myriad WOMEN'S HEALTH

RESULTS RECIPIENT
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**Report Date:** 11/08/2021

MALE
**DONOR 14281**
**DOB:**
**Ethnicity:** South Asian
**Sample Type:** EDTA Blood
**Date of Collection:** 10/20/2021
**Date Received:** 10/21/2021
**Date Tested:** 11/06/2021
**Barcode:** 11004513001914
**Accession ID:** CSLQY9HHU32QPY4
**Indication:** Egg or sperm donor

FEMALE
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 | DONOR 14281 | Partner |
|---|---|---|
| Panel Information | Foresight Carrier Screen<br>Universal Panel<br>Fundamental Plus Panel<br>Fundamental Panel<br>**(175 conditions tested)** | N/A |
| **POSITIVE: CARRIER**<br>GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness<br>**Reproductive Risk: 1 in 160**<br>**Inheritance: Autosomal Recessive** | **CARRIER***<br>NM_004004.5(GJB2):c.35delG (aka p.G12Vfs*2) 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**<br>Spinal Muscular Atrophy<br>**Reproductive Risk: 1 in 200**<br>**Inheritance: Autosomal Recessive** | **CARRIER***<br>SMN1: 1 copy | 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**<br>Alpha Thalassemia, HBA1/HBA2-related<br>**Reproductive Risk: Not Calculated**<br>**Inheritance: Autosomal Recessive** | **CARRIER***<br>1 disease-causing mutation detected.<br>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 13.

### 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.

**POSITIVE: CARRIER**

# GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness

**Reproductive risk: 1 in 160**
Risk before testing: 1 in 6,600

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

| **Patient** | **DONOR 14281** | **No partner tested** |
|---|---|---|
| **Result** | ✚ Carrier | N/A |
| **Variant(s)** | NM_004004.5(GJB2):c.35delG(aka p.G12Vfs*2) heterozygote | N/A |
| **Methodology** | Sequencing with copy number analysis (v3.1) | 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.

**POSITIVE: CARRIER**

# Spinal Muscular Atrophy

**Reproductive risk: 1 in 200**
Risk before testing: 1 in 9,800

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

| Patient | DONOR 14281 | No partner tested |
|---|---|---|
| **Result** | ✚ Carrier | N/A |
| **Variant(s)** | SMN1: 1 copy | N/A |
| **Methodology** | Spinal muscular atrophy (v3.0) | N/A |
| **Interpretation** | This individual is a carrier of spinal muscular atrophy. Carriers generally do not experience symptoms. | N/A |
| **Detection rate** | 93% | N/A |
| **Variants tested** | SMN1 copy number. | N/A |

## What Is Spinal Muscular Atrophy?

Spinal muscular atrophy (SMA) is a condition that causes a loss 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. 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.

With all types of SMA, there may be difficulties with sleeping and gaining weight. Frequent pneumonia is common, as is curvature of the spine (scoliosis) and stiff joints. 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.

### 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, ALSO CALLED WERDNIG-HOFFMANN DISEASE

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; they cannot sit without support and will not achieve any motor-skill milestones.

### TYPE II, ALSO CALLED DUBOWITZ DISEASE

In children with type II SMA, muscle weakness becomes apparent between the ages of 6 and 12 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, ALSO CALLED KUGELBERG-WELANDER DISEASE

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 one's 20s or 30s, 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?

In the United States, the prevalence of SMA is estimated to be between 1 in every 6000 to 10,000 individuals. The condition is found in individuals of every race and ethnic background, but it is most common among Caucasians.

## How Is Spinal Muscular Atrophy Treated?

There is no cure for SMA. The majority of 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 symptomatic treatments for SMA, a medication is now available that has been shown to improve motor development in infants and children with the condition. This medication, known as nusinersen (market name Spinraza™), has been approved in the United States for use in pediatric and adult 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 of SMA he or she has and their treatment course.

#### TYPE 0

Type 0 SMA is typically fatal between two and six months of age. These infants do not develop any motor skills expected of infants their age.

#### TYPE I

This type of SMA is usually fatal within two years. However, children with type I SMA may live longer with the aid of mechanical breathing aids and other available therapies. There are a few known cases in which the individual survived to adolescence or early adulthood.

