myriad WOMEN'S HEALTH

RESULTS RECIPIENT
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Report Date: 06/30/2020

MALE
DONOR 12605
DOB:
Ethnicity: Mixed or Other Caucasian
Sample Type: EDTA Blood
Date of Collection: 06/23/2020
Date Received: 06/24/2020
Date Tested: 06/29/2020
Barcode: 11004512623018
Accession ID: CSLFNHX3LKKJ6ZQ
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 12605	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel **(175 conditions tested)**	N/A
POSITIVE: CARRIER Primary Hyperoxaluria Type 3 **Reproductive Risk: 1 in 520** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_138413.3(HOGA1):c.569C>T(P190L) 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 Dihydrolipoamide Dehydrogenase Deficiency **Reproductive Risk: 1 in 2,000** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000108.3(DLD):c.803_804delAG heterozygote †	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".
POSITIVE: CARRIER Alpha Thalassemia, HBA1/HBA2-related **Reproductive Risk: Not Calculated** **Inheritance: Autosomal Recessive**	**CARRIER*** -alpha3.7 [chr16:g.(?_226678)_(227520_?)del] heterozygote 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".

†Likely to have a negative impact on gene function.
*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 11.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner, as both parents must be carriers before a child is at high risk of developing the disease.
- Genetic counseling is recommended and patients may wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

POSITIVE: CARRIER

Primary Hyperoxaluria Type 3

Reproductive risk: 1 in 520
Risk before testing: 1 in 67,000

Gene: HOGA1 | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 12605	No partner tested
Result	✚ Carrier	N/A
Variant(s)	NM_138413.3(HOGA1):c.569C>T(P190L) heterozygote †	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of primary hyperoxaluria type 3. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_138413:1-7.	N/A

†Likely to have a negative impact on gene function.

What is Primary Hyperoxaluria Type 3?

Primary hyperoxaluria type 3 (PH3) is an inherited disease where lack of a particular liver enzyme causes the body to accumulate excess amounts of a substance called oxalate. Oxalate leads to a buildup of insoluble calcium salts in the kidneys. There are three types of primary hyperoxaluria. Unlike types 1 and 2, which can impact other organs, buildup of oxalate in PH3 has not been seen outside of the kidneys.

People with PH3 are at increased risk for developing kidney stones. Symptoms can develop anytime from infancy to adulthood. Approximately 50% of affected individuals developed kidney stones by age 5, but many experience a decrease by adulthood. Some people with the disease do not have symptoms until adulthood. Kidney function can be impacted by frequent kidney stones; however, kidney failure has rarely been reported in individuals with PH3.

How common is Primary Hyperoxaluria Type 3?

Approximately 1 in 165,000 individuals worldwide are expected to be affected with PH3. Reports of higher and lower carrier frequencies have been reported in the Ashkenazi Jewish and African American populations, respectively.

How is Primary Hyperoxaluria Type 3 treated?

People with the condition should drink plenty of water. Intravenous (IV) fluids may be necessary during periods of illness or times of limited fluid intake. A physician may prescribe medication or vitamins to help lower oxalate levels and inhibit the formation of kidney stones. Dietary restriction of foods high in oxalate may be beneficial. Unlike with other type of primary hyperoxaluria, individuals with PH3 rarely require dialysis or kidney/liver transplantation.

What is the prognosis for a person with Primary Hyperoxaluria Type 3?

Individuals with PH3 often have formation of multiple kidney stones, which can be managed by increased fluid intake and supplements. Kidney stone formation in many individuals with PH3 often decrease as they reach adulthood. Thus far, only one individual with PH3 has been reported to have progressed to kidney failure, and transplant is not necessary in most individuals with PH3.

POSITIVE: CARRIER

Dihydrolipoamide Dehydrogenase Deficiency

Reproductive risk: 1 in 2,000
Risk before testing: < 1 in 1,000,000

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

Patient	DONOR 12605	No partner tested
Result	✚ Carrier	N/A
Variant(s)	NM_000108.3(DLD):c.803_804delAG heterozygote †	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of dihydrolipoamide dehydrogenase deficiency. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000108:1-14.	N/A

†Likely to have a negative impact on gene function.

