# Expanded Carrier Screen by Next Generation Sequencing with Fragile X

**Patient:** [redacted]
DOB: [redacted] Age: 32 Sex: F
**Patient Identifiers:** [redacted]

**Visit Number (FIN):** [redacted]

Client: [redacted]

Physician: [redacted]

ARUP Test Code: 2014677

Collection Date: 10/30/2025
Received in lab: 11/02/2025
Completion Date: 11/12/2025

## 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).**

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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:** 11/11/2025

FEMALE
[redacted]
**DOB:** [redacted]
**Ethnicity:** African or African American
**Sample Type:** EDTA Blood
**Date of Collection:** 10/30/2025
**Date Received:** 11/04/2025
**Date Tested:** 11/10/2025
**Barcode:** [redacted]
**Accession ID:** [redacted]
**Indication:** Family history and/or partner positive

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<br>Universal Panel<br>Fundamental Plus Panel<br>Fundamental Panel<br>Fragile X Syndrome<br>**(176 conditions tested)** | N/A |
| **POSITIVE: CARRIER**<br>Alpha Thalassemia, HBA1/HBA2-related<br>**Reproductive Risk: Not Calculated**<br>**Inheritance: Autosomal Recessive** | ⊞ **CARRIER***<br>2 disease-causing mutations detected.<br>Alpha globin status: -a/-a. | Reproductive risk can be more accurately assessed after carrier screening of the partner. Carrier testing should be considered. See "Next Steps". |
| **UNCERTAIN CARRIER STATUS**<br>Spinal Muscular Atrophy<br>**Reproductive Risk: 1 in 8,800**<br>**Inheritance: Autosomal Recessive** | **UNCERTAIN CARRIER STATUS**<br>Positive for g.27134T>G SNP †<br>SMN1: 2 copies | 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". |

*Carriers generally do not experience symptoms.
†See the "Methods and Limitations" section below for more details.

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.

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**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] homozygote | N/A |
| **Alpha globin status** | -a/-a | 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/-a (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."

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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.

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## 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.

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## UNCERTAIN CARRIER STATUS

# Spinal Muscular Atrophy

**Reproductive risk: 1 in 8,800**
Risk before testing: 1 in 17,000

**Gene:** SMN1 | **Inheritance Pattern:** Autosomal Recessive

| **Patient** | [redacted] | **No partner tested** |
|---|---|---|
| **Result** | Uncertain Carrier Status | N/A |
| **Variant(s)** | Positive for g.27134T>G SNP †<br>SMN1: 2 copies | N/A |
| **Methodology** | Spinal muscular atrophy (v4.0) | N/A |
| **Interpretation** | This individual has 2 copies of SMN1 but is also positive for the g.27134T>G SNP. Based on this combination of results and African or African American ethnicity, the residual risk to be a carrier is 1 in 34. Consultation with a genetic counselor is recommended. | N/A |
| **Detection rate** | 71% | N/A |
| **Variants tested** | SMN1 copy number. | N/A |

†See the "Methods and Limitations" section below for more details.

## What is Spinal Muscular Atrophy?

Spinal muscular atrophy (SMA) is a condition that causes a loss (atrophy) of motor neurons, which are specific nerves in the brain and spinal cord that control movement. It is caused by a deficiency of the SMN protein, which is most often the result of a deletion (or loss) of part of the *SMN1* gene. Without motor neurons, messages cannot be passed from the brain to the muscles of the body. In severe cases, a patient will not be able to sit independently, and their breathing and swallowing may be impaired. In the mildest cases, symptoms begin in adulthood and independent movement, such as walking, may become more difficult (but still possible). With all types of SMA, there may be difficulties with sleeping and gaining weight (failure to thrive). Frequent pneumonia is common, as is curvature of the spine (scoliosis) and stiff joints (joint contractures). Intelligence is generally unaffected in individuals with SMA. Women with the milder forms of the condition have been known to give birth to healthy children, although many of the pregnancies have complications.

