

RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Dr. Jeffrey Olliffe
 4915 25th Ave NE, Suite 204W
 Seattle, WA 98105
 Phone: (206) 588-1484
 Fax: (206) 466-4696
 NPI: 1306838271
 Report Date: 09/10/2018

MALE
DONOR 10280
 DOB: ██████████
 Ethnicity: Mixed or Other
 Caucasian
 Sample Type: EDTA Blood
 Date of Collection: 08/31/2018
 Date Received: 09/04/2018
 Date Tested: 09/10/2018
 Barcode: 11004212489042
 Accession ID:
 CSLWK4YHKPGRR2Q
 Indication: Egg or sperm donor

FEMALE
 N/A

Foresight™ Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Counsyl 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 10280	Partner
Panel Information	Foresight Carrier Screen Universal Panel (175 conditions tested)	N/A
POSITIVE: CARRIER Gaucher Disease Reproductive Risk: 1 in 450 Inheritance: Autosomal Recessive	+ CARRIER* NM_001005741.2(GBA):c.1226A>G (N409S, aka N370S) 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 510 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 Familial Mediterranean Fever Reproductive Risk: 1 in 2,000 Inheritance: Autosomal Recessive	+ CARRIER* NM_000243.2(MEFV):c.2080A>G (M694V, aka MED) 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 11.

CLINICAL NOTES

- None

NEXT STEPS

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

POSITIVE: CARRIER

Gaucher Disease

Reproductive risk: 1 in 450
 Risk before testing: 1 in 50,000

Gene: GBA | Inheritance Pattern: Autosomal Recessive

Patient	DONOR 10280	No partner tested
Result	Carrier	N/A
Variant(s)	NM_001005741.2(GBA):c.1226A>G(N409S, aka N370S) heterozygote	N/A
Methodology	Analysis of homologous regions	N/A
Interpretation	This individual is a carrier of Gaucher disease. Carriers generally do not experience symptoms. The N370S mutation is associated with Type 1 Gaucher disease.	N/A
Detection rate	60%	N/A
Variants tested	D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18.	N/A

What Is Gaucher Disease?

Gaucher disease is an inherited condition in which the body fails to properly produce a particular enzyme needed to break down a fatty substance called glucocerebroside. Without this enzyme, glucocerebroside and several other associated substances will build up in the body, causing a wide range of symptoms. Gaucher disease is caused by mutations in the *GBA* gene.

There are five main types of Gaucher disease, each with different manifestations. These types are described below.

TYPE 1 FORM

Type 1 Gaucher disease is the most common form. It can affect individuals at any age, and its symptoms vary widely from mild to severe.

Many individuals with type 1 Gaucher disease have symptoms related to their bones. Symptoms may include bone pain, low bone-mineral density, and an increased risk for fractures. On the mild end of the spectrum, some individuals experience only a small drop in their bone-mineral density. In more severe cases, blood supply to the bones is lost, leading to permanent damage. Bone problems are often the most debilitating aspect of the disease.

Individuals with type 1 Gaucher disease often have an enlarged liver and spleen. They may also have a lowered number of red blood cells and platelets. With fewer red blood cells (anemia), a patient will often be tired and weak. Fewer platelets in the blood will make the individual more prone to bruising and excessive bleeding. Lung disease is another possible symptom.

Type 1 is distinct from other forms of Gaucher disease in that it usually does not affect an individual's brain or spinal cord.

TYPE 2 FORM

Type 2 is known as the infantile or acute neuropathic form of Gaucher disease. Symptoms usually appear before age two and progress rapidly. Like individuals with type 1 Gaucher disease, children with type 2 Gaucher disease may have an enlarged liver and spleen, a lowered number of red blood cells (anemia) leading to weakness and tiredness, a lowered number of platelets leading to bleeding and bruising, and lung disease.

While type 2 Gaucher disease does not cause bone problems, it does cause neurological problems. Neurological symptoms often include limited cognitive and motor development, brainstem abnormalities that can cause breathing problems and difficulty swallowing, constant arching of the back and tilting back of the head, uncontrollable tightening and releasing of the muscles, and an inability to open the mouth. As the nervous system deteriorates, children with type 2 Gaucher disease may develop dementia and the inability to coordinate their own movements.