What is Dihydrolipoamide Dehydrogenase Deficiency (E3 Deficiency)?

Dihydrolipoamide Dehydrogenase Deficiency also known as E3 deficiency, caused by mutations in the *DLD* gene, is an inherited condition resulting in the deficiency of the enzyme dihydrolipoamide dehydrogenase, which disrupts multiple enzyme complexes that help break down substances in cells. This condition can cause metabolic abnormalities, neurological damage, poor muscle tone, developmental delay, and movement problems. Infants with E3 deficiency often appear normal until the age of eight weeks to six months, when they develop a severe buildup of lactic acid in the body (lactic acidosis) that causes vomiting, abdominal pain, and rapid breathing. If untreated, this condition can be fatal. Infants and children with the disease can have developmental delay and a progressive breakdown of their nervous system. They often have poor muscle tone (hypotonia) and abnormal movements. The disease is also called maple syrup urine disease type 3, due to the characteristic maple-syrup-like smell of their urine.

How Common Is E3 Deficiency?

The prevalence of E3 deficiency in the general population is unknown. The majority of known cases come from families of Ashkenazi Jewish background, where the prevalence is 1 in 35,000 to 1 in 48,000.

How Is E3 Deficiency Treated?

There is no established treatment for E3 deficiency. Combinations of diet, vitamins, and supplements have been tried without much success.

What Is the Prognosis for an Individual with E3 Deficiency?

While the number of known cases does not allow for a well-established prognosis, it is thought that most individuals with E3 deficiency will die during childhood.

POSITIVE: CARRIER

Alpha Thalassemia, HBA1/HBA2-related

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

Patient	DONOR 12605	No partner tested
Result	✚ Carrier	N/A
Variant(s)	-alpha3.7 [chr16:g.(?_226678)_(227520_?)del] heterozygote	N/A
Methodology	Analysis of homologous regions	N/A
Interpretation	This individual is a carrier of alpha thalassemia. Carriers do not experience symptoms, but may have hematologic abnormalities. -alpha3.7 is classified as an alpha+ mutation. Based on this result, the patient's alpha globin status is -a/aa (carrier), where "-" indicates a deleted or nonfunctional alpha globin gene.	N/A
Detection rate	Unknown due to rarity of disease	N/A
Variants tested	-(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, anti3.7, anti4.2, del HS-40.	N/A

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, caused by harmful genetic changes (mutations) in the *HBA1* and *HBA2* genes, 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 a disruption in the normal production of alpha globin. The *HBA1* and *HBA2* genes work together to make a functioning alpha globin protein.

There are various forms of alpha thalassemia, which have a variety of symptoms. The type of alpha thalassemia an individual has depends on the combination of mutations they inherit in the *HBA1* and *HBA2* genes. 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*). In order to have symptoms of alpha thalassemia, an individual must have mutations in three or four of their gene copies. Carriers generally have either two or three functional alpha globin genes and do not have any symptoms of the conditions.

SILENT CARRIER: THREE FUNCTIONAL ALPHA GLOBIN GENES

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): TWO FUNCTIONAL ALPHA GLOBIN GENES

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. There are reports of individuals with two mutations in *HBA1* and/or *HBA2* who have a diagnosis of 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: ONE FUNCTIONAL ALPHA GLOBIN GENE

Hemoglobin H (HbH) disease is typically the result of mutations in three of the four alpha globin genes. This form is highly variable. Many individuals with HbH do not have any symptoms, while some may have mild to moderate anemia. Other symptoms of HbH include yellowing of the skin or eyes (jaundice), enlargement of the spleen, bone deformities, fatigue, and other minor complications.

HEMOGLOBIN BART SYNDROME: ZERO FUNCTIONAL ALPHA GLOBIN GENES

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. Patients who do not receive any treatment will have poor outcomes, and many will not live past five years of age. However, 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 12605 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions (DTS v3.1).

