



RESULTS RECIPIENT
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 Report Date: 11/06/2020

MALE
DONOR 12661
 DOB: [REDACTED]
 Ethnicity: Ashkenazi Jewish
 Sample Type: EDTA Blood
 Date of Collection: 10/27/2020
 Date Received: 10/29/2020
 Date Tested: 11/05/2020
 Barcode: 11004512732147
 Accession ID:
 CSLHN9Y9P3MEWGL
 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 12661	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested)	N/A
POSITIVE: CARRIER HADHA-related Disorders Reproductive Risk: 1 in 1,000 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_000182.4(HADHA):c.1528G>C(E510Q, aka E474Q) 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 Biotinidase Deficiency Reproductive Risk: 1 in 1,700 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_000060.2(BTD):c.1330G>C(D444H) 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 Nephrotic Syndrome, NPHS2-related Reproductive Risk: 1 in 110,000 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_014625.2(NPHS2):c.686G>A(R229Q) 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 10.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- 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 HADHA-related Disorders

Reproductive risk: 1 in 1,000
 Risk before testing: 1 in 250,000

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

Patient	DONOR 12661	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000182.4(HADHA):c.1528G>C(E510Q, aka E474Q) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of HADHA-related disorders. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000182:1-20.	N/A

What are HADHA-Related Disorders?

HADHA-related disorders result from the body lacking an enzyme called mitochondrial trifunctional protein. Without this enzyme, the body has trouble turning a specific type of fat from foods, known as long-chain fatty acids, into energy. This process, called fatty acid oxidation, normally breaks down long-chain fatty acids stepwise until they can be turned into usable energy. When mitochondrial trifunctional protein is missing, fatty acids build up in the body and cause damage to organs and tissues. HADHA-related disorders are caused by mutations in the *HADHA* gene.

LONG-CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY

Individuals with long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD) have trouble converting long-chain fatty acids into energy, especially when they are not eating for a period of time (fasting), when they are ill, or when they do strenuous exercise. Symptoms can begin in infancy and include poor feeding, muscle weakness (hypotonia), low blood sugar (hypoglycemia), and low energy (lethargy). There can also be liver problems, vision loss due to damage to the retina, muscle breakdown, seizures, and sensory problems in the arms and legs (peripheral neuropathy). In more severe situations, individuals with LCHADD can have problems breathing and trouble with their heart muscle (cardiomyopathy) or rhythm (arrhythmia), which can lead to coma or death.

MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY (MTPD)

Individuals with MTPD tend to have similar symptoms to those with LCHADD, though the symptoms may be more severe. Most babies with the neonatal form of MTPD die in infancy. Children with a less-severe form of MTPD may not show symptoms until times of fasting, when they are ill, or when they do strenuous exercise. Some children may not have symptoms between these episodes, but repeated crisis events can lead to brain damage and intellectual and developmental disabilities.

How Common Are HADHA-Related Disorders?

The incidence of HADHA-related disorders in the United States is estimated at approximately 1 in 90,000 individuals. A similar carrier frequency and incidence (1 in 91,700) is seen in Estonia while the incidence in Germany and Poland ranges from 1 in 250,000 to 1 in 109,090 births with certain regions of Poland seeing a higher incidence of up to 1 in 16,900 births.

How Are HADHA-Related Disorders Treated?

The main method of management for LCHADD and MTPD is a special diet and avoidance of fasting. A physician or nutritionist will recommend a diet low in fats and high in carbohydrates, which are easier for an affected individual to break down, and a feeding schedule with frequent meals. Often it is necessary to have an additional dietary protocol in place for illness or other stressful times. A physician may also prescribe medium-chain triglyceride oil, L-carnitine, or other supplements for additional energy.

What Is the Prognosis for an Individual with a HADHA-Related Disorder?

Untreated, LCHADD and MTPD are often fatal in infancy or childhood. When symptoms appear in infancy, treatment is often not effective because the disease causes irreparable damage to the heart and leads to cognitive impairments.

For some cases of LCHADD and less-severe forms of MTPD, early detection and early treatment can prevent many of the severe complications and allow affected individuals to have typical growth and development. Even with careful treatment, there may still be some episodes of low blood sugar and damage to the heart, liver, and muscle. Recurrent acute episodes of low blood sugar can lead to cognitive impairments over time. With treatment, some individuals with LCHADD or MTPD may live into adulthood.