#### TYPE II

With type II SMA, 75% of those affected live to the age of 25. They are often able to sit independently when placed in a sitting position, but lose this ability by their mid-teens.

**TYPE III**

Individuals with type III SMA may have a normal lifespan. Many learn to walk independently, although most lose the ability to do so by their thirties or forties.

**TYPE IV**

A normal lifespan is also possible for individuals with type IV SMA. They do not develop symptoms until their twenties or thirties and usually retain the ability to walk independently.

**POSITIVE: CARRIER**

# Alpha Thalassemia, HBA1/HBA2-related

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

| Patient | DONOR 14281 | 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 (v3.2) | 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 as alpha and beta globin. Alpha thalassemia is caused by harmful genetic changes (mutations) 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 each individual has four gene copies that make up the alpha chain of their hemoglobin (two *HBA1* and two *HBA2*). Individuals can inherit a mutation in one, two, three, or all four gene copies. There are also different types of mutations within the *HBA1* and *HBA2* genes. Larger mutations that remove most or all of a gene are called “deletional,” while smaller mutations are called “non-deletional.”

The symptoms associated with alpha thalassemia can range from mild anemia to fetal death. The symptoms depend on how many and what types of mutations an individual inherits. Typically, individuals with mutations in more of their alpha globin genes have more severe symptoms. Additionally, non-deletional mutations generally cause more severe symptoms than deletional mutations. Individuals can inherit a combination of deletional and non-deletional mutations.

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 mutations in *HBA1* and *HBA2* are recommended to consult with a genetics professional to determine both their personal risk for disease and their reproductive risk.

#### SILENT CARRIER

Silent carriers of alpha thalassemia have a mutation 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 mutation in two of the four alpha globin genes. Individuals with this finding generally have RBCs that are pale and small when visualized (mild anemia). 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 mutations in *HBA1* and/or *HBA2* who have a diagnosis of hemoglobin H (HbH) disease (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 mutations in three of the four alpha globin genes. This form is highly variable, and symptoms depend on the type of mutations 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 fatigue, anemia, yellowing of the skin or eyes (jaundice), enlargement of the spleen, and other more rare or minor complications.

#### HEMOGLOBIN BART SYNDROME

Hemoglobin Bart (Hb Bart) syndrome is typically the result of mutations 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). 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. Most babies with this condition are stillborn or die soon after birth. When fetal blood transfusions are successful, survival is possible; however, there is high risk for intellectual and physical disability in these survivors.

## 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, iron chelation therapy (removal of excess iron from the body), 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 *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 build-up 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

**DONOR 14281 [Foresight Carrier Screen]**: Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

## Sequencing with copy number analysis

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 a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected non-coding bases 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 regions of homology impede reliable variant detection may be assayed using alternate technology, or they may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with an average sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel and such deletions are detected with a sensitivity of ≥75%. Selected founder deletions may be detected at slightly higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from junction reads, where available, or using the positions of affected probes. Only exons known to be included in the region affected by a copy number variant are provided 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.

## Spinal muscular atrophy

Targeted copy number analysis via high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. Other genetic variants 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 copies of *SMN1*.

## Analysis of homologous regions

A combination of high-throughput sequencing, read-depth-based copy number analysis, and targeted genotyping is used to determine the number of functional gene copies and/or the presence of selected loss-of-function variants in certain genes that have 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 CAH, depending on the chromosomal location of the variants (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 similar rare carrier states, where complementary changes exist between the 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 numbers on the report are based only 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

High-throughput sequencing and read-depth-based 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 listed exons plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g , long 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 similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay.

## Interpretation of reported variants

The classification and interpretation of all variants identified in this assay reflects the current state of Myriad's scientific understanding at the time this report was issued. 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, 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 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 more patients. f 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 an updated report is issued, the variants reported may change based on their current classification. 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 MWH 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 lie on the same chromosome or different chromosomes (phase). This test is not designed to detect sex-chromosome copy number variations. If present, sex-chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Variant interpretation and residual and reproductive risk estimations assume a normal karyotype and may be different for individuals with abnormal karyotypes. The test does not fully address all inherited forms of intellectual disability, birth defects, or heritable diseases. Furthermore, not all forms of genetic variation are detected by this assay (i.e., 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 the 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. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: #05D1102604.