TYPE 3 FORM

Type 3 Gaucher disease is known as the juvenile or chronic neuropathic form. Symptoms often begin before age two, though this is variable. Usually, the symptoms associated with type 3 Gaucher disease progress more slowly than with type 2 Gaucher disease.

Like individuals with type 1 Gaucher disease, children with type 3 Gaucher disease may have an enlarged liver and spleen; a lowered number of red blood cells (anemia) leading to weakness and tiredness; a lowered number of platelets, leading to bleeding and bruising; lung disease; and bone problems, including pain, fractures, and arthritis.

As with type 2 Gaucher disease, type 3 Gaucher disease also causes neurological problems, which may include seizures that worsen over time, progressive cognitive problems, and difficulty controlling eye movement. Toward the end of their lives, individuals with type 3 Gaucher disease may also develop dementia.

PERINATAL-LETHAL FORM

The perinatal-lethal form is a rare but severe form of Gaucher disease. This form usually leads to death *in utero* or shortly after birth. Infants with the disease have symptoms including an enlarged liver and spleen, a lowered number of red blood cells and platelets, neurological problems, skin abnormalities, and often distinct facial features.

CARDIOVASCULAR FORM

The cardiovascular form of Gaucher disease causes symptoms involving the heart, notably a hardening of the mitral and aortic valves. If this symptom is severe, heart-valve replacement may be required. In addition, individuals with the cardiovascular form may have a slightly enlarged liver and spleen; bone problems including pain, fractures, and arthritis; and difficulty controlling eye movement and a clouding of the eye's cornea, which can affect vision. The cardiovascular form of the disease is sometimes called type 3C.

How Common Is Gaucher Disease?

The incidence of Gaucher disease is 1 in 40,000 to 1 in 60,000 in the general population. It is more common in people with Ashkenazi Jewish ancestry, where the incidence is 1 in 800.

How Is Gaucher Disease Treated?

Gaucher disease is treated with enzyme replacement therapy (ERT) and substrate reduction therapy. ERT is given by infusion and helps eliminate the buildup of glucocerebrosides in the body. Oral forms of ERT are also available. For many affected individuals, ERT is effective in treating disease symptoms and preventing complications, particularly bone and organ damage. ERT does not improve or prevent the neurological symptoms found in type 2 and type 3 Gaucher disease.

Individuals with the cardiovascular form of Gaucher disease often need heart-valve replacements, after which ERT can be helpful.

Additional treatments for the symptoms of Gaucher disease include blood transfusions for tiredness and excessive bleeding; joint replacement to relieve pain and restore movement; and medication to treat bone pain.

What Is the Prognosis for an Individual with Gaucher Disease?

Because symptoms of Gaucher disease vary widely in type and severity, both among the different types and among individuals with the same type, the outlook is similarly varied. The prognosis for an individual with Gaucher disease depends on the type of Gaucher disease, the severity of symptoms in that particular individual, and the availability and effectiveness of treatment.

Those with a milder form of type 1 Gaucher disease are expected to have a normal lifespan, particularly if ERT is administered when necessary. Some individuals with severe cases of type 1 Gaucher disease may have debilitating symptoms that are more difficult to manage.

Those with type 2 Gaucher disease often have significant developmental delays and die between the ages of two and four. In the most severe type 2 Gaucher disease cases, death may occur before or shortly after birth.

People with type 3 Gaucher disease usually develop symptoms in childhood that slowly worsen over time. While some type 3 Gaucher disease patients have died in childhood, others have lived into their thirties and forties.

For those with the cardiovascular form of the disease, the prognosis depends upon the success of their valve-replacement surgery.

With the perinatal-lethal form, death occurs before or shortly after birth. Women with milder cases of Gaucher disease can have successful pregnancies.

Some individuals affected with Gaucher disease may also develop Parkinson's disease or Lewy body dementia, but the vast majority of individuals with Gaucher disease never develop these conditions.