Individuals with later-onset disease and symptoms limited to muscle weakness and pain are typically healthy and do not have problems with the heart, liver, or changes in cognitive ability or intellect.

Additional Considerations for Carriers

Carriers of fatty-acid oxidation defects, including HADHA-related disorders, 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 HADHA-related disorder. 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 HADHA-related disorder, should speak with her physician for recommendations and may benefit from consultation with a high-risk physician.

POSITIVE: CARRIER

Biotinidase Deficiency

Reproductive risk: 1 in 1,700
 Risk before testing: 1 in 13,000

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

Patient	DONOR 12661	No partner tested
Result	Carrier	N/A
Variant(s)	NM_000060.2(<i>BTD</i>):c.1330G>C(D444H) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of biotinidase deficiency. Carriers generally do not experience symptoms. D444H is a partial biotinidase deficiency mutation.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000060:1-4.	N/A

What Is Biotinidase Deficiency?

Biotinidase deficiency is a highly treatable inherited disease in which the body cannot process biotin (vitamin B7), due to a deficiency in an enzyme called biotinidase. Biotinidase deficiency is caused by mutations in the *BTD* gene.

PROFOUND BIOTINIDASE DEFICIENCY

Individuals who have less than 10% of the normal amount of the enzyme biotinidase are said to have profound biotinidase deficiency. Without treatment, their symptoms tend to be significant. Individuals with biotinidase deficiency can experience seizures, poor muscle tone, difficulty with movement and balance, vision loss, hearing loss, skin rashes, breathing problems, hair loss, fungal infections, and intellectual and/or developmental delays. These symptoms often begin after the first few weeks or months of life and can be life-threatening if untreated.

PARTIAL BIOTINIDASE DEFICIENCY

Individuals who have between 10% and 30% of the normal amounts of biotinidase have a milder form of the disease known as partial biotinidase deficiency. They may experience less-severe symptoms, or they may not show any symptoms until they become ill or stressed.

How Common Is Biotinidase Deficiency?

The incidence of profound biotinidase deficiency is approximately 1 in 137,000 births. The prevalence of partial biotinidase deficiency is approximately 1 in 110,000 people. Since partial biotinidase deficiency can be mild, it is possible that the true prevalence is more common.

How Is Biotinidase Deficiency Treated?

Biotinidase deficiency is treated with a biotin pill taken daily by mouth. A physician can determine the proper dosage and adjust that dosage over time if necessary. This treatment is lifelong and highly effective. Both people with profound biotinidase deficiency and partial biotinidase deficiency should take biotin supplements.

It is important to start biotin supplementation as soon as possible. Treatment with biotin supplements can help improve some symptoms of biotinidase deficiency. If there is delayed treatment, symptoms such as vision loss, hearing loss, and developmental delay are not reversible.

For people who have vision or hearing loss, vision aids or hearing aids may be helpful. Learning specialists can help patients with intellectual delay learn as effectively as possible.

What Is the Prognosis for a Person with Biotinidase Deficiency?

With early detection and treatment, a person with biotinidase deficiency can live a completely normal life. If left untreated, the disease can cause life-threatening complications. When the disease is not detected early, patients may experience permanent damage to their hearing, vision, and intellectual ability. In cases where the disease is entirely unrecognized, it can be life-threatening.

POSITIVE: CARRIER

Nephrotic Syndrome, NPHS2-related

Reproductive risk: 1 in 110,000

Risk before testing: 1 in 310,000

Gene: NPHS2 | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 12661	No partner tested
Result	Carrier	N/A
Variant(s)	NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of nephrotic syndrome, NPHS2-related. Carriers generally do not experience symptoms. The pathogenicity of R229Q is dependent on the variant observed on the other chromosome.	N/A
Detection rate	>99%	N/A
Exons tested	NM_014625:1-8.	N/A

What Is Nephrotic Syndrome, NPHS2-Related?

