†' symbol indicates a positive result. See the full clinical report for interpretation and details. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Disease	[redacted] Residual Risk	Reproductive Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	-alpha3.7 [chr16:g.(?_226678)_(227520_?)del] heterozygote † Alpha globin status: -a/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 34,000	< 1 in 1,000,000
Alport Syndrome, COL4A3-related	1 in 5,800	< 1 in 1,000,000
Alport Syndrome, COL4A4-related	1 in 35,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	1 in 12,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 29,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 4,200	< 1 in 1,000,000
ATP7A-related Disorders	1 in 400,000	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1	1 in 18,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 8,900	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 39,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 2,500	1 in 260,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 17,000	< 1 in 1,000,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 11,000	< 1 in 1,000,000
Canavan Disease	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 18,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 12,000	< 1 in 1,000,000
CLN3-related Disorders	1 in 13,000	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP11B1-related	1 in 8,400	< 1 in 1,000,000

Disease	[redacted] Residual Risk	Reproductive Risk
Congenital Adrenal Hyperplasia, CYP21A2-related	1 in 1,200	1 in 290,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation, MPI-related	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation, PMM2-related	1 in 14,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 5,200	< 1 in 1,000,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	1 in 8,400	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated
ERCC6-related Disorders	1 in 8,400	< 1 in 1,000,000
ERCC8-related Disorders	1 in 12,000	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,800	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	1 in 9,800	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	< 1 in 1,000,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Hyperinsulinism, ABCC8-related	1 in 17,000	< 1 in 1,000,000
Familial Hyperinsulinism, KCNJ11-related	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	1 in 2,800	1 in 330,000
Fanconi Anemia Complementation Group A	1 in 2,900	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 38,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Fragile X Syndrome	Normal: 28 and 31 repeats	Not calculated
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 44,000	< 1 in 1,000,000
Galactosemia	1 in 11,000	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,300	< 1 in 1,000,000
Gaucher Disease	1 in 310	1 in 150,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	NM_004004.5(GJB2):c.139G>T(E47*) heterozygote †	1 in 130
GLB1-related Disorders	1 in 17,000	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycine Encephalopathy, AMT-related	1 in 26,000	< 1 in 1,000,000
Glycine Encephalopathy, GLDC-related	1 in 2,500	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 8,700	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	< 1 in 50,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 25,000	< 1 in 1,000,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 28,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 10,000	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 23,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 26,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMB3-related	1 in 31,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 17,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 11,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 18,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 36,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 12,000	< 1 in 1,000,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency	1 in 6,000	< 1 in 1,000,000

Disease	[redacted] Residual Risk	Reproductive Risk
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, MMUT-related	1 in 12,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis III Gamma	< 1 in 20,000	< 1 in 1,000,000
Mucolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	1 in 670,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIA	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 26,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	< 1 in 50,000	< 1 in 1,000,000
Muscular Dystrophy, LAMA2-related	1 in 5,700	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 1,200	1 in 400,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 1 in 1,000,000
Neuronal Ceroid Lipofuscinosis, CLN6-related	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease Type C1	1 in 17,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-related	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	< 1 in 50,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	< 1 in 1,000,000
PCCA-related Propionic Acidemia	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 3,300	< 1 in 1,000,000
Pendred Syndrome	1 in 6,400	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 1	1 in 16,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 7,100	< 1 in 1,000,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 6,500	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 16,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 13,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 20,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Sandhoff Disease	1 in 18,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 9,700	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 14,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 17,000	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 1,[redacted]	1 in 820,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	< 1 in 1,000,000

Disease	[redacted] Residual Risk	Reproductive Risk
USH2A-related Disorders	1 in 7,200	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 14,000	< 1 in 1,000,000
Wilson Disease	1 in 9,000	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 36,000	1 in 140,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Juvenile Retinoschisis	1 in 840,000	< 1 in 1,000,000
X-linked Myotubular Myopathy	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000