Additional Considerations for Carriers

Studies have shown that carriers of Gaucher disease may have an increased risk of developing Parkinson's disease, above the level of risk observed in the general population. This risk could be between 7 and 15% by age 80 (in comparison to the general population's risk of 1 to 2%), but most carriers of Gaucher disease never develop Parkinson's disease. If there is a family history of Parkinson's disease or significant concern about the development of Parkinson's disease, a referral to a neurologist for evaluation is recommended.

POSITIVE: CARRIER

Biotinidase Deficiency

Reproductive risk: 1 in 510
Risk before testing: 1 in 3,200

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

Patient	DONOR 10280	No partner tested
Result	Carrier	N/A
Variant(s)	NM_000060.2(BTD):c.1330G>C(D444H) heterozygote	N/A
Methodology	Sequencing with copy number analysis	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 the vitamin biotin due to a deficiency in a particular enzyme. If left untreated, the disease can cause numerous life-threatening complications. By taking daily supplements of biotin before symptoms occur, however, all symptoms of the disease can be avoided. With early detection and treatment, a person with biotinidase deficiency can live a completely normal life.

PROFOUND BIOTINIDASE DEFICIENCY

People 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. People with biotinidase deficiency can experience seizures, poor muscle tone, difficulty with movement and balance, vision and/or hearing loss, skin rashes, breathing problems, 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.

If symptoms have already appeared, treatment with biotin can reverse damage to the body already done by the disease. Vision loss, hearing loss, and developmental delay are irreversible.

PARTIAL BIOTINIDASE DEFICIENCY

People 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 may be asymptomatic until periods of illness or stress.

How common is Biotinidase Deficiency?

Profound biotinidase deficiency occurs in about 1 in 137,000 births. Studies report that the milder partial biotinidase deficiency occurs in about 1 in 110,000 people. Counsyl's internal data suggests that partial biotinidase deficiency 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. Biotin is non-toxic, so it is recommended that people with partial biotinidase deficiency also take biotin supplements.

If treatment is begun after symptoms appear, some symptoms, such as skin problems and hair loss, will disappear. If the disease has already caused irreversible hearing or vision loss, low vision aids or hearing aids may be helpful. Learning specialists can assist with any irreversible developmental deficits.

What is the prognosis for a person with Biotinidase Deficiency?

With early diagnosis and treatment, people with biotinidase deficiency can live completely normal lives with no symptoms. Those in whom the disease is not detected early may experience permanent damage to their hearing, vision, or intellect. In cases where the disease is entirely unrecognized, it can be life-threatening.

POSITIVE: CARRIER

Familial Mediterranean Fever

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

Gene: MEFV | Inheritance Pattern: Autosomal Recessive

Patient	DONOR 10280	No partner tested
Result	Carrier	N/A
Variant(s)	NM_000243.2(MEFV):c.2080A>G(M694V, aka MED) heterozygote	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of familial Mediterranean fever. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000243:1-10.	N/A

What is Familial Mediterranean Fever?

Familial Mediterranean fever (FMF) is an inherited condition which causes episodic attacks of fever and painful inflammation of the abdomen, chest, and joints. People with FMF may also develop a rash during these attacks. The attacks last for 1 to 3 days and can vary in severity. Between attacks, the person typically feels normal. These symptom-free periods can last for days or even years.

In 80-90% of people affected by FMF, the first attack occurs by the age of 20. Less commonly, symptoms begin later in life. Children who have FMF may experience periodic fever as their only symptom.

Some people with FMF develop a protein buildup in various parts of the body, notably the kidney. If left untreated, this can lead to life-threatening kidney failure. People who do not experience the characteristic attacks of FMF can still develop this particular form of kidney failure. This symptom is most common among people of Turkish and North African Jewish heritage, affecting 60% and 75% respectively.

Other symptoms that can occur during an attack of FMF include headache and inflammation of the heart and/or testicles. Affected people may also develop an inflammation of the membrane that surrounds the brain and spinal cord, though this is not usually serious or damaging. People with FMF who go untreated may experience decreased fertility.