Nephrotic syndrome, NPHS2-related is an inherited condition that causes issues with kidney function often leading to kidney failure. Mutations in the *NPHS2* gene cause a form of nephrotic syndrome that is unresponsive to steroid treatment known as steroid-resistant nephrotic syndrome (SRNS). Symptoms of the condition typically begin between 4 and 12 months of age, but in some cases occur later in childhood.

Symptoms of the condition include an excess of protein in the urine (proteinuria), low levels of protein in the blood, kidney failure, and swelling of the body (edema). The swelling can also cause weight gain and high blood pressure. Individuals with nephrotic syndrome are prone to infection due to their inability to retain sufficient amounts of serum antibodies. They are also prone to develop harmful blood clots. Kidney failure typically occurs before the age of 20, and kidney transplantation may allow for a more normal lifespan.

How Common Is Nephrotic Syndrome, NPHS2-Related?

The incidence of all childhood nephrotic syndrome is 2 to 16 per 100,000 individuals worldwide of which 10-20% have SRNS. Approximately 10% of individuals with SRNS carry mutations in the *NPHS2* gene.

How Is Nephrotic Syndrome, NPHS2-Related Treated?

The goal of treatment is to minimize damage to the kidneys. Medication to control blood pressure and high cholesterol may be prescribed. Often children with nephrotic syndrome with protein loss require antibiotics to control for infection. A physician may recommend infusions of protein for children with SRNS to help replace what is lost in the urine. Diuretic drugs may help eliminate excess water and thus reduce swelling while blood thinners may be required to aid in blood clotting. Typically, kidney failure will occur, and a kidney transplant will be required though symptoms of the disease can recur after transplant.



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MALE
DONOR 12661
DOB: [REDACTED]
Ethnicity: Ashkenazi Jewish
Barcode: 11004512732147

FEMALE
N/A

What Is the Prognosis for Nephrotic Syndrome, NPHS2-Related?

The prognosis for an individual with nephrotic syndrome, NPHS2-related varies, but with transplantation and careful medical management, affected children can live into adulthood.

Methods and Limitations

DONOR 12661 [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, large upstream deletions involving the genes *GJB6* and/or *CRYL1* that affect the expression of *GJB2* 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.



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Ethnicity: Ashkenazi Jewish
Barcode: 11004512732147