**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.

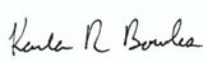
If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

# 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, PhD, FACMG, CGMB

Report content approved by Maria Alfaro, PhD, FACMG, CGMB on Nov 8, 2021

# Conditions Tested

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**CLN3-related Neuronal Ceroid Lipofuscinosis** - **Gene:** CLN3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001042432 2-16. **Detection Rate:** South Asian >99%.

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

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

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

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

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

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

**Congenital Adrenal Hyperplasia, CYP11B1-related** - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** South Asian 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:** South Asian 88%.

**Congenital Disorder of Glycosylation Type Ia** - **Gene:** PMM2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000303:1-8. **Detection Rate:** South Asian >99%.

**Congenital Disorder of Glycosylation Type Ic** - **Gene:** ALG6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_013339:2-15. **Detection Rate:** South Asian >99%.
**Congenital Disorder of Glycosylation, MPI-related** - **Gene:** MPI. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002435:1-8. **Detection Rate:** South Asian >99%.
**Costeff Optic Atrophy Syndrome** - **Gene:** OPA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_025136:1-2. **Detection Rate:** South Asian >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:** South Asian >99%.
**Cystinosis** - **Gene:** CTNS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004937:3-12. **Detection Rate:** South Asian >99%.
**D-bifunctional Protein Deficiency** - **Gene:** HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000414:1-24. **Detection Rate:** South Asian 98%.
**Delta-sarcoglycanopathy** - **Gene:** SGCD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000337:2-9. **Detection Rate:** South Asian 96%.
**Dihydrolipoamide Dehydrogenase Deficiency** - **Gene:** DLD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000108:1-14. **Detection Rate:** South Asian >99%.
**Dysferlinopathy** - **Gene:** DYSF. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003494:1-55. **Detection Rate:** South Asian 98%.
**Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)** - **Gene:** DMD. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_004006:1-79. **Detection Rate:** South Asian 99%.
**ERCC6-related Disorders** - **Gene:** ERCC6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000124:2-21. **Detection Rate:** South Asian 96%.
**ERCC8-related Disorders** - **Gene:** ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000082:1-12. **Detection Rate:** South Asian 97%.
**EVC-related Ellis-van Creveld Syndrome** - **Gene:** EVC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153717:1-21. **Detection Rate:** South Asian 96%.
**EVC2-related Ellis-van Creveld Syndrome** - **Gene:** EVC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_147127:1-22. **Detection Rate:** South Asian 98%.
**Fabry Disease** - **Gene:** GLA. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000169:1-7. **Detection Rate:** South Asian 98%.
**Familial Dysautonomia** - **Gene:** ELP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003640:2-37. **Detection Rate:** South Asian >99%.
**Familial Hyperinsulinism, ABCC8-related** - **Gene:** ABCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000352:1-39. **Detection Rate:** South Asian >99%.
**Familial Hyperinsulinism, KCNJ11-related** - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000525:1. **Detection Rate:** South Asian >99%.
**Familial Mediterranean Fever** - **Gene:** MEFV. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000243:1-10. **Detection Rate:** South Asian >99%.
**Fanconi Anemia Complementation Group A** - **Gene:** FANCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000135:1-43. **Detection Rate:** South Asian 92%.