There are four main subtypes of SMA, each described below. It is not always possible to predict which type of SMA an individual will have based on their genetic testing results.

### TYPE 0

Type 0 is the most severe form of SMA. Symptoms can often be seen in the later stages of pregnancy, as the fetus is less active than expected. Once born, the infant will have little ability to move and may not be able to breathe and swallow independently. Infants with SMA type 0 often die before six months of age.

### TYPE I

Type I is another severe form of the condition. Symptoms typically develop within the first six months of life. Infants with type I SMA often have trouble breathing and swallowing. Their muscle tone and strength are extremely poor (hypotonia and muscle weakness, respectively); they cannot sit without support and will not achieve any motor skill milestones.

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**TYPE II**

In children with type II SMA, muscle weakness becomes apparent between the ages of six and twelve months. When placed in a sitting position, children with the condition can usually maintain the position without support; however, they often lose this ability by their mid-teens. Individuals with type II SMA cannot stand or walk without assistance. They have poor muscle tone and strength, and their fingers usually tremble uncontrollably.

**TYPE III**

Type III is a milder form of the condition. Symptoms begin sometime between the age of one year and early adulthood. As young children, these individuals may fall repeatedly and have trouble walking downstairs. While their muscles are weaker than normal, individuals with type III SMA can usually stand and walk without assistance, although they may lose this ability later in life. The legs are often more severely affected than the arms.

**TYPE IV**

Type IV is the mildest form of SMA. With this form of the condition, muscle weakness does not begin until the twenties or thirties, or potentially even later. This weakness is often mild to moderate, and the individual is generally able to walk and move independently. They may also experience mild to moderate tremors and/or twitching of the muscles.

## How common is Spinal Muscular Atrophy?

The incidence of SMA in the population is between 1 in every 6,000 to 20,000 individuals. SMA is more common in White populations.

## How is Spinal Muscular Atrophy treated?

There is no cure for SMA. Many available treatments are supportive in nature and are aimed at improving the symptoms that are present in individuals with the condition. For children with the more severe forms of SMA, mechanical breathing aids may help with sleep and prolong lifespan. In addition, placement of a feeding tube may ensure proper nutrition in those with swallowing problems or feeding difficulties. For individuals with milder forms of SMA, certain types of respiratory assistance may help with sleep problems and surgery may be used to treat orthopedic issues.

In addition to the treatments for SMA symptoms, medications are now available that have been shown to improve motor development in individuals with the condition. These medications, known as nusinersen (market name Spinraza®), onasemnogene abeparvovec (market name Zolgensma®), and risdiplam (market name Evrysdi®), have been approved in the United States for use in patients with SMA.

## What is the prognosis for an individual with Spinal Muscular Atrophy?

The prognosis for an individual with SMA varies greatly depending on which type they have and their treatment course. With newly available treatments, the prognosis of the condition may change over time. Type 0 SMA is typically fatal between two and six months of age. Children with type I SMA may live longer with the aid of mechanical breathing aids and other available therapies. With type II SMA, 75% of those affected live to the age of 25.

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# Methods and Limitations

[redacted] **[Foresight Carrier Screen]**: Sequencing with copy number analysis, FMR1 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 v5.0).

## 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. 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 genomic features (e.g., homology or homopolymers) impede reliable variant detection may be assayed using alternate technology, have reduced sensitivity, or be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for full exon-level deletions and duplications with a sensitivity of ~99%. Only full exon-level deletions and larger are assayed for other genes on the panel with a sensitivity of ≥75%. Partial exon and single or multi-exon deletions in regions of genomic or assay complexity may have reduced sensitivity or may be excluded from the ROI. Selected founder deletions may be detected at higher sensitivity. Affected exons and/or breakpoints of copy number variants (CNVs) are estimated from tiled regions and, when available, using junction reads. Only exons included in the region affected by the CNV are included in the variant nomenclature. In some cases, the CNV may be larger or smaller than indicated and/or may involve breakpoint sequence complexity that is not reflected in the variant nomenclature. If *GJB2* is in the ordered ROI, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed. If in the ordered ROI, the following are not assessed: CCG trinucleotide repeat expansions in *AFF2*; polyalanine expansions in exon 2 of *ARX*; variants in the *ORF15* region of *RPGR*; introns 1 and 22 inversions in *F8*.