FEMALE
N/A

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. 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. 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 Nov 6, 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:** Ashkenazi Jewish 94%.
- 6-pyruvoyl-tetrahydropterin Synthase Deficiency** - Gene: PTS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000317:1-6. **Detection Rate:** Ashkenazi Jewish >99%.
- ABCC8-related Familial Hyperinsulinism** - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000352:1-39. **Detection Rate:** Ashkenazi Jewish >99%.
- Adenosine Deaminase Deficiency** - Gene: ADA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000022:1-12. **Detection Rate:** Ashkenazi Jewish >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:** Ashkenazi Jewish >99%.
- Alpha-sarcoglycanopathy** - Gene: SGCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000023:1-9. **Detection Rate:** Ashkenazi Jewish >99%.
- Alstrom Syndrome** - Gene: ALMS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015120:1-23. **Detection Rate:** Ashkenazi Jewish >99%.
- AMT-related Glycine Encephalopathy** - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000481:1-9. **Detection Rate:** Ashkenazi Jewish >99%.
- Andermann Syndrome** - Gene: SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_133647:1-25. **Detection Rate:** Ashkenazi Jewish >99%.
- Argininemia** - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000045:1-8. **Detection Rate:** Ashkenazi Jewish 97%.
- Argininosuccinic Aciduria** - Gene: ASL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001024943:1-16. **Detection Rate:** Ashkenazi Jewish >99%.
- Aspartylglucosaminuria** - Gene: AGA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000027:1-9. **Detection Rate:** Ashkenazi Jewish >99%.
- Ataxia with Vitamin E Deficiency** - Gene: TTPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000370:1-5. **Detection Rate:** Ashkenazi Jewish >99%.
- Ataxia-telangiectasia** - Gene: ATM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000051:2-63. **Detection Rate:** Ashkenazi Jewish >99%.
- ATP7A-related Disorders** - Gene: ATP7A. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000052:2-23. **Detection Rate:** Ashkenazi Jewish 92%.
- Autoimmune Polyglandular Syndrome Type 1** - Gene: AIRE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000383:1-14. **Detection Rate:** Ashkenazi Jewish >99%.
- Autosomal Recessive Osteopetrosis Type 1** - Gene: TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006019:2-20. **Detection Rate:** Ashkenazi Jewish >99%.
- Autosomal Recessive Polycystic Kidney Disease, PKHD1-related** - Gene: PKHD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_138694 2-67. **Detection Rate:** Ashkenazi Jewish >99%.
- Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay** - Gene: SACS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_014363 2-10. **Detection Rate:** Ashkenazi Jewish 99%.
- Bardet-Biedl Syndrome, BBS1-related** - Gene: BBS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024649:1-17. **Detection Rate:** Ashkenazi Jewish >99%.
- Bardet-Biedl Syndrome, BBS10-related** - Gene: BBS10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024685:1-2. **Detection Rate:** Ashkenazi Jewish >99%.
- Bardet-Biedl Syndrome, BBS12-related** - Gene: BBS12. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_152618:2. **Detection Rate:** Ashkenazi Jewish >99%.
- Bardet-Biedl Syndrome, BBS2-related** - Gene: BBS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_031885:1-17. **Detection Rate:** Ashkenazi Jewish >99%.
- BCS1L-related Disorders** - Gene: BCS1L. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004328:3-9. **Detection Rate:** Ashkenazi Jewish >99%.
- Beta-sarcoglycanopathy** - Gene: SGCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000232:1-6. **Detection Rate:** Ashkenazi Jewish >99%.
- Biotinidase Deficiency** - Gene: BTD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000060:1-4. **Detection Rate:** Ashkenazi Jewish >99%.
- Bloom Syndrome** - Gene: BLM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000057:2-22. **Detection Rate:** Ashkenazi Jewish >99%.
- Calpainopathy** - Gene: CAPN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000070:1-24. **Detection Rate:** Ashkenazi Jewish >99%.
- Canavan Disease** - Gene: ASPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000049:1-6. **Detection Rate:** Ashkenazi Jewish 98%.
- Carbamoylphosphate Synthetase I Deficiency** - Gene: CPS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001875:1-38. **Detection Rate:** Ashkenazi Jewish >99%.
- Carnitine Palmitoyltransferase IA Deficiency** - Gene: CPT1A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001876:2-19. **Detection Rate:** Ashkenazi Jewish >99%.
- Carnitine Palmitoyltransferase II Deficiency** - Gene: CPT2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000098:1-5. **Detection Rate:** Ashkenazi Jewish >99%.
- Cartilage-hair Hypoplasia** - Gene: RMRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NR_003051:1. **Detection Rate:** Ashkenazi Jewish >99%.
- Cerebrotendinous Xanthomatosis** - Gene: CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000784:1-9. **Detection Rate:** Ashkenazi Jewish >99%.
- Citrullinemia Type 1** - Gene: ASS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000050:3-16. **Detection Rate:** Ashkenazi Jewish >99%.
- CLN3-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001042432 2-16. **Detection Rate:** Ashkenazi Jewish >99%.
- CLN5-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006493:1-4. **Detection Rate:** Ashkenazi Jewish >99%.
- CLN6-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017882:1-7. **Detection Rate:** Ashkenazi Jewish >99%.
- CLN8-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_018941:2-3. **Detection Rate:** Ashkenazi Jewish >99%.
- Cohen Syndrome** - Gene: VPS13B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017890:2-62. **Detection Rate:** Ashkenazi Jewish 97%.
- COL4A3-related Alport Syndrome** - Gene: COL4A3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000091:1-52. **Detection Rate:** Ashkenazi Jewish 97%.

COL4A4-related Alport Syndrome - Gene: COL4A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000092:2-48. **Detection Rate:** Ashkenazi Jewish 98%.

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

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:** Ashkenazi Jewish >99%.

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

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

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

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

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

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

Delta-sarcoglycanopathy - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000337:2-9. **Detection Rate:** Ashkenazi Jewish 99%.

Dihydroipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000108:1-14. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

ERCC6-related Disorders - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000124:2-21. **Detection Rate:** Ashkenazi Jewish 99%.

ERCC8-related Disorders - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000082:1-12. **Detection Rate:** Ashkenazi Jewish 95%.

