



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
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 Seattle, WA 98105
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 NPI: 1306838271
 Report Date: 12/10/2021

MALE
DONOR 12821
 DOB:
 Ethnicity: African or African American
 Sample Type: EDTA Blood
 Date of Collection: 11/30/2021
 Date Received: 12/02/2021
 Date Tested: 12/10/2021
 Barcode: 11004513015678
 Accession ID: CSLFG6RH2RKQ2EN
 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 12821	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested)	N/A
POSITIVE: CARRIER Medium Chain Acyl-CoA Dehydrogenase Deficiency Reproductive Risk: 1 in 240 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_000016.4(ACADM):c.985A>G(K329E, aka K304E) 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 Congenital Adrenal Hyperplasia, CYP21A2-related Reproductive Risk: 1 in 550 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_000500.7(CYP21A2):c.844G>T(V282L) 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 USH2A-related Disorders Reproductive Risk: 1 in 580 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_206933.2(USH2A):c.2299delG(E767Sfs*21) 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".

*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 12.

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



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also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

POSITIVE: CARRIER

Medium Chain Acyl-CoA Dehydrogenase Deficiency

Reproductive risk: 1 in 240
 Risk before testing: 1 in 15,000

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

Patient	DONOR 12821	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000016.4(ACADM):c.985A>G(K329E, aka K304E) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of medium chain acyl-CoA dehydrogenase deficiency. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000016:1-12.	N/A

What Is Medium-Chain Acyl-CoA Dehydrogenase Deficiency?

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is an inherited metabolic disorder in which fats cannot be broken down into energy to fuel the body. It is part of a group of disorders called fatty-acid oxidation defects and is caused by mutations in the *ACADM* gene. Mutations in this gene lead to low levels of the medium-chain acyl-CoA dehydrogenase enzyme resulting in a buildup of partially metabolized fatty acids that can accumulate in body tissues and cause damage to the brain, liver, and other organs.

For most individuals with MCAD deficiency, symptoms first appear in infancy or early childhood and can be triggered by long periods without eating (fasting) or by illness. Symptoms of metabolic crises include vomiting, lack of energy, low blood sugar, and an enlarged liver. Without treatment, metabolic crises can quickly develop into life-threatening problems including seizures, breathing problems, brain damage, coma, and death. A small percentage of sudden infant death syndrome (SIDS) is likely due to undiagnosed MCAD deficiency. In some cases, symptoms may not appear until late childhood or in adulthood and can be milder.

How Common Is Medium-Chain Acyl-CoA Dehydrogenase Deficiency?

MCAD deficiency is most common in Caucasians from Northern Europe. In the United States, the disease prevalence is 1 in 17,000 individuals. Affected Americans are often of Northern European ancestry. The disease is rare among Hispanics, African Americans, Asians, and Native Americans in the United States.

Studies have found high rates of MCAD deficiency in Northern Germany (1 in 4,900 individuals) and Southern Germany (1 in 8,500 individuals).

How Is Medium-Chain Acyl-CoA Dehydrogenase Deficiency Treated?

The key treatment for individuals with MCAD deficiency is to avoid fasting. Infants must be frequently fed a formula low in fat but high in carbohydrates. For children and adults, consuming cornstarch can also provide a sustained release of energy and allow for longer gaps between meals. If an individual is unable to eat or drink food on their own, it may be necessary to give them glucose by intravenous fluids.

What Is the Prognosis for an Individual with Medium-Chain Acyl-CoA Dehydrogenase Deficiency?

Early diagnosis and dietary management are crucial for the best outcome. If dietary management starts early (especially before the first crisis) and is consistent, individuals with MCAD deficiency have a good prognosis with normal or near-normal lifespan. In undiagnosed and untreated cases of MCAD deficiency, metabolic crises can quickly cause irreversible damage or even lead to death.

Additional Considerations for Carriers

Carriers of fatty-acid oxidation defects, including MCAD deficiency, do not typically show symptoms of the disease. However, there is an increased risk of serious pregnancy complications, particularly in the third trimester, in women carrying a fetus affected with MCAD deficiency. These complications can include HELLP syndrome and acute fatty liver of pregnancy. A woman whose pregnancy may be affected by a fatty-acid oxidation defect, such as MCAD deficiency, should speak with her physician for recommendations and may benefit from consultation with a high-risk physician.