Sequencing with copy number analysis

High-throughput sequencing and read depth-based copy number analysis are used to analyze the listed exons, as well as selected intergenic and intronic regions, of the genes in the Conditions Tested section of the report. The region of interest (ROI) of the test comprises these regions, in addition to the 20 intronic bases flanking each exon. 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 high coverage and the sequences are compared to standards and references of normal variation (Genome Reference Consortium Human Build 37 (GRCh37)/hg19). More than 99% of all bases in the ROI are sequenced at greater than the minimum read depth. Mutations may not be detected in areas of lower sequence coverage. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes may be addressed by a different method. *CFTR* and *DMD* testing includes analysis for both large (exon-level) deletions and duplications with an average sensitivity of 99%, while other genes are only analyzed for large deletions with a sensitivity of >75%. However, the sensitivity may be higher for selected founder deletions. The breakpoints of copy number variants and exons affected are estimated from probe positions. Only exons known to be included in the copy number variant are provided in the name. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, two large upstream deletions which overlap *GJB6* and affect the expression of *GJB2*, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854), are also analyzed. Mosaicism or somatic variants present at low levels may not be detected. If detected, these may not be reported.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect. Detection rates only account for analytical sensitivity and 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 mutations.

All variants that are a recognized cause of the disease will be reported. In addition, variants that have not previously been established as a recognized cause of disease may be identified. In these cases, only variants classified as "likely" pathogenic are reported. Likely pathogenic variants are described elsewhere in the report as "likely to have a negative impact on gene function". Likely pathogenic variants are evaluated and classified by assessing the nature of the variant and reviewing reports of allele frequencies in cases and controls, functional studies, variant annotation and effect prediction, and segregation studies. Exon level duplications are assumed to be in tandem and are classified according to their predicted effect on the reading frame. Benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Curation summaries of reported variants are available upon request.

Spinal muscular atrophy

Targeted copy number analysis is used to determine the copy number of exon 7 of the *SMN1* gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of *SMN1* are carriers with two *SMN1* genes on one chromosome and a *SMN1* deletion on the other chromosome. This is more likely in individuals who have 2 copies of the *SMN1* gene and are positive for the g.27134T>G SNP, 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 2 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 mutations in certain genes that have homology to other regions. The precise breakpoints of large deletions in these genes cannot be determined, but are estimated from copy number analysis. High numbers of pseudogene copies may interfere with this analysis.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a loss of function mutation may not actually be a carrier of 21-hydroxylase-deficient congenital adrenal hyperplasia (CAH). Because the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are only based on published incidences 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 of overall chances for 21-hydroxylase-deficient CAH, especially in the aforementioned populations, as they do not account for non-classic CAH. If *HBA1*/*HBA2* are tested, some individuals with four alpha globin genes may be carriers, with three genes on one chromosome and a deletion on the other chromosome. This and similar, but rare, carrier states, where complementary changes exist in both the gene and a pseudogene, may not be detected by the assay.

Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these may not be reported. f 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. 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. Residual and reproductive risks provided assume a normal karyotype. Risks for individuals with abnormal karyotypes may be different. The test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet.Gynecol. 2007;109:229-37*).

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**.

Resources

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

Patients can share their reports via research registries such as Genome Connect, an online research registry working to build the knowledge base about genetics and health. 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

Jack Ji, PhD, FACMG

Report content approved by Jack Ji, PhD, FACMG on Jun 30, 2020

Conditions Tested

11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** Mixed or Other Caucasian 94%.

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

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

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

Alpha Thalassemia, HBA1/HBA2-related - **Genes:** HBA1, HBA2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, anti3.7, anti4.2, del HS-40. **Detection Rate:** Unknown due to rarity of disease.

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

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

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

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

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

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

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

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

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

Ataxia-telangiectasia - **Gene:** ATM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000051:2-63. **Detection Rate:** Mixed or Other Caucasian 98%.

ATP7A-related Disorders - **Gene:** ATP7A. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000052:2-23. **Detection Rate:** Mixed or Other Caucasian 96%.