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

EVC2-related Ellis-van Creveld Syndrome - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_147127:1-22. **Detection Rate:** Ashkenazi Jewish >99%.

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

Familial Dysautonomia - Gene: IKBKAP. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003640:2-37. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

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

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

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

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

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

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

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. **Detection Rate:** Ashkenazi Jewish 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:** Ashkenazi Jewish 95%.

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - Gene: GJB2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004004:1-2. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

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

Glycogen Storage Disease Type Ia - Gene: G6PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000151:1-5. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

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

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

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

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

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

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA.

Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000520:1-14. **Detection Rate:** Ashkenazi Jewish >99%.

HMG-CoA Lyase Deficiency - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000191:1-9. **Detection Rate:** Ashkenazi Jewish 98%.

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

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

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

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

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

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

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

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

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

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

LAMA2-related Muscular Dystrophy - Gene: LAMA2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000426:1-65. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

Lysosomal Acid Lipase Deficiency - Gene: LIPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000235:2-10. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

Maple Syrup Urine Disease Type II - Gene: DBT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001918:1-11. **Detection Rate:** Ashkenazi Jewish 96%.

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

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

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

Methylmalonic Acidemia, cblA Type - Gene: MMAA. Autosomal Recessive.

Sequencing with copy number analysis. Exons: NM_172250:2-7. **Detection Rate:** Ashkenazi Jewish >99%.

Methylmalonic Acidemia, cblB Type - Gene: MMAB. Autosomal Recessive.

Sequencing with copy number analysis. Exons: NM_052845:1-9. **Detection Rate:** Ashkenazi Jewish >99%.

Methylmalonic Aciduria and Homocystinuria, cblC Type - Gene: MMACHC.

Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015506:1-4. **Detection Rate:** Ashkenazi Jewish >99%.

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

Mucopolidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032520:1-11. **Detection Rate:** Ashkenazi Jewish >99%.

Mucopolidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_020533:1-14. **Detection Rate:** Ashkenazi Jewish >99%.

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

Mucopolysaccharidosis Type II - Gene: IDS. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000202:1-9. **Detection Rate:** Ashkenazi Jewish 88%.

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

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

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

MUT-related Methylmalonic Acidemia - Gene: MUT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000255:2-13. **Detection Rate:** Ashkenazi Jewish >99%.

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

NEB-related Nemaline Myopathy - Gene: NEB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001271208:3-80,117-183. **Detection Rate:** Ashkenazi Jewish >99%.

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

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

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

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

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

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

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

PCCA-related Propionic Acidemia - Gene: PCCA. Autosomal Recessive.

Sequencing with copy number analysis. Exons: NM_000282:1-24. **Detection Rate:** Ashkenazi Jewish 95%.

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

PCDH15-related Disorders - Gene: PCDH15. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_033056:2-33. Detection Rate: Ashkenazi Jewish 93%.

Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000441:2-21. Detection Rate: Ashkenazi Jewish >99%.

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

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

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

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

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

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

POMGNT-related Disorders - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017739:2-22. Detection Rate: Ashkenazi Jewish 96%.

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000152:2-20. Detection Rate: Ashkenazi Jewish >99%.

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

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

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

Primary Hyperoxaluria Type 2 - Gene: GRHR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012203:1-9. Detection Rate: Ashkenazi Jewish >99%.

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

Pycnodysostosis - Gene: CTSK. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000396:2-8. Detection Rate: Ashkenazi Jewish >99%.

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

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

RTEL1-related Disorders - Gene: RTEL1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032957:2-35. Detection Rate: Ashkenazi Jewish >99%.

Sandhoff Disease - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000521:1-14. Detection Rate: Ashkenazi Jewish 99%.

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

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000382:1-10. Detection Rate: Ashkenazi Jewish 96%.

SLC26A2-related Disorders - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000112:2-3. Detection Rate: Ashkenazi Jewish >99%.

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

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

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. Detection Rate: Ashkenazi Jewish 94%.

Spondylothoracic Dysostosis - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001039958:1-2. Detection Rate: Ashkenazi Jewish >99%.

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

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

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

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

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

USH1C-related Disorders - Gene: USH1C. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_005709:1-21. Detection Rate: Ashkenazi Jewish >99%.