POSITIVE: CARRIER

Congenital Adrenal Hyperplasia, CYP21A2-related

Reproductive risk: 1 in 550
 Risk before testing: 1 in 75,000

Gene: CYP21A2 | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 12821	No partner tested
Result	Carrier	N/A
Variant(s)	NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote	N/A
Methodology	Analysis of homologous regions (v3.2)	N/A
Interpretation	This individual is a carrier of congenital adrenal hyperplasia, CYP21A2-related. Carriers generally do not experience symptoms. NM_000500.7(CYP21A2):c.844G>T(V282L) is a non-classic congenital adrenal hyperplasia, CYP21A2-related mutation.	N/A
Detection rate	92%	N/A
Variants tested	CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G.	N/A

What Is Congenital Adrenal Hyperplasia, CYP21A2-Related?

Congenital adrenal hyperplasia (CAH) refers to a group of genetic disorders that affect the body's adrenal glands. The adrenal glands regulate essential functions in the body, including the production of several important hormones. CAH occurs when the adrenal glands are unable to produce these hormones properly, resulting in a hormone imbalance. CAH, CYP21A2-related is caused by mutations in the *CYP21A2* gene. The *CYP21A2* gene produces the 21-hydroxylase enzyme. Another name for this disorder is 21-hydroxylase-deficient CAH (21-OHD CAH).

When the 21-hydroxylase enzyme is missing or present at low levels, the adrenal glands are unable to produce two critical hormones, cortisol and aldosterone. The body responds to this deficiency by producing an excess of male sex hormones, called androgens. Collectively, the excess androgen production and hormone deficiencies can lead to a variety of medical problems, which vary in severity depending on the form of CAH. CAH associated with *CYP21A2* (21-OHD CAH) has two major forms: classic and non-classic.

CLASSIC FORM

The most severe form referred to as classic 21-OHD CAH, can be further divided into two different subtypes: salt wasting and simple virilizing (non-salt wasting) types. The classic salt-wasting type is associated with near-to-complete deficiency of the 21-hydroxylase enzyme, resulting in the complete inability to produce the hormones cortisol and aldosterone. In this type, the body cannot retain enough sodium (salt) and when too much salt is lost in the urine, it may lead to dehydration, vomiting, diarrhea, poor growth, heart-rhythm abnormalities (arrhythmias), and shock (salt wasting). If not properly treated, salt wasting can lead to death in some cases.

Additionally, female newborns often have external genitals that do not clearly appear either male or female (ambiguous genitalia), whereas male newborns may present with enlarged genitals. Signs of early puberty and the exaggerated development of male characteristics (virilization) occur in both males and females with CAH. These symptoms may include: rapid growth and development

in early childhood, but shorter-than-average height in adulthood, abnormal menstruation cycles for females, excess facial hair for females, early facial-hair growth for males, severe acne, and infertility in both men and women. Male characteristics such as muscle bulk and a deep voice can occur in females and in boys (masculinization).

The simple virilizing type of CAH is associated with partial 21-hydroxylase deficiency. Unlike the salt-wasting type, individuals with this condition typically do not experience severe and life-threatening sodium-deficiency symptoms as newborns. However, the majority of female newborns with this type will have ambiguous genitalia, and both male and female children may show signs of early puberty.

NON-CLASSIC FORM

The non-classic type (late-onset type) is the the least-severe form of 21-OHD CAH and is caused by a mild deficiency of the 21-hydroxylase enzyme. Individuals with this type may start experiencing symptoms related to excess androgen production in childhood, adolescence, or adulthood. Both males and females may exhibit rapid growth in childhood, shorter-than-average stature in adulthood, virilization, and infertility. Additionally, girls may experience symptoms of masculinization and abnormal menstruation. However, some individuals with non-classic CAH may never know they are affected because the symptoms are so mild.

How Common Is Congenital Adrenal Hyperplasia, CYP21A2-Related?

The incidence of 21-OHD CAH varies by type and ethnicity. The incidence for the classic form is approximately 1 in 15,000 births worldwide. The prevalence of the classic form varies from 1 in 300 for Yupik Eskimos in Alaska to 1 in 21,000 in Japanese. The non-classic form of 21-OHD CAH is much more common, with an incidence of approximately 1 in 1000 births. The prevalence of the non-classic form is much higher in some ethnicities, namely in the Ashkenazi Jewish (1 in 27), Hispanic (1 in 40), Slavic (1 in 50), and Italian (1 in 300) ethnicities. Mutations in *CYP21A2* account for about 90% of CAH cases.

How Is Congenital Adrenal Hyperplasia, CYP21A2-Related Treated?