About half of people with FMF have mild symptoms preceding an attack. These may include a mild, unpleasant sensation in parts of the body that will soon be affected or may consist of other physical and emotional symptoms.

How common is Familial Mediterranean Fever?

FMF is most common among ethnic groups from the Mediterranean region, notably people of Armenian, Arab, Turkish, Iraqi Jewish, and North African Jewish ancestry. One in every 200 to 1,000 people in these groups is affected by the disease and carrier rates in some populations have been estimated as high as 1 in 5.

Cases of FMF have also been found in other populations, including Italians, Greeks, Spaniards, Cypriots, and less commonly, Northern Europeans and Japanese.

How is Familial Mediterranean Fever treated?

There is no cure for FMF, however the drug colchicine has been very effective in preventing the disease's characteristic attacks. With daily doses of colchicine, 75% of people with FMF can avoid attacks with an additional 15% showing an improvement in their symptoms. Colchicine also prevents the dangerous buildup of proteins in the kidneys which could otherwise lead to kidney failure.

Episodic attacks of fever and inflammation can be treated with non-steroidal anti-inflammatory drugs. Those who do develop serious kidney failure may be helped by kidney transplantation.

What is the prognosis for a person with Familial Mediterranean Fever?

With early and regular treatment, people with FMF can live a normal lifespan and may even be symptom-free. The disease has the potential to be life-threatening only if the person is untreated (or does not respond to treatment) and develops kidney failure.

Methods and Limitations

DONOR 10280 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions.

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. 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. If *GJB2* is tested, two large upstream deletions which overlap *GJB6* and affect the expression of *GJB2*, *del(GJB6-D13S1830)* and *del(GJB6-D13S1854)*, are also analyzed. Mosaicism or somatic variants present at low levels may not be detected. If detected, these may not be reported.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect. Detection rates only account for analytical sensitivity and certain variants that have been previously described in the literature may not be reported if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease-specific rates of de novo mutations.

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

Spinal muscular atrophy

Targeted copy number analysis is used to determine the copy number of exon 7 of the *SMN1* gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of *SMN1* are carriers with two *SMN1* genes on one chromosome and a *SMN1* deletion on the other chromosome. This is more likely in individuals who have 2 copies of the *SMN1* gene and are positive for the g.27134T>G SNP, which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have 2 copies of *SMN1*.

Analysis of homologous regions

A combination of high-throughput sequencing, read depth-based copy number analysis, and targeted genotyping is used to determine the number of functional gene copies and/or the presence of selected loss of function mutations in certain genes that have homology to other regions. The precise breakpoints of large deletions in these genes cannot be determined, but are estimated from copy number analysis. High numbers of pseudogene copies may interfere with this analysis.

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

Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these may not be reported. 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. 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**.

LABORATORY DIRECTOR



H. Peter Kang, MD, MS, FCAP

Report content approved by Saurav Guha, PhD, FACMG on Sep 10, 2018

Conditions Tested

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

21-hydroxylase-deficient Congenital Adrenal Hyperplasia - 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, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Mixed or Other Caucasian 96%.

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

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

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

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

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

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

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

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

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

Argininemia - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001244438:1-8. **Detection Rate:** Mixed or Other Caucasian 97%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

COL4A3-related Alport Syndrome - Gene: COL4A3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000091:1-52. **Detection Rate:** Mixed or Other Caucasian 97%.

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

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

Congenital Disorder of Glycosylation Type Ib - Gene: MPI. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002435:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

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

Congenital Finnish Nephrosis - Gene: NPHS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004646:1-29. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

Delta-sarcoglycanopathy - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000337:2-9. **Detection Rate:** Mixed or Other Caucasian 99%.

Dysferlinopathy - Gene: DYSF. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001130987:1-56. **Detection Rate:** Mixed or Other Caucasian 98%.

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

ERCC6-related Disorders - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000124:2-21. **Detection Rate:** Mixed or Other Caucasian 99%.

ERCC8-related Disorders - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000082:1-12. **Detection Rate:** Mixed or Other Caucasian 95%.