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

Autosomal Recessive Osteopetrosis Type 1 - **Gene:** TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006019:2-20. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

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

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

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

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

Calpainopathy - **Gene:** CAPN3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000070:1-24. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

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

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

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

CLN6-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017882:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.
CLN8-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_018941:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Cohen Syndrome - **Gene:** VPS13B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017890:2-62. **Detection Rate:** Mixed or Other Caucasian 97%.
COL4A3-related Alport Syndrome - **Gene:** COL4A3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000091:1-52. **Detection Rate:** Mixed or Other Caucasian 97%.
COL4A4-related Alport Syndrome - **Gene:** COL4A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000092:2-48. **Detection Rate:** Mixed or Other Caucasian 98%.
Combined Pituitary Hormone Deficiency, PROP1-related - **Gene:** PROP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006261:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Congenital Adrenal Hyperplasia, CYP21A2-related - **Gene:** CYP21A2. Autosomal Recessive. Analysis of homologous regions. **Variants (12):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, c.293-13C>G. **Detection Rate:** Mixed or Other Caucasian 96%.
Congenital Disorder of Glycosylation Type Ia - **Gene:** PMM2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000303:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Congenital Disorder of Glycosylation Type Ic - **Gene:** ALG6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_013339:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.
Congenital Disorder of Glycosylation, MPI-related - **Gene:** MPI. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002435:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Costeff Optic Atrophy Syndrome - **Gene:** OPA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_025136:1-2. **Detection Rate:** Mixed or Other Caucasian >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:** Mixed or Other Caucasian >99%.
Cystinosis - **Gene:** CTNS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004937:3-12. **Detection Rate:** Mixed or Other Caucasian >99%.
D-bifunctional Protein Deficiency - **Gene:** HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000414:1-24. **Detection Rate:** Mixed or Other Caucasian 98%.
Delta-sarcoglycanopathy - **Gene:** SGCD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000337:2-9. **Detection Rate:** Mixed or Other Caucasian 99%.
Dihydrolipoamide Dehydrogenase Deficiency - **Gene:** DLD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000108:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Dysferlinopathy - **Gene:** DYSF. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003494:1-55. **Detection Rate:** Mixed or Other Caucasian 98%.
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - **Gene:** DMD. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_004006:1-79. **Detection Rate:** Mixed or Other Caucasian >99%.
ERCC6-related Disorders - **Gene:** ERCC6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000124:2-21. **Detection Rate:** Mixed or Other Caucasian 99%.
ERCC8-related Disorders - **Gene:** ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000082:1-12. **Detection Rate:** Mixed or Other Caucasian 95%.
EVC-related Ellis-van Creveld Syndrome - **Gene:** EVC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153717:1-21. **Detection Rate:** Mixed or Other Caucasian 96%.
EVC2-related Ellis-van Creveld Syndrome - **Gene:** EVC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_147127:1-22. **Detection Rate:** Mixed or Other Caucasian >99%.
Fabry Disease - **Gene:** GLA. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000169:1-7. **Detection Rate:** Mixed or Other Caucasian 98%.
Familial Dysautonomia - **Gene:** IKBKAP. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003640:2-37. **Detection Rate:** Mixed or Other Caucasian >99%.
Familial Mediterranean Fever - **Gene:** MEFV. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000243:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Fanconi Anemia Complementation Group A - **Gene:** FANCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000135:1-43. **Detection Rate:** Mixed or Other Caucasian 92%.
Fanconi Anemia, FANCC-related - **Gene:** FANCC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000136:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.
FKRP-related Disorders - **Gene:** FKRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_024301:4. **Detection Rate:** Mixed or Other Caucasian >99%.
FKTN-related Disorders - **Gene:** FKTN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001079802:3-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Free Sialic Acid Storage Disorders - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012434:1-11. **Detection Rate:** Mixed or Other Caucasian 98%.
Galactokinase Deficiency - **Gene:** GALK1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000154:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Galactosemia - **Gene:** GALT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000155:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Gamma-sarcoglycanopathy - **Gene:** SGCG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000231:2-8. **Detection Rate:** Mixed or Other Caucasian 88%.
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:** Mixed or Other Caucasian 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:** Mixed or Other Caucasian >99%.
GLB1-related Disorders - **Gene:** GLB1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000404:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.
GLDC-related Glycine Encephalopathy - **Gene:** GLDC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000170:1-25. **Detection Rate:** Mixed or Other Caucasian 94%.
Glutaric Acidemia, GCDH-related - **Gene:** GCDH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000159:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Glycogen Storage Disease Type Ia - **Gene:** G6PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000151:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Glycogen Storage Disease Type Ib - **Gene:** SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001164277 3-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Glycogen Storage Disease Type III - **Gene:** AGL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000642:2-34. **Detection Rate:** Mixed or Other Caucasian >99%.