Currently, there is no cure for CAH. However, treatments are available to address some of the associated symptoms. Patients benefit from taking hormone-replacement medications, which work to increase levels of deficient hormones and suppress the overproduction of male hormones. Most individuals with classic CAH will need to take hormone medications for the rest of their lives. Those with the less-severe forms of CAH are sometimes able to stop taking these medications in adulthood and are typically treated with lower doses. Some individuals with non-classic CAH do not require any treatment. A multidisciplinary team of physicians, including an endocrinologist, will need to monitor the medication dosage, medication side effects, growth, and sexual development of patients who continue to receive treatment.

Newborn females with ambiguous genitalia may need surgery to correct the function and appearance of the external genitalia. Surgery, if needed, is most often performed during infancy, but can be performed later in life. Treatments provided during pregnancy may reduce the degree of virilization in female fetuses. However, because the long-term safety of prenatal treatment is unknown, these therapies are considered experimental and are not recommended by professional guidelines.

What Is the Prognosis for an Individual with Congenital Adrenal Hyperplasia, CYP21A2-Related?

With early diagnosis and proper medication management, most individuals with 21-OHD CAH will have a normal life expectancy. Early death can occur during periods of significant sodium loss (salt crises) if medication dosage is not adequately adjusted, especially during times of illness or trauma. Problems with growth and development, ambiguous genitalia, and virilization are monitored by physicians on an ongoing basis. Females with 21-OHD CAH can become pregnant, but fertility is reduced.

POSITIVE: CARRIER USH2A-related Disorders

Reproductive risk: 1 in 580
 Risk before testing: 1 in 83,000

Gene: USH2A | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 12821	No partner tested
Result	Carrier	N/A
Variant(s)	NM_206933.2(USH2A):c.2299delG(E767Sfs*21) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of USH2A-related disorders. Carriers generally do not experience symptoms.	N/A
Detection rate	98%	N/A
Exons tested	NM_206933:2-72.	N/A

What are USH2A-related Disorders?

USH2A-related disorders are an inherited group of conditions associated with vision loss and, in many cases, hearing loss. This group of disorders does not affect intelligence or cause any other primary health problems. USH2A-related disorders are caused by harmful genetic changes (mutations) in the *USH2A* gene. *USH2A* plays an important role in the development of the inner ear and the light-sensitive tissue of the eyes (retina). The two types of USH2A-related disorders are Usher syndrome type II and retinitis pigmentosa type 39.

USHER SYNDROME TYPE II

Mutations in *USH2A* are the most common cause of Usher syndrome type II (USH2). Individuals with USH2 have mild to severe hearing loss beginning at birth (congenital). The hearing loss is usually not progressive, and it mainly affects the ability to detect high-pitched (high-frequency) sounds. Individuals with USH2 do not typically have balance issues associated with other types of Usher syndrome.

USH2 also causes vision loss, typically beginning in adolescence or early adulthood. This is due to a condition called retinitis pigmentosa (RP). RP is an eye disease that causes night blindness and a gradual loss of side vision (peripheral vision). Eventually only the central vision remains, creating "tunnel vision." This central vision can also become impaired and can lead to total blindness in a small number of individuals with the disease. In some cases, individuals with USH2 develop clouding in the lens of the eye (cataracts), which can further impair vision.

RETINITIS PIGMENTOSA 39 (RP39)

Some individuals with *USH2A* mutations have retinitis pigmentosa without hearing loss, a condition known as retinitis pigmentosa 39 (RP39).

How common are USH2A-related Disorders?

Several genes are known to cause Usher syndrome, which affects approximately 3-6 in 100,000 individuals worldwide. The proportion of Usher syndrome cases caused by *USH2A* differs significantly between ethnic groups.

The exact incidence of RP39 is not known. The prevalence of RP that occurs in the absence of other symptoms (non-syndromic RP) is approximately 1 in 3,000 and 1 in 7,000 worldwide. RP39 is thought to be the most common cause of non-syndromic RP, accounting for 10-15% of cases.

How are USH2A-related Disorders treated?

Currently there is no cure for hearing loss associated with USH2, but early treatment is important, as it provides a child the best opportunity to develop communication skills. While a child is young, his or her brain is most receptive to learning spoken language or sign language. Hearing aids can improve hearing. Some individuals with more severe hearing loss may benefit from surgical implantation of a small device that stimulates the hearing nerve (a cochlear implant). Specialists can introduce those with hearing loss to other available tools and methods of instruction. To help the child adapt, it is often helpful if the family undergoes such instruction together.