## FMR1 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 ± one 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.

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## 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. In some instances, additional unreported pseudogene-derived variants may be present in the same gene copy as the reported variant (e.g. chimeric alleles). The ability of other assays to detect this complex genotype is dependent on the specific test methodology utilized.

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. If the 30kb deletion (aka *CYP21A2* deletion) is reported, the extent of impacted gene sequence may be attenuated in length or may extend through the end of the gene (i.e. full gene deletion). If present, an attenuated deletion may be associated with less severe or non-classic CAH (PMID: 22156666, 20970527, 30611409, 29450859). If further testing is pursued for the 30kb deletion, the methodology should accommodate deletions of variable size. 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, more than two additional copies of either gene may be present but will not be reported. 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. Variants and/or haplotypes associated with a non-Mendelian risk for disease are only reported on request. 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.

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## 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 and/or classification leading to false negative or false positive results. 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, variants that interfere with analysis, technical or analytical errors, and other sources.

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 Erik Zmuda, PhD, FACMG, CGMB on Nov 11, 2025

Genetic testing was completed by CLIA accredited laboratories in the United States located at: 233 E Grand Ave, South San Francisco, CA 94080. CLIA IDs: 05D2299797.
The following personnel codes and digital laboratory director signature may reflect remote review of digital data: 6609, 6633, 30523

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# Conditions Tested

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

**Adenosine Deaminase Deficiency** - **Gene:** ADA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000022:1-12. **Detection Rate:** African or African American 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:** African or African American >99%.

**Alpha-mannosidosis** - **Gene:** MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000528:1-23. **Detection Rate:** African or African American 98%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Canavan Disease** - **Gene:** ASPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000049:1-6. **Detection Rate:** African or African American 89%.

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

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

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

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

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

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

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

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

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

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

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

**Congenital Adrenal Hyperplasia, CYP11B1-related** - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** African or African American 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:** African or African American 92%.

**Congenital Disorder of Glycosylation, ALG6-related** - **Gene:** ALG6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_013339:2-15. **Detection Rate:** African or African American >99%.

**Congenital Disorder of Glycosylation, MPI-related** - **Gene:** MPI. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002435:1-8. **Detection Rate:** African or African American >99%.

**Congenital Disorder of Glycosylation, PMM2-related** - **Gene:** PMM2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000303:1-8. **Detection Rate:** African or African American >99%.

**Costeff Optic Atrophy Syndrome** - **Gene:** OPA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_025136:1-2. **Detection Rate:** African or African American >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:** African or African American >99%.

**Cystinosis** - **Gene:** CTNS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004937:3-12. **Detection Rate:** African or African American >99%.

**Delta-sarcoglycanopathy** - **Gene:** SGCD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000337:2-9. **Detection Rate:** African or African American 96%.

**Dihydrolipoamide Dehydrogenase Deficiency** - **Gene:** DLD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000108:1-14. **Detection Rate:** African or African American >99%.

**Dysferlinopathy** - **Gene:** DYSF. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003494:1-55. **Detection Rate:** African or African American 98%.

**Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)** - **Gene:** DMD. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_004006:1-79. **Detection Rate:** African or African American 99%.

**ERCC6-related Disorders** - **Gene:** ERCC6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000124:2-21. **Detection Rate:** African or African American 96%.

**ERCC8-related Disorders** - **Gene:** ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000082:1-12. **Detection Rate:** African or African American 97%.