**Fanconi Anemia, FANCC-related** - **Gene:** FANCC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000136:2-15. **Detection Rate:** South Asian >99%.
**FKRP-related Disorders** - **Gene:** FKRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_024301:4. **Detection Rate:** South Asian >99%.
**FKTN-related Disorders** - **Gene:** FKTN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001079802:3-11. **Detection Rate:** South Asian >99%.
**Free Sialic Acid Storage Disorders** - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012434:1-11. **Detection Rate:** South Asian 98%.
**Galactokinase Deficiency** - **Gene:** GALK1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000154:1-8. **Detection Rate:** South Asian >99%.
**Galactosemia** - **Gene:** GALT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000155:1-11. **Detection Rate:** South Asian >99%.
**Gamma-sarcoglycanopathy** - **Gene:** SGCG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000231:2-8. **Detection Rate:** South Asian 87%.
**Gaucher Disease** - **Gene:** GBA. Autosomal Recessive. Analysis of homologous regions. **Variants (10):** D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. **Detection Rate:** South Asian 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:** South Asian >99%.
**GLB1-related Disorders** - **Gene:** GLB1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000404:1-16. **Detection Rate:** South Asian >99%.
**Glutaric Acidemia, GCDH-related** - **Gene:** GCDH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000159:2-12. **Detection Rate:** South Asian >99%.
**Glycine Encephalopathy, AMT-related** - **Gene:** AMT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000481:1-9. **Detection Rate:** South Asian >99%.
**Glycine Encephalopathy, GLDC-related** - **Gene:** GLDC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000170:1-25. **Detection Rate:** South Asian 94%.
**Glycogen Storage Disease Type Ia** - **Gene:** G6PC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000151:1-5. **Detection Rate:** South Asian 98%.
**Glycogen Storage Disease Type Ib** - **Gene:** SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001164277 3-11. **Detection Rate:** South Asian >99%.
**Glycogen Storage Disease Type III** - **Gene:** AGL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000642:2-34. **Detection Rate:** South Asian >99%.
**GNE Myopathy** - **Gene:** GNE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001128227:1-12. **Detection Rate:** South Asian >99%.
**GNPTAB-related Disorders** - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024312:1-21. **Detection Rate:** South Asian >99%.
**HADHA-related Disorders** - **Gene:** HADHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000182:1-20. **Detection Rate:** South Asian >99%.
**Hb Beta Chain-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:** South Asian >99%.
**Hereditary Fructose Intolerance** - **Gene:** ALDOB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000035:2-9. **Detection Rate:** South Asian >99%.
**Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** - **Gene:** HEXA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000520:1-14. **Detection Rate:** South Asian >99%.
**HMG-CoA Lyase Deficiency** - **Gene:** HMGCL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000191:1-9. **Detection Rate:** South Asian >99%.
**Holocarboxylase Synthetase Deficiency** - **Gene:** HLCS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000411:4-12. **Detection Rate:** South Asian >99%.
**Homocystinuria, CBS-related** - **Gene:** CBS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000071:3-17. **Detection Rate:** South Asian >99%.
**Hydrolethalus Syndrome** - **Gene:** HYLS1. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_145014:4. **Detection Rate:** South Asian >99%.
**Hypophosphatasia** - **Gene:** ALPL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000478:2-12. **Detection Rate:** South Asian >99%.
**Isovaleric Acidemia** - **Gene:** IVD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002225:1-12. **Detection Rate:** South Asian >99%.