For vision loss due to RP, visual aids and specialized instruction (for example, in tactile signing) help individuals adapt to their limited vision. Use of UVA- and UVB-blocking sunglasses is recommended, and other optical aids may increase eye comfort. For some, therapy with vitamin A palmitate may slow retinal degeneration.

Research is currently being done to determine whether Usher syndrome can be treated with specific medications or gene therapy in the future.

What is the prognosis for an individual with a USH2A-related Disorder?

USH2 results in hearing and vision impairment, while RP39 results in vision impairment with normal hearing. However, neither of these conditions affect one's lifespan or intelligence.

Methods and Limitations

DONOR 12821 [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 $\geq 75\%$. 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. If the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If 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.



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FEMALE
N/A

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

A handwritten signature in black ink that reads "Karla R. Bowles".

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Erik Zmuda, PhD, Diplomate of the American Board of Medical Genetics and Genomics, CGMB on Dec 10, 2021

Conditions Tested

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Congenital Adrenal Hyperplasia, CYP21A2-related - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. Variants (13): CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** African or African American 92%.

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

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

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

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

Cystic Fibrosis - Gene: CFTR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** African or African American >99%.

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

D-bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000414:1-24. **Detection Rate:** African or African American 98%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. **Detection Rate:** African or African American 87%.

Gaucher Disease - Gene: 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:** African or African American 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:** African or African American >99%.

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

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

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

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

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

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001164277 3-11. **Detection Rate:** African or African American >99%.

Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000642:2-34. **Detection Rate:** African or African American >99%.

GNE Myopathy - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001128227:1-12. **Detection Rate:** African or African American >99%.

GNPTAB-related Disorders - Gene: GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024312:1-21. **Detection Rate:** African or African American >99%.

HADHA-related Disorders - Gene: HADHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000182:1-20. **Detection Rate:** African or African American >99%.

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:** African or African American >99%.

Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000035:2-9. **Detection Rate:** African or African American >99%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000520:1-14. **Detection Rate:** African or African American >99%.

HMG-CoA Lyase Deficiency - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000191:1-9. **Detection Rate:** African or African American >99%.

Holocarboxylase Synthetase Deficiency - Gene: HLCS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000411:4-12. **Detection Rate:** African or African American >99%.

Homocystinuria, CBS-related - Gene: CBS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000071:3-17. **Detection Rate:** African or African American >99%.

Hydrolethalus Syndrome - Gene: HYL1. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_145014:4. **Detection Rate:** African or African American >99%.

Hypophosphatasia - Gene: ALPL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000478:2-12. **Detection Rate:** African or African American >99%.

Isovaleric Acidemia - Gene: IVD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002225:1-12. **Detection Rate:** African or African American >99%.

Joubert Syndrome 2 - Gene: TMEM216. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001173990:1-5. **Detection Rate:** African or African American >99%.

Junctional Epidermolysis Bullosa, LAMA3-related - Gene: LAMA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000227:1-38. **Detection Rate:** African or African American >99%.

Junctional Epidermolysis Bullosa, LAMB3-related - Gene: LAMB3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000228:2-23. **Detection Rate:** African or African American >99%.

Junctional Epidermolysis Bullosa, LAMC2-related - Gene: LAMC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_005562:1-23. **Detection Rate:** African or African American >99%.

Krabbe Disease - Gene: GALC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000153:1-17. **Detection Rate:** African or African American >99%.

Leigh Syndrome, French-Canadian Type - Gene: LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_133259:1-38. **Detection Rate:** African or African American >99%.

Lipoid Congenital Adrenal Hyperplasia - Gene: STAR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000349:1-7. **Detection Rate:** African or African American >99%.

Lysosomal Acid Lipase Deficiency - Gene: LIPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000235:2-10. **Detection Rate:** African or African American 98%.

Maple Syrup Urine Disease Type Ia - Gene: BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000709:1-9. **Detection Rate:** African or African American >99%.

Maple Syrup Urine Disease Type Ib - Gene: BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_183050:1-10. **Detection Rate:** African or African American >99%.

Maple Syrup Urine Disease Type II - Gene: DBT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001918:1-11. **Detection Rate:** African or African American 97%.

Medium Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000016:1-12. **Detection Rate:** African or African American >99%.

Megalencephalic Leukoencephalopathy with Subcortical Cysts - Gene: MLC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015166 2-12. **Detection Rate:** African or African American >99%.

Metachromatic Leukodystrophy - Gene: ARSA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000487:1-8. **Detection Rate:** African or African American >99%.