**EVC-related Ellis-van Creveld Syndrome** - **Gene:** EVC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153717:1-21. **Detection Rate:** African or African American 96%.

**EVC2-related Ellis-van Creveld Syndrome** - **Gene:** EVC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_147127:1-22. **Detection Rate:** African or African American 98%.

**Fabry Disease** - **Gene:** GLA. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000169:1-7. **Detection Rate:** African or African American 98%.

**Familial Dysautonomia** - **Gene:** ELP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003640:2-37. **Detection Rate:** African or African American >99%.

**Familial Hyperinsulinism, ABCC8-related** - **Gene:** ABCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000352:1-39. **Detection Rate:** African or African American >99%.

**Familial Hyperinsulinism, KCNJ11-related** - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000525:1. **Detection Rate:** African or African American >99%.

**Familial Mediterranean Fever** - **Gene:** MEFV. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000243:1-10. **Detection Rate:** African or African American >99%.

**Fanconi Anemia Complementation Group A** - **Gene:** FANCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000135:1-43. **Detection Rate:** African or African American 92%.

**Fanconi Anemia, FANCC-related** - **Gene:** FANCC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000136:2-15. **Detection Rate:** African or African American >99%.

**FKRP-related Disorders** - **Gene:** FKRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_024301:4. **Detection Rate:** African or African American >99%.

**FKTN-related Disorders** - **Gene:** FKTN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001079802:3-11. **Detection Rate:** African or African American >99%.

**Fragile X Syndrome** - **Gene:** FMR1. X-linked Dominant. FMR1 Triplet repeat detection. **Variant (1):** FMR1 CGG repeat number. **Detection Rate:** African or African American >99%.

**Free Sialic Acid Storage Disorders** - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012434:1-11. **Detection Rate:** African or African American 98%.

**Galactokinase Deficiency** - **Gene:** GALK1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000154:1-8. **Detection Rate:** African or African American >99%.

**Galactosemia** - **Gene:** GALT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000155:1-11. **Detection Rate:** African or African American >99%.

**Gamma-sarcoglycanopathy** - **Gene:** SGCG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000231:2-8. **Detection Rate:** African or African American 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:** African or African American 60%.

**GLB1-related Disorders** - **Gene:** GLB1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000404:1-16. **Detection Rate:** African or African American >99%.

**Glutaric Acidemia, GCDH-related** - **Gene:** GCDH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000159:2-12. **Detection Rate:** African or African American >99%.

**Glycine Encephalopathy, AMT-related** - **Gene:** AMT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000481:1-9. **Detection Rate:** African or African American >99%.

**Glycine Encephalopathy, GLDC-related** - **Gene:** GLDC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000170:1-25. **Detection Rate:** African or African American 94%.

**Glycogen Storage Disease Type Ia** - **Gene:** G6PC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000151:1-5. **Detection Rate:** African or African American 98%.

**Glycogen Storage Disease Type Ib** - **Gene:** SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001164277:3-10. **Detection Rate:** African or African American >99%.