**Joubert Syndrome 2** - **Gene:** TMEM216. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001173990:1-5. **Detection Rate:** South Asian >99%.
**Junctional Epidermolysis Bullosa, LAMA3-related** - **Gene:** LAMA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000227:1-38. **Detection Rate:** South Asian >99%.
**Junctional Epidermolysis Bullosa, LAMB3-related** - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228:2-23. **Detection Rate:** South Asian >99%.
**Junctional Epidermolysis Bullosa, LAMC2-related** - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** South Asian >99%.
**Krabbe Disease** - **Gene:** GALC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000153:1-17. **Detection Rate:** South Asian >99%.
**Leigh Syndrome, French-Canadian Type** - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133259:1-38. **Detection Rate:** South Asian >99%.
**Lipoid Congenital Adrenal Hyperplasia** - **Gene:** STAR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000349:1-7. **Detection Rate:** South Asian >99%.
**Lysosomal Acid Lipase Deficiency** - **Gene:** LIPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000235:2-10. **Detection Rate:** South Asian 98%.
**Maple Syrup Urine Disease Type Ia** - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000709:1-9. **Detection Rate:** South Asian >99%.
**Maple Syrup Urine Disease Type Ib** - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_183050:1-10. **Detection Rate:** South Asian >99%.
**Maple Syrup Urine Disease Type II** - **Gene:** DBT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001918:1-11. **Detection Rate:** South Asian 97%.
**Medium Chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000016:1-12. **Detection Rate:** South Asian >99%.
**Megalencephalic Leukoencephalopathy with Subcortical Cysts** - **Gene:** MLC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015166 2-12. **Detection Rate:** South Asian >99%.
**Metachromatic Leukodystrophy** - **Gene:** ARSA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000487:1-8. **Detection Rate:** South Asian >99%.
**Methylmalonic Acidemia, cblA Type** - **Gene:** MMAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_172250:2-7. **Detection Rate:** South Asian >99%.
**Methylmalonic Acidemia, cblB Type** - **Gene:** MMAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_052845:1-9. **Detection Rate:** South Asian >99%.
**Methylmalonic Acidemia, MMUT-related** - **Gene:** MMUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** South Asian >99%.
**Methylmalonic Aciduria and Homocystinuria, cblC Type** - **Gene:** MMACHC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015506:1-4. **Detection Rate:** South Asian >99%.
**MKS1-related Disorders** - **Gene:** MKS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017777:1-18. **Detection Rate:** South Asian >99%.
**Mucolipidosis III Gamma** - **Gene:** GNPTG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032520:1-11. **Detection Rate:** South Asian 98%.
**Mucolipidosis IV** - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_020533:1-14. **Detection Rate:** South Asian >99%.
**Mucopolysaccharidosis Type I** - **Gene:** IDUA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000203:1-14. **Detection Rate:** South Asian >99%.
**Mucopolysaccharidosis Type II** - **Gene:** IDS. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000202:1-9. **Detection Rate:** South Asian 89%.
**Mucopolysaccharidosis Type IIIA** - **Gene:** SGSH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000199:1-8. **Detection Rate:** South Asian >99%.
**Mucopolysaccharidosis Type IIIB** - **Gene:** NAGLU. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000263:1-6. **Detection Rate:** South Asian >99%.
**Mucopolysaccharidosis Type IIIC** - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_152419:1-18. **Detection Rate:** South Asian >99%.
**Muscular Dystrophy, LAMA2-related** - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-43,45-65. **Detection Rate:** South Asian 98%.
**MYO7A-related Disorders** - **Gene:** MYO7A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000260:2-49. **Detection Rate:** South Asian >99%.
**NEB-related Nemaline Myopathy** - **Gene:** NEB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** South Asian 92%.
**Nephrotic Syndrome, NPHS1-related** - **Gene:** NPHS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004646:1-29. **Detection Rate:** South Asian >99%.
**Nephrotic Syndrome, NPHS2-related** - **Gene:** NPHS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014625:1-8. **Detection Rate:** South Asian >99%.
**Neuronal Ceroid Lipofuscinosis, CLN6-related** - **Gene:** CLN6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017882:1-7. **Detection Rate:** South Asian >99%.
**Niemann-Pick Disease Type C1** - **Gene:** NPC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000271:1-25. **Detection Rate:** South Asian >99%.
**Niemann-Pick Disease Type C2** - **Gene:** NPC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006432:1-5. **Detection Rate:** South Asian >99%.
**Niemann-Pick Disease, SMPD1-related** - **Gene:** SMPD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000543:1-6. **Detection Rate:** South Asian >99%.
**Nijmegen Breakage Syndrome** - **Gene:** NBN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002485:1-16. **Detection Rate:** South Asian >99%.
**Ornithine Transcarbamylase Deficiency** - **Gene:** OTC. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000531:1-10. **Detection Rate:** South Asian 97%.
**PCCA-related Propionic Acidemia** - **Gene:** PCCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000282:1-24. **Detection Rate:** South Asian 95%.
**PCCB-related Propionic Acidemia** - **Gene:** PCCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000532:1-15. **Detection Rate:** South Asian >99%.
**PCDH15-related Disorders** - **Gene:** PCDH15. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_033056:2-33. **Detection Rate:** South Asian 93%.
**Pendred Syndrome** - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000441:2-21. **Detection Rate:** South Asian >99%.
**Peroxisome Biogenesis Disorder Type 1** - **Gene:** PEX1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000466:1-24. **Detection Rate:** South Asian >99%.
**Peroxisome Biogenesis Disorder Type 3** - **Gene:** PEX12. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000286:1-3. **Detection Rate:** South Asian >99%.
**Peroxisome Biogenesis Disorder Type 4** - **Gene:** PEX6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000287:1-17. **Detection Rate:** South Asian 97%.