Methylmalonic Acidemia, cblA Type - Gene: MMAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_172250:2-7. **Detection Rate:** African or African American >99%.

Methylmalonic Acidemia, cblB Type - Gene: MMAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_052845:1-9. **Detection Rate:** African or African American >99%.

Methylmalonic Acidemia, MMUT-related - Gene: MMUT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000255:2-13. **Detection Rate:** African or African American >99%.

Methylmalonic Aciduria and Homocystinuria, cblC Type - Gene: MMACHC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015506:1-4. **Detection Rate:** African or African American >99%.

MKS1-related Disorders - Gene: MKS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017777:1-18. **Detection Rate:** African or African American >99%.

Mucopolipidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032520:1-11. **Detection Rate:** African or African American 98%.

Mucopolipidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_020533:1-14. **Detection Rate:** African or African American >99%.

Mucopolysaccharidosis Type I - Gene: IDUA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000203:1-14. **Detection Rate:** African or African American >99%.

Mucopolysaccharidosis Type II - Gene: IDS. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000202:1-9. **Detection Rate:** African or African American 89%.

Mucopolysaccharidosis Type IIIA - Gene: SGSH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000199:1-8. **Detection Rate:** African or African American >99%.

Mucopolysaccharidosis Type IIIB - Gene: NAGLU. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000263:1-6. **Detection Rate:** African or African American >99%.

Mucopolysaccharidosis Type IIIC - Gene: HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_152419:1-18. **Detection Rate:** African or African American >99%.

Muscular Dystrophy, LAMA2-related - Gene: LAMA2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000426:1-43,45-65. **Detection Rate:** African or African American 98%.

MYO7A-related Disorders - Gene: MYO7A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000260:2-49. **Detection Rate:** African or African American >99%.

NEB-related Nemaline Myopathy - Gene: NEB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001271208:3-80,117-183. **Detection Rate:** African or African American 92%.

Nephrotic Syndrome, NPHS1-related - Gene: NPHS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004646:1-29. **Detection Rate:** African or African American >99%.

Nephrotic Syndrome, NPHS2-related - Gene: NPHS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_014625:1-8. **Detection Rate:** African or African American >99%.

Neuronal Ceroid Lipofuscinosis, CLN6-related - Gene: CLN6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017882:1-7. **Detection Rate:** African or African American >99%.

Niemann-Pick Disease Type C1 - Gene: NPC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000271:1-25. **Detection Rate:** African or African American >99%.

Niemann-Pick Disease Type C2 - Gene: NPC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006432:1-5. **Detection Rate:** African or African American >99%.

Niemann-Pick Disease, SMPD1-related - Gene: SMPD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000543:1-6. **Detection Rate:** African or African American >99%.

Nijmegen Breakage Syndrome - Gene: NBN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002485:1-16. **Detection Rate:** African or African American >99%.

Ornithine Transcarbamylase Deficiency - Gene: OTC. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000531:1-10. **Detection Rate:** African or African American 97%.

PCCA-related Propionic Acidemia - Gene: PCCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000282:1-24. **Detection Rate:** African or African American 95%.

PCCB-related Propionic Acidemia - Gene: PCCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000532:1-15. **Detection Rate:** African or African American >99%.

PCDH15-related Disorders - Gene: PCDH15. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_033056:2-33. **Detection Rate:** African or African American 93%.

Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000441:2-21. **Detection Rate:** African or African American >99%.

Peroxisome Biogenesis Disorder Type 1 - Gene: PEX1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000466:1-24. **Detection Rate:** African or African American >99%.

Peroxisome Biogenesis Disorder Type 3 - Gene: PEX12. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000286:1-3. **Detection Rate:** African or African American >99%.

Peroxisome Biogenesis Disorder Type 4 - Gene: PEX6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000287:1-17. **Detection Rate:** African or African American 97%.

Peroxisome Biogenesis Disorder Type 5 - Gene: PEX2. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_000318:4. **Detection Rate:** African or African American >99%.

Peroxisome Biogenesis Disorder Type 6 - Gene: PEX10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_153818:1-6. **Detection Rate:** African or African American >99%.

Phenylalanine Hydroxylase Deficiency - Gene: PAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000277:1-13. **Detection Rate:** African or African American >99%.

POMGNT-related Disorders - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017739:2-22. **Detection Rate:** African or African American 96%.

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000152:2-20. **Detection Rate:** African or African American >99%.

PPT1-related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000310:1-9. **Detection Rate:** African or African American >99%.