GNE Myopathy - **Gene:** GNE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001128227:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.
GNPTAB-related Disorders - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024312:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.
HADHA-related Disorders - **Gene:** HADHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000182:1-20. **Detection Rate:** Mixed or Other Caucasian >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:** Mixed or Other Caucasian >99%.
Hereditary Fructose Intolerance - **Gene:** ALDOB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000035:2-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228 2-23. **Detection Rate:** Mixed or Other Caucasian >99%.
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000520:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
HMG-CoA Lyase Deficiency - **Gene:** HMGCL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000191:1-9. **Detection Rate:** Mixed or Other Caucasian 98%.
Holocarboxylase Synthetase Deficiency - **Gene:** HLCS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000411:4-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Homocystinuria, CBS-related - **Gene:** CBS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000071:3-17. **Detection Rate:** Mixed or Other Caucasian >99%.
Hydrolethalus Syndrome - **Gene:** HYLS1. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_145014:4. **Detection Rate:** Mixed or Other Caucasian >99%.
Hypophosphatasia - **Gene:** ALPL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000478:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Isovaleric Acidemia - **Gene:** IVD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002225:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Joubert Syndrome 2 - **Gene:** TMEM216. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001173990:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Junctional Epidermolysis Bullosa, LAMA3-related - **Gene:** LAMA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000227:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.
Junctional Epidermolysis Bullosa, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.
KCNJ11-related Familial Hyperinsulinism - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000525:1. **Detection Rate:** Mixed or Other Caucasian >99%.
Krabbe Disease - **Gene:** GALC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000153:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.
LAMA2-related Muscular Dystrophy - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-65. **Detection Rate:** Mixed or Other Caucasian >99%.
Leigh Syndrome, French-Canadian Type - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133259:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.
Lipoid Congenital Adrenal Hyperplasia - **Gene:** STAR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000349:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Lysosomal Acid Lipase Deficiency - **Gene:** LIPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000235:2-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Maple Syrup Urine Disease Type Ia - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000709:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Maple Syrup Urine Disease Type Ib - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_183050:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Maple Syrup Urine Disease Type II - **Gene:** DBT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001918:1-11. **Detection Rate:** Mixed or Other Caucasian 96%.
Medium Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000016:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Megalencephalic Leukoencephalopathy with Subcortical Cysts - **Gene:** MLC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015166 2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Metachromatic Leukodystrophy - **Gene:** ARSA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000487:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Methylmalonic Acidemia, cblA Type - **Gene:** MMAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_172250:2-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Methylmalonic Acidemia, cblB Type - **Gene:** MMAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_052845:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Methylmalonic Aciduria and Homocystinuria, cblC Type - **Gene:** MMACHC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015506:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.
MKS1-related Disorders - **Gene:** MKS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017777:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.
Mucolipidosis III Gamma - **Gene:** GNPTG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032520:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Mucolipidosis IV - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_020533:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Mucopolysaccharidosis Type I - **Gene:** IDUA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000203:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Mucopolysaccharidosis Type II - **Gene:** IDS. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000202:1-9. **Detection Rate:** Mixed or Other Caucasian 88%.
Mucopolysaccharidosis Type IIIA - **Gene:** SGSH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000199:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Mucopolysaccharidosis Type IIIB - **Gene:** NAGLU. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000263:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Mucopolysaccharidosis Type IIIC - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_152419:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.
MUT-related Methylmalonic Acidemia - **Gene:** MUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** Mixed or Other Caucasian >99%.
MYO7A-related Disorders - **Gene:** MYO7A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000260:2-49. **Detection Rate:** Mixed or Other Caucasian >99%.
NEB-related Nemaline Myopathy - **Gene:** NEB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** Mixed or Other Caucasian 92%.