**Peroxisome Biogenesis Disorder Type 5** - **Gene:** PEX2. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000318:4. **Detection Rate:** South Asian >99%.
**Peroxisome Biogenesis Disorder Type 6** - **Gene:** PEX10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153818:1-6. **Detection Rate:** South Asian >99%.
**Phenylalanine Hydroxylase Deficiency** - **Gene:** PAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000277:1-13. **Detection Rate:** South Asian >99%.
**POMGNT-related Disorders** - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017739:2-22. **Detection Rate:** South Asian 96%.
**Pompe Disease** - **Gene:** GAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000152:2-20. **Detection Rate:** South Asian >99%.
**PPT1-related Neuronal Ceroid Lipofuscinosis** - **Gene:** PPT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000310:1-9. **Detection Rate:** South Asian >99%.
**Primary Carnitine Deficiency** - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003060:1-10. **Detection Rate:** South Asian >99%.
**Primary Hyperoxaluria Type 1** - **Gene:** AGXT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000030:1-11. **Detection Rate:** South Asian >99%.
**Primary Hyperoxaluria Type 2** - **Gene:** GRHPR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012203:1-9. **Detection Rate:** South Asian >99%.
**Primary Hyperoxaluria Type 3** - **Gene:** HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** South Asian >99%.
**Pycnodysostosis** - **Gene:** CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000396:2-8. **Detection Rate:** South Asian >99%.
**Pyruvate Carboxylase Deficiency** - **Gene:** PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000920:3-22. **Detection Rate:** South Asian >99%.
**Rhizomelic Chondrodysplasia Punctata Type 1** - **Gene:** PEX7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000288:1-10. **Detection Rate:** South Asian >99%.
**RTEL1-related Disorders** - **Gene:** RTEL1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032957:2-35. **Detection Rate:** South Asian >99%.
**Sandhoff Disease** - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** South Asian 98%.
**Short-chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000017:1-10. **Detection Rate:** South Asian >99%.
**Sjogren-Larsson Syndrome** - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** South Asian 96%.
**SLC26A2-related Disorders** - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000112:2-3. **Detection Rate:** South Asian >99%.
**Smith-Lemli-Opitz Syndrome** - **Gene:** DHCR7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001360:3-9. **Detection Rate:** South Asian >99%.
**Spastic Paraplegia Type 15** - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015346:2-42. **Detection Rate:** South Asian >99%.
**Spinal Muscular Atrophy** - **Gene:** SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** South Asian 93%.
**Spondylothoracic Dysostosis** - **Gene:** MESP2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** South Asian >99%.
**TGM1-related Autosomal Recessive Congenital Ichthyosis** - **Gene:** TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000359 2-15. **Detection Rate:** South Asian >99%.
**TPP1-related Neuronal Ceroid Lipofuscinosis** - **Gene:** TPP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000391:1-13. **Detection Rate:** South Asian >99%.
**Tyrosine Hydroxylase Deficiency** - **Gene:** TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_199292:1-14. **Detection Rate:** South Asian >99%.
**Tyrosinemia Type I** - **Gene:** FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** South Asian >99%.
**Tyrosinemia Type II** - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** South Asian >99%.
**USH1C-related Disorders** - **Gene:** USH1C. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005709:1-21. **Detection Rate:** South Asian >99%.
**USH2A-related Disorders** - **Gene:** USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_206933:2-72. **Detection Rate:** South Asian 98%.
**Usher Syndrome Type 3** - **Gene:** CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** South Asian >99%.
**Very-long-chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADVL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000018:1-20. **Detection Rate:** South Asian >99%.
**Wilson Disease** - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** South Asian >99%.
**X-linked Adrenal Hypoplasia Congenita** - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** South Asian 97%.
**X-linked Adrenoleukodystrophy** - **Gene:** ABCD1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000033:1-6. **Detection Rate:** South Asian 77%.
**X-linked Alport Syndrome** - **Gene:** COL4A5. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000495:1-51. **Detection Rate:** South Asian 96%.
**X-linked Juvenile Retinoschisis** - **Gene:** RS1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000330:1-6. **Detection Rate:** South Asian 98%.
**X-linked Myotubular Myopathy** - **Gene:** MTM1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000252:2-15. **Detection Rate:** South Asian 96%.
**X-linked Severe Combined Immunodeficiency** - **Gene:** IL2RG. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000206:1-8. **Detection Rate:** South Asian >99%.
**Xeroderma Pigmentosum Group A** - **Gene:** XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** South Asian >99%.
**Xeroderma Pigmentosum Group C** - **Gene:** XPC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004628:1-16. **Detection Rate:** South Asian 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 | DONOR 14281<br>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 †<br>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 |
| **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 800,000 | 1 in 150,000 |
| **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 50,000 | < 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-sarcoglycanopathy** | 1 in 39,000 | < 1 in 1,000,000 |
| **Biotinidase Deficiency** | 1 in 17,000 | 1 in 990,000 |
| **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 Neuronal Ceroid Lipofuscinosis** | 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 |
| **COL4A3-related Alport Syndrome** | 1 in 5,800 | < 1 in 1,000,000 |
| **COL4A4-related Alport Syndrome** | 1 in 35,000 | < 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 530 | 1 in 130,000 |
| **Congenital Disorder of Glycosylation Type Ia** | 1 in 16,000 | < 1 in 1,000,000 |