USH2A-related Disorders - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_206933:2-72. Detection Rate: Ashkenazi Jewish 94%.

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

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

Wilson Disease - Gene: ATP7B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000053:1-21. Detection Rate: Ashkenazi Jewish >99%.

X-linked Adrenoleukodystrophy - Gene: ABCD1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000033:1-6. Detection Rate: Ashkenazi Jewish 77%.

X-linked Alport Syndrome - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000495:1-51. Detection Rate: Ashkenazi Jewish 95%.

X-linked Congenital Adrenal Hypoplasia - Gene: NROB1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. Detection Rate: Ashkenazi Jewish 99%.

X-linked Juvenile Retinoschisis - Gene: RS1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000330:1-6. Detection Rate: Ashkenazi Jewish 98%.

X-linked Myotubular Myopathy - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000252:2-15. Detection Rate: Ashkenazi Jewish 98%.



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 11/06/2020

MALE
DONOR 12661
DOB: [REDACTED]
Ethnicity: Ashkenazi Jewish
Barcode: 11004512732147

FEMALE
N/A

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

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

Xeroderma Pigmentosum Group C - Gene: XPC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004628:1-16. **Detection Rate:** Ashkenazi Jewish 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 12661 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,300	< 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 4,400	1 in 770,000
Adenosine Deaminase Deficiency	1 in 39,000	< 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
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 20,000	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autoimmune Polyglandular Syndrome Type 1	1 in 18,000	< 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 39,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	1 in 14,000	< 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	NM_000060.2(BTD):c.1330G>C(D444H) heterozygote †	1 in 1,700
Bloom Syndrome	1 in 11,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	1 in 3,300	1 in 730,000
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 4,600	1 in 860,000
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
CLN6-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 5,500	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 21,000	< 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 6,100	< 1 in 1,000,000
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
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

Disease	DONOR 12661 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 2,300	1 in 220,000
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	1 in 9,300	< 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 19,000	< 1 in 1,000,000
ERCC8-related Disorders	1 in 7,300	< 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 3,000	1 in 360,000
Familial Mediterranean Fever	1 in 1,000	1 in 42,000
Fanconi Anemia Complementation Group A	1 in 3,100	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	1 in 9,300	< 1 in 1,000,000
FKRP-related Disorders	1 in 19,000	< 1 in 1,000,000
FKTN-related Disorders	1 in 6,300	< 1 in 1,000,000
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 35,000	< 1 in 1,000,000
Galactosemia	1 in 16,000	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	< 1 in 1,000,000
Gaucher Disease	1 in 250	1 in 13,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,000	1 in 160,000
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 7,000	< 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 50,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 32,000	< 1 in 1,000,000
HADHA-related Disorders	E474Q heterozygote †	1 in 1,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 6,600	< 1 in 1,000,000
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 3,000	1 in 350,000
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 27,000	< 1 in 1,000,000
Hydrolethals Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 22,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 26,000	< 1 in 1,000,000
Joubert Syndrome 2	1 in 9,600	< 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 20,000	< 1 in 1,000,000
Krabbe Disease	1 in 17,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	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 30,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 32,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 9,700	< 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 6,000	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000



RESULTS RECIPIENT
 SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 11/06/2020

MALE
 DONOR 12661
 DOB: ██████████
 Ethnicity: Ashkenazi Jewish
 Barcode: 11004512732147

FEMALE
 N/A

Disease	DONOR 12661 Residual Risk	Reproductive Risk
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucopolidosis III Gamma	< 1 in 50,000	< 1 in 1,000,000
Mucopolidosis IV	1 in 8,900	< 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 31,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 43,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 18,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 11,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote †	1 in 110,000
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 10,000	< 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 1,200	1 in 360,000
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 12,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 22,000	< 1 in 1,000,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 10,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 16,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 13,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 8,600	< 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 10,000	< 1 in 1,000,000
Sandhoff Disease	< 1 in 47,000	< 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 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 580	1 in 94,000
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 13,000	< 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 Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 11/06/2020

MALE
DONOR 12661
 DOB: [REDACTED]
 Ethnicity: Ashkenazi Jewish
 Barcode: 11004512732147

FEMALE
 N/A

Disease	DONOR 12661 Residual Risk	Reproductive Risk
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