Nephrotic Syndrome, NPHS1-related - **Gene:** NPHS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004646:1-29. **Detection Rate:** Mixed or Other Caucasian >99%.
Nephrotic Syndrome, NPHS2-related - **Gene:** NPHS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014625:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Niemann-Pick Disease Type C1 - **Gene:** NPC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000271:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.
Niemann-Pick Disease Type C2 - **Gene:** NPC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006432:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Niemann-Pick Disease, SMPD1-related - **Gene:** SMPD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000543:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Nijmegen Breakage Syndrome - **Gene:** NBN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002485:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.
Ornithine Transcarbamylase Deficiency - **Gene:** OTC. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000531:1-10. **Detection Rate:** Mixed or Other Caucasian 97%.
PCCA-related Propionic Acidemia - **Gene:** PCCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000282:1-24. **Detection Rate:** Mixed or Other Caucasian 95%.
PCCB-related Propionic Acidemia - **Gene:** PCCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000532:1-15. **Detection Rate:** Mixed or Other Caucasian >99%.
PCDH15-related Disorders - **Gene:** PCDH15. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_033056:2-33. **Detection Rate:** Mixed or Other Caucasian 93%.
Pendred Syndrome - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000441:2-21. **Detection Rate:** Mixed or Other Caucasian >99%.
Peroxisome Biogenesis Disorder Type 1 - **Gene:** PEX1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000466:1-24. **Detection Rate:** Mixed or Other Caucasian >99%.
Peroxisome Biogenesis Disorder Type 3 - **Gene:** PEX12. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000286:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Peroxisome Biogenesis Disorder Type 4 - **Gene:** PEX6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000287:1-17. **Detection Rate:** Mixed or Other Caucasian 97%.
Peroxisome Biogenesis Disorder Type 5 - **Gene:** PEX2. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000318:4. **Detection Rate:** Mixed or Other Caucasian >99%.
Peroxisome Biogenesis Disorder Type 6 - **Gene:** PEX10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153818:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Phenylalanine Hydroxylase Deficiency - **Gene:** PAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000277:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.
POMGNT-related Disorders - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017739:2-22. **Detection Rate:** Mixed or Other Caucasian 96%.
Pompe Disease - **Gene:** GAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000152:2-20. **Detection Rate:** Mixed or Other Caucasian 98%.
PPT1-related Neuronal Ceroid Lipofuscinosis - **Gene:** PPT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000310:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Primary Carnitine Deficiency - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003060:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Primary Hyperoxaluria Type 1 - **Gene:** AGXT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000030:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012203:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Primary Hyperoxaluria Type 3 - **Gene:** HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Pycnodysostosis - **Gene:** CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000396:2-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Pyruvate Carboxylase Deficiency - **Gene:** PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000920:3-22. **Detection Rate:** Mixed or Other Caucasian >99%.
Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000288:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
RTEL1-related Disorders - **Gene:** RTEL1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032957:2-35. **Detection Rate:** Mixed or Other Caucasian >99%.
Sandhoff Disease - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Short-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000017:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** Mixed or Other Caucasian 96%.
SLC26A2-related Disorders - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000112:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Smith-Lemli-Opitz Syndrome - **Gene:** DHCR7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001360:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Spastic Paraplegia Type 15 - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015346:2-42. **Detection Rate:** Mixed or Other Caucasian >99%.
Spinal Muscular Atrophy - **Gene:** SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** Mixed or Other Caucasian 95%.
Spondylothoracic Dysostosis - **Gene:** MESP2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.
TGM1-related Autosomal Recessive Congenital Ichthyosis - **Gene:** TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000359 2-15. **Detection Rate:** Mixed or Other Caucasian >99%.
TPP1-related Neuronal Ceroid Lipofuscinosis - **Gene:** TPP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000391:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.
Tyrosine Hydroxylase Deficiency - **Gene:** TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_199292:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Tyrosinemia Type I - **Gene:** FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Tyrosinemia Type II - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
USH1C-related Disorders - **Gene:** USH1C. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005709:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.