Primary Carnitine Deficiency - Gene: SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003060:1-10. **Detection Rate:** African or African American >99%.

Primary Hyperoxaluria Type 1 - Gene: AGXT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000030:1-11. **Detection Rate:** African or African American >99%.

Primary Hyperoxaluria Type 2 - Gene: GRHPR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012203:1-9. **Detection Rate:** African or African American >99%.

Primary Hyperoxaluria Type 3 - Gene: HOGA1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_138413:1-7. **Detection Rate:** African or African American >99%.

Pycnodysostosis - Gene: CTSK. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000396:2-8. **Detection Rate:** African or African American >99%.

Pyruvate Carboxylase Deficiency - Gene: PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000920:3-22. **Detection Rate:** African or African American >99%.

Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000288:1-10. **Detection Rate:** African or African American >99%.

RTEL1-related Disorders - Gene: RTEL1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032957:2-35. **Detection Rate:** African or African American >99%.

Sandhoff Disease - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000521:1-14. **Detection Rate:** African or African American 98%.

Short-chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000017:1-10. **Detection Rate:** African or African American >99%.

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000382:1-10. **Detection Rate:** African or African American 96%.

SLC26A2-related Disorders - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000112:2-3. **Detection Rate:** African or African American >99%.

Smith-Lemli-Opitz Syndrome - Gene: DHCR7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001360:3-9. **Detection Rate:** African or African American >99%.

Spastic Paraplegia Type 15 - Gene: ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015346:2-42. **Detection Rate:** African or African American >99%.

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. **Detection Rate:** African or African American 71%.

Spondylothoracic Dysostosis - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001039958:1-2. **Detection Rate:** African or African American >99%.

TGM1-related Autosomal Recessive Congenital Ichthyosis - Gene: TGM1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000359 2-15. **Detection Rate:** African or African American >99%.

TPP1-related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000391:1-13. **Detection Rate:** African or African American >99%.

Tyrosine Hydroxylase Deficiency - Gene: TH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_199292:1-14. **Detection Rate:** African or African American >99%.

Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000137:1-14. **Detection Rate:** African or African American >99%.



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 12/10/2021

MALE
DONOR 12821
DOB:
Ethnicity: African or African
American
Barcode: 11004513015678

FEMALE
N/A

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

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

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

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

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

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

X-linked Adrenal Hypoplasia Congenita - Gene: NROB1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. **Detection Rate:** African or African American 97%.

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

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

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

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

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

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

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

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 12821 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	Alpha globin status: aa/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 41,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 44,000	< 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 38,000	< 1 in 1,000,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 7,400	< 1 in 1,000,000
Canavan Disease	1 in 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	NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote †	1 in 550
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



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 12/10/2021

MALE
DONOR 12821
 DOB:
 Ethnicity: African or African
 American
 Barcode: 11004513015678

FEMALE
 N/A

Disease	DONOR 12821 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 6,100	< 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
Dihydroliipoamide 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 530	1 in 12,000
Fanconi Anemia Complementation Group A	1 in 3,100	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 42,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 44,000	< 1 in 1,000,000
Galactosemia	1 in 7,000	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 2,600	< 1 in 1,000,000
Gaucher Disease	1 in 310	1 in 150,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 4,100	1 in 670,000
GLB1-related Disorders	1 in 17,000	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycine Encephalopathy, AMT-related	1 in 26,000	< 1 in 1,000,000
Glycine Encephalopathy, GLDC-related	1 in 2,500	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 8,700	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	< 1 in 50,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 25,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 700	1 in 21,000
Hereditary Fructose Intolerance	1 in 23,000	< 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 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 17,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 26,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 36,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 13,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	K304E heterozygote †	1 in 240
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



RESULTS RECIPIENT
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MALE
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 American
 Barcode: 11004513015678

FEMALE
 N/A

Disease	DONOR 12821 Residual Risk	Reproductive Risk
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, MMUT-related	1 in 18,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucopolipidosis III Gamma	< 1 in 20,000	< 1 in 1,000,000
Mucopolipidosis 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 11,000	< 1 in 1,000,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 5,900	< 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 20,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	< 1 in 50,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
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 18,000	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 400	1 in 100,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	< 1 in 1,000,000
USH2A-related Disorders	NM_206933.2(USH2A):c.2299delG(E767Sfs*21) heterozygote †	1 in 580



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Disease	DONOR 12821 Residual Risk	Reproductive Risk
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 14,000	< 1 in 1,000,000
Wilson Disease	1 in 9,000	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 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 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000