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**Glycogen Storage Disease Type III** - **Gene:** AGL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000642:2-34. **Detection Rate:** African or African American >99%.
**GNE Myopathy** - **Gene:** GNE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001128227:1-12. **Detection Rate:** African or African American >99%.
**GNPTAB-related Disorders** - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024312:1-21. **Detection Rate:** African or African American >99%.
**HADHA-related Disorders** - **Gene:** HADHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000182:1-20. **Detection Rate:** African or African American >99%.
**Hereditary Fructose Intolerance** - **Gene:** ALDOB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000035:2-9. **Detection Rate:** African or African American >99%.
**Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** - **Gene:** HEXA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000520:1-14. **Detection Rate:** African or African American >99%.
**HMG-CoA Lyase Deficiency** - **Gene:** HMGCL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000191:1-9. **Detection Rate:** African or African American >99%.
**Holocarboxylase Synthetase Deficiency** - **Gene:** HLCS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000411:4-12. **Detection Rate:** African or African American >99%.
**Homocystinuria, CBS-related** - **Gene:** CBS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000071:3-17. **Detection Rate:** African or African American >99%.
**HSD17B4-related Disorders** - **Gene:** HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000414:1-24. **Detection Rate:** African or African American 98%.
**Hydrolethalus Syndrome** - **Gene:** HYLS1. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_145014:4. **Detection Rate:** African or African American >99%.
**Hypophosphatasia** - **Gene:** ALPL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000478:2-12. **Detection Rate:** African or African American >99%.
**Isovaleric Acidemia** - **Gene:** IVD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002225:1-12. **Detection Rate:** African or African American >99%.
**Joubert Syndrome 2** - **Gene:** TMEM216. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001173990:1-5. **Detection Rate:** African or African American >99%.
**Junctional Epidermolysis Bullosa, LAMA3-related** - **Gene:** LAMA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000227:1-38. **Detection Rate:** African or African American >99%.
**Junctional Epidermolysis Bullosa, LAMB3-related** - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228:2-23. **Detection Rate:** African or African American >99%.
**Junctional Epidermolysis Bullosa, LAMC2-related** - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** African or African American >99%.
**Krabbe Disease** - **Gene:** GALC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000153:1-17. **Detection Rate:** African or African American >99%.
**Leigh Syndrome, French-Canadian Type** - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133259:1-38. **Detection Rate:** African or African American >99%.
**Lipoid Congenital Adrenal Hyperplasia** - **Gene:** STAR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000349:1-7. **Detection Rate:** African or African American >99%.
**Lysosomal Acid Lipase Deficiency** - **Gene:** LIPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000235:2-10. **Detection Rate:** African or African American 98%.
**Maple Syrup Urine Disease Type Ia** - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000709:1-9. **Detection Rate:** African or African American >99%.
**Maple Syrup Urine Disease Type Ib** - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_183050:1-10. **Detection Rate:** African or African American >99%.
**Maple Syrup Urine Disease Type II** - **Gene:** DBT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001918:1-11. **Detection Rate:** African or African American 97%.
**Medium-chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000016:1-12. **Detection Rate:** African or African American >99%.
**Megalencephalic Leukoencephalopathy with Subcortical Cysts** - **Gene:** MLC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015166:2-12. **Detection Rate:** African or African American >99%.
**Metachromatic Leukodystrophy** - **Gene:** ARSA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000487:1-8. **Detection Rate:** African or African American >99%.
**Methylmalonic Acidemia, cblA Type** - **Gene:** MMAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_172250:2-7. **Detection Rate:** African or African American >99%.
**Methylmalonic Acidemia, cblB Type** - **Gene:** MMAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_052845:1-9. **Detection Rate:** African or African American >99%.
**Methylmalonic Acidemia, MMUT-related** - **Gene:** MMUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** African or African American >99%.
**Methylmalonic Aciduria and Homocystinuria, cblC Type** - **Gene:** MMACHC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015506:1-4. **Detection Rate:** African or African American >99%.
**MKS1-related Disorders** - **Gene:** MKS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017777:1-18. **Detection Rate:** African or African American >99%.
**Mucolipidosis III Gamma** - **Gene:** GNPTG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032520:1-11. **Detection Rate:** African or African American 98%.
**Mucolipidosis IV** - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_020533:1-14. **Detection Rate:** African or African American >99%.
**Mucopolysaccharidosis Type I** - **Gene:** IDUA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000203:1-14. **Detection Rate:** African or African American 97%.
**Mucopolysaccharidosis Type II** - **Gene:** IDS. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000202:1-9. **Detection Rate:** African or African American 89%.
**Mucopolysaccharidosis Type IIIA** - **Gene:** SGSH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000199:1-8. **Detection Rate:** African or African American >99%.
**Mucopolysaccharidosis Type IIIB** - **Gene:** NAGLU. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000263:1-6. **Detection Rate:** African or African American >99%.
**Mucopolysaccharidosis Type IIIC** - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_152419:1-18. **Detection Rate:** African or African American >99%.
**Muscular Dystrophy, LAMA2-related** - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-43,45-65. **Detection Rate:** African or African American 98%.
**MYO7A-related Disorders** - **Gene:** MYO7A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000260:2-49. **Detection Rate:** African or African American >99%.
**NEB-related Nemaline Myopathy** - **Gene:** NEB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** African or African American 92%.