USH2A-related Disorders - **Gene:** USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_206933:2-72. **Detection Rate:** Mixed or Other Caucasian 94%.
Usher Syndrome Type 3 - **Gene:** CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Very-long-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADVL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000018:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.
Wilson Disease - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.
X-linked Adrenoleukodystrophy - **Gene:** ABCD1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000033:1-6. **Detection Rate:** Mixed or Other Caucasian 77%.
X-linked Alport Syndrome - **Gene:** COL4A5. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000495:1-51. **Detection Rate:** Mixed or Other Caucasian 95%.
X-linked Congenital Adrenal Hypoplasia - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** Mixed or Other Caucasian 99%.
X-linked Juvenile Retinoschisis - **Gene:** RS1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000330:1-6. **Detection Rate:** Mixed or Other Caucasian 98%.
X-linked Myotubular Myopathy - **Gene:** MTM1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000252:2-15. **Detection Rate:** Mixed or Other Caucasian 98%.
X-linked Severe Combined Immunodeficiency - **Gene:** IL2RG. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000206:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Xeroderma Pigmentosum Group A - **Gene:** XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Xeroderma Pigmentosum Group C - **Gene:** XPC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004628:1-16. **Detection Rate:** Mixed or Other Caucasian 97%.

Risk Calculations

Below are the risk calculations for all conditions tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's 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 after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

†Indicates a positive result. See the full clinical report for interpretation and details.

Disease	DONOR 12605 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,800	< 1 in 1,000,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Familial Hyperinsulinism	1 in 17,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	-alpha3.7 [chr16:g.(?_226678)_(227520_?)del] heterozygote † Alpha globin status: -a/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
AMT-related Glycine Encephalopathy	1 in 22,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	< 1 in 17,000	< 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 11,000	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	< 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 32,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 650,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 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 25,000	< 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 14,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 8,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN6-related Neuronal Ceroid Lipofuscinosis	1 in 43,000	< 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 6,200	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 12,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	1 in 1,300	1 in 280,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 12605 Residual Risk	Reproductive Risk
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 3,000	1 in 360,000
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 40,000	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	NM_000108.3(DLD):c.803_804delAG heterozygote †	1 in 2,000
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 26,000	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 9,900	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,500	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	< 1 in 50,000	< 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 Mediterranean Fever	< 1 in 50,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	< 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 10,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	< 1 in 1,000,000
Gaucher Disease	1 in 260	1 in 110,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,500	1 in 260,000
GLB1-related Disorders	1 in 19,000	< 1 in 1,000,000
GLDC-related Glycine Encephalopathy	1 in 2,800	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 18,000	< 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 23,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 32,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 20,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 3,100	1 in 390,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 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 33,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 9,400	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 27,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 32,000	< 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, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
KCNJ11-related Familial Hyperinsulinism	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	1 in 34,000	< 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 18,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 42,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 13,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 4,400	1 in 790,000
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

Disease	DONOR 12605 Residual Risk	Reproductive Risk
Methylmalonic Acidemia, cblB Type	1 in 48,000	< 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 50,000	< 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 600,000	1 in 150,000
Mucopolysaccharidosis Type IIIA	1 in 12,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 25,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 37,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	< 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
Niemann-Pick Disease Type C1	1 in 19,000	< 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 15,000	< 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 8,200	< 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 4,800	1 in 940,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 4,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 11,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 17,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	NM_138413.3(HOGA1) c.569C>T(P190L) heterozygote †	1 in 520
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 32,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 11,000	< 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 9,400	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 35,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 2,200	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 18,000	< 1 in 1,000,000
Wilson Disease	1 in 6,500	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000

Disease	DONOR 12605 Residual Risk	Reproductive Risk
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Congenital Adrenal Hypoplasia	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 40,000
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 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000