| Disease | DONOR 14281 Residual Risk | Reproductive Risk |
|---|---|---|
| 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 |
| Costeff Optic Atrophy Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Cystic Fibrosis | 1 in 10,000 | < 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 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 80,000 |
| 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 50,000 | < 1 in 1,000,000 |
| 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 32,000 | < 1 in 1,000,000 |
| FKTN-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| 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 3,800 | < 1 in 1,000,000 |
| Gaucher Disease | 1 in 310 | 1 in 150,000 |
| GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness | p.G12Vfs*2 heterozygote † | 1 in 160 |
| 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 |
| Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) | 1 in 2,400 | 1 in 230,000 |
| Hereditary Fructose Intolerance | 1 in 7,900 | < 1 in 1,000,000 |
| Hexosaminidase A Deficiency (Including Tay-Sachs Disease) | 1 in 30,000 | < 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 |
| 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 34,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type Ia | 1 in 9,400 | < 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 4,100 | < 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 |

| Disease | DONOR 14281 Residual Risk | Reproductive Risk |
|---|---|---|
| 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 10,000 | < 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 1,000,000 | 1 in 300,000 |
| 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 140,000 |
| 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 8,600 | < 1 in 1,000,000 |
| POMGNT-related Disorders | < 1 in 12,000 | < 1 in 1,000,000 |
| Pompe Disease | 1 in 10,000 | < 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 16,000 | < 1 in 1,000,000 |
| Smith-Lemli-Opitz Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Spastic Paraplegia Type 15 | < 1 in 50,000 | < 1 in 1,000,000 |
| Spinal Muscular Atrophy | SMN1: 1 copy † | 1 in 200 |
| 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 |
| 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 |

| Disease | DONOR 14281 Residual Risk | Reproductive Risk |
|---|---|---|
| **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 90,000 | 1 in 42,000 |
| **X-linked Alport Syndrome** | Not calculated | Not calculated |
| **X-linked Juvenile Retinoschisis** | < 1 in 1,000,000 | 1 in 50,000 |
| **X-linked Myotubular Myopathy** | Not calculated | Not calculated |
| **X-linked Severe Combined Immunodeficiency** | < 1 in 1,000,000 | 1 in 200,000 |
| **Xeroderma Pigmentosum Group A** | 1 in 28,000 | < 1 in 1,000,000 |
| **Xeroderma Pigmentosum Group C** | 1 in 7,300 | < 1 in 1,000,000 |