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**Nephrotic Syndrome, NPHS1-related** - **Gene:** NPHS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004646:1-29. **Detection Rate:** African or African American >99%.
**Nephrotic Syndrome, NPHS2-related** - **Gene:** NPHS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014625:1-8. **Detection Rate:** African or African American >99%.
**Neuronal Ceroid Lipofuscinosis, CLN6-related** - **Gene:** CLN6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017882:1-7. **Detection Rate:** African or African American 96%.
**Neuronal Ceroid Lipofuscinosis, PPT1-related** - **Gene:** PPT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000310:1-9. **Detection Rate:** African or African American >99%.
**Niemann-Pick Disease Type C1** - **Gene:** NPC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000271:1-25. **Detection Rate:** African or African American >99%.
**Niemann-Pick Disease Type C2** - **Gene:** NPC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006432:1-5. **Detection Rate:** African or African American >99%.
**Niemann-Pick Disease, SMPD1-related** - **Gene:** SMPD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000543:1-6. **Detection Rate:** African or African American >99%.
**Nijmegen Breakage Syndrome** - **Gene:** NBN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002485:1-16. **Detection Rate:** African or African American >99%.
**Nonsyndromic Hearing Loss, GJB2-related** - **Gene:** GJB2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004004:1-2. **Detection Rate:** African or African American >99%.
**Ornithine Transcarbamylase Deficiency** - **Gene:** OTC. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000531:1-10. **Detection Rate:** African or African American 97%.
**PCCA-related Propionic Acidemia** - **Gene:** PCCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000282:1-24. **Detection Rate:** African or African American 95%.
**PCCB-related Propionic Acidemia** - **Gene:** PCCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000532:1-15. **Detection Rate:** African or African American >99%.
**PCDH15-related Disorders** - **Gene:** PCDH15. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_033056:2-33. **Detection Rate:** African or African American 93%.
**Pendred Syndrome** - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000441:2-21. **Detection Rate:** African or African American >99%.
**Peroxisome Biogenesis Disorder Type 1** - **Gene:** PEX1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000466:1-24. **Detection Rate:** African or African American >99%.
**Peroxisome Biogenesis Disorder Type 3** - **Gene:** PEX12. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000286:1-3. **Detection Rate:** African or African American >99%.
**Peroxisome Biogenesis Disorder Type 4** - **Gene:** PEX6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000287:1-17. **Detection Rate:** African or African American 97%.
**Peroxisome Biogenesis Disorder Type 5** - **Gene:** PEX2. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000318:4. **Detection Rate:** African or African American >99%.
**Peroxisome Biogenesis Disorder Type 6** - **Gene:** PEX10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153818:1-6. **Detection Rate:** African or African American >99%.
**PEX7-related Disorders** - **Gene:** PEX7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000288:1-10. **Detection Rate:** African or African American >99%.
**Phenylalanine Hydroxylase Deficiency** - **Gene:** PAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000277:1-13. **Detection Rate:** African or African American >99%.
**POMGNT1-related Disorders** - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017739:2-22. **Detection Rate:** African or African American 96%.
**Pompe Disease** - **Gene:** GAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000152:2-20. **Detection Rate:** African or African American >99%.
**Primary Carnitine Deficiency** - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003060:1-10. **Detection Rate:** African or African American >99%.
**Primary Hyperoxaluria Type 1** - **Gene:** AGXT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000030:1-11. **Detection Rate:** African or African American >99%.
**Primary Hyperoxaluria Type 2** - **Gene:** GRHPR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012203:1-9. **Detection Rate:** African or African American >99%.
**Primary Hyperoxaluria Type 3** - **Gene:** HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** African or African American >99%.
**Pycnodysostosis** - **Gene:** CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000396:2-8. **Detection Rate:** African or African American >99%.
**Pyruvate Carboxylase Deficiency** - **Gene:** PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000920:3-22. **Detection Rate:** African or African American >99%.
**RTEL1-related Disorders** - **Gene:** RTEL1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001283009:2-35. **Detection Rate:** African or African American 99%.
**Sandhoff Disease** - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** African or African American 98%.
**Short-chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000017:1-10. **Detection Rate:** African or African American >99%.
**Sjogren-Larsson Syndrome** - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** African or African American 96%.
**SLC26A2-related Disorders** - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000112:2-3. **Detection Rate:** African or African American >99%.
**Smith-Lemli-Opitz Syndrome** - **Gene:** DHCR7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001360:3-9. **Detection Rate:** African or African American >99%.
**Spastic Paraplegia Type 15** - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015346:2-42. **Detection Rate:** African or African American >99%.
**Spinal Muscular Atrophy** - **Gene:** SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** African or African American 71%.
**Spondylothoracic Dysostosis** - **Gene:** MESP2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** African or African American 93%.
**TGM1-related Autosomal Recessive Congenital Ichthyosis** - **Gene:** TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000359:2-15. **Detection Rate:** African or African American >99%.
**TPP1-related Neuronal Ceroid Lipofuscinosis** - **Gene:** TPP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000391:1-13. **Detection Rate:** African or African American >99%.
**Tyrosine Hydroxylase Deficiency** - **Gene:** TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_199292:1-14. **Detection Rate:** African or African American >99%.
**Tyrosinemia Type I** - **Gene:** FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** African or African American >99%.

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**Tyrosinemia Type II** - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** African or African American >99%.

**USH1C-related Disorders** - **Gene:** USH1C. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005709:1-21. **Detection Rate:** African or African American >99%.

**USH2A-related Disorders** - **Gene:** USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_206933:2-72. **Detection Rate:** African or African American 98%.

**Usher Syndrome Type 3** - **Gene:** CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** African or African American >99%.

**Very-long-chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADVL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000018:1-20. **Detection Rate:** African or African American >99%.

**Wilson Disease** - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** African or African American >99%.

**X-linked Adrenal Hypoplasia Congenita** - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** African or African American 97%.

**X-linked Adrenoleukodystrophy** - **Gene:** ABCD1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000033:1-6. **Detection Rate:** African or African American 77%.

**X-linked Alport Syndrome** - **Gene:** COL4A5. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000495:1-51. **Detection Rate:** African or African American 96%.

**X-linked Juvenile Retinoschisis** - **Gene:** RS1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000330:1-6. **Detection Rate:** African or African American 98%.

**X-linked Myotubular Myopathy** - **Gene:** MTM1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000252:2-15. **Detection Rate:** African or African American 96%.

**X-linked Severe Combined Immunodeficiency** - **Gene:** IL2RG. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000206:1-8. **Detection Rate:** African or African American >99%.

**Xeroderma Pigmentosum Group A** - **Gene:** XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** African or African American >99%.

**Xeroderma Pigmentosum Group C** - **Gene:** XPC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004628:1-16. **Detection Rate:** African or African American 97%.

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# 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.

*See the "Methods and Limitations" section above for more details.

| 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] homozygote †<br>Alpha globin status: -a/-a. | Not calculated |
| Alpha-mannosidosis | 1 in 15,000 | < 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 13,000 | < 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 41,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS10-related | < 1 in 50,000 | < 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 700 | 1 in 21,000 |
| Beta-sarcoglycanopathy | 1 in 39,000 | < 1 in 1,000,000 |
| Biotinidase Deficiency | 1 in 38,000 | < 1 in 1,000,000 |
| Bloom Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Calpainopathy | 1 in 7,400 | < 1 in 1,000,000 |
| Canavan Disease | 1 in 1,400 | 1 in 900,000 |
| 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 |

Chart continues on following page(s)

| Disease | [redacted]<br>Residual Risk | Reproductive Risk |
|---|---|---|
| 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 |
| Congenital Adrenal Hyperplasia, CYP21A2-related | 1 in 1,700 | 1 in 930,000 |
| Congenital Disorder of Glycosylation, ALG6-related | < 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 6,100 | < 1 in 1,000,000 |
| Cystinosis | 1 in 22,000 | < 1 in 1,000,000 |
| Delta-sarcoglycanopathy | < 1 in 13,000 | < 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 530 | 1 in 12,000 |
| Fanconi Anemia Complementation Group A | 1 in 3,100 | < 1 in 1,000,000 |
| Fanconi Anemia, FANCC-related | < 1 in 50,000 | < 1 in 1,000,000 |
| FKRP-related Disorders | 1 in 42,000 | < 1 in 1,000,000 |
| FKTN-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| Fragile X Syndrome | Normal: 32 and 32 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 7,000 | < 1 in 1,000,000 |
| Gamma-sarcoglycanopathy | 1 in 2,600 | < 1 in 1,000,000 |
| Gaucher Disease | 1 in 310 | 1 in 150,000 |
| 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 23,000 | < 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 27,000 | < 1 in 1,000,000 |
| HSD17B4-related Disorders | 1 in 9,000 | < 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 17,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type Ia | 1 in 26,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type Ib | 1 in 36,000 | < 1 in 1,000,000 |

Chart continues on following page(s)

| Disease | [redacted]<br>Residual Risk | Reproductive Risk |
|---|---|---|
| Maple Syrup Urine Disease Type II | 1 in 13,000 | < 1 in 1,000,000 |
| Medium-chain Acyl-CoA Dehydrogenase Deficiency | 1 in 6,000 | < 1 in 1,000,000 |
| 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 18,000 | < 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 5,600 | < 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 11,000 | < 1 in 1,000,000 |
| Neuronal Ceroid Lipofuscinosis, PPT1-related | 1 in 7,700 | < 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 |
| Nonsyndromic Hearing Loss, GJB2-related | 1 in 3,700 | 1 in 560,000 |
| Ornithine Transcarbamylase Deficiency | 1 in 530,000 | < 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 |
| PEX7-related Disorders | 1 in 16,000 | < 1 in 1,000,000 |
| Phenylalanine Hydroxylase Deficiency | 1 in 11,000 | < 1 in 1,000,000 |
| POMGNT1-related Disorders | < 1 in 12,000 | < 1 in 1,000,000 |
| Pompe Disease | 1 in 5,900 | < 1 in 1,000,000 |
| Primary Carnitine Deficiency | 1 in 16,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 1 | 1 in 20,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 2 | < 1 in 50,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 3 | < 1 in 50,000 | < 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 |
| RTEL1-related Disorders | < 1 in 37,000 | < 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 16,000 | < 1 in 1,000,000 |
| Smith-Lemli-Opitz Syndrome | 1 in 18,000 | < 1 in 1,000,000 |
| Spastic Paraplegia Type 15 | < 1 in 50,000 | < 1 in 1,000,000 |
| Spinal Muscular Atrophy | Positive for g.27134T>G SNP [*]<br>SMN1: 2 copies [†]<br>1 in 34 | 1 in 8,800 |
| Spondylothoracic Dysostosis | < 1 in 6,800 | < 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 |

Chart continues on following page(s)

| Disease | [redacted]<br>Residual Risk | Reproductive Risk |
|---|---|---|
| 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 |
| USH2A-related Disorders | 1 in 5,900 | < 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 |

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