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

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

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

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

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

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

Fanconi Anemia Type C - Gene: FANCC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000136:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

Galactokinase Deficiency - Gene: GALK1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000154:1-8. **Detection Rate:** Mixed or Other Caucasian 88%.

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

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. **Detection Rate:** Mixed or Other Caucasian 88%.

Gaucher Disease - Gene: GBA. Autosomal Recessive. Analysis of homologous regions. Variants (10): D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. **Detection Rate:** Mixed or Other Caucasian 60%.

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

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

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

Glutaric Acidemia Type 1 - Gene: GCDH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000159:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000518:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

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

Homocystinuria Caused by Cystathionine Beta-synthase Deficiency - Gene: CBS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000071:3-17. **Detection Rate:** Mixed or Other Caucasian >99%.

Hydroletharus Syndrome - Gene: HYL51. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_001134793:3. **Detection Rate:** Mixed or Other Caucasian >99%.

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

Inclusion Body Myopathy 2 - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001128227:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

Lipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000108:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

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

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

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

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

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

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

Mucopolysaccharidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032520:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.

Mucopolysaccharidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_020533:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

Muscle-eye-brain Disease - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017739:2-22. **Detection Rate:** Mixed or Other Caucasian 96%.

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

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

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

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

Niemann-Pick Disease Type C - Gene: NPC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000271:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.

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

Niemann-Pick Disease, SMPD1-associated - Gene: SMPD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000543:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

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

Northern Epilepsy - Gene: CLN8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_018941:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

PCCB-related Propionic Acidemia - Gene: PCCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001178014:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

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

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

PEX1-related Zellweger Syndrome Spectrum - Gene: PEX1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000466:1-24. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000152:2-20. **Detection Rate:** Mixed or Other Caucasian 98%.

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

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

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

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

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

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

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

Pyruvate Carboxylase Deficiency - Gene: PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_022172:2-21. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

Salla Disease - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012434:1-11. **Detection Rate:** Mixed or Other Caucasian 98%.

Sandhoff Disease - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000521:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

Segawa Syndrome - Gene: TH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000360:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.

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

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000382:1-10. **Detection Rate:** Mixed or Other Caucasian 97%.

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

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

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** Mixed or Other Caucasian 95%.

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

Sulfate Transporter-related Osteochondrodysplasia - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000112:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

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

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

USH1C-related Disorders - Gene: USH1C. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_153676:1-27. **Detection Rate:** Mixed or Other Caucasian >99%.

USH2A-related Disorders - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_206933:2-72. **Detection Rate:** Mixed or Other Caucasian 94%.

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

Very Long Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000018:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.

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

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

X-linked Alport Syndrome - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000495:1-51. **Detection Rate:** Mixed or Other Caucasian 95%.

X-linked Congenital Adrenal Hypoplasia - Gene: NROB1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. **Detection Rate:** Mixed or Other Caucasian 99%.

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

X-linked Myotubular Myopathy - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000252:2-15. **Detection Rate:** Mixed or Other Caucasian 98%.

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

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

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

Risk Calculations

Below are the risk calculations for all conditions tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

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

Disease	DONOR 10280 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,800	< 1 in 1,000,000
21-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 310,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Hyperinsulinism	1 in 11,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	< 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
ARSACS	< 1 in 44,000	< 1 in 1,000,000
Aspartylglycosaminuria	< 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 8,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 16,000	< 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
Beta-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Biotinidase Deficiency	NM_000060.2(BTD):c.1330G>C(D444H) heterozygote †	1 in 510
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	< 1 in 31,000	< 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 50,000	< 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 22,000	< 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 43,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 6,200	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 12,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 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 2,700	1 in 290,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000

Disease	DONOR 10280 Residual Risk	Reproductive Risk
Delta-sarcoglycanopathy	< 1 in 40,000	< 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 26,000	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 9,900	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,500	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	< 1 in 50,000	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 80,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	MED heterozygote †	1 in 2,000
Fanconi Anemia Complementation Group A	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia Type C	1 in 16,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 10,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	< 1 in 1,000,000
Gaucher Disease	N370S heterozygote †	1 in 450
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 3,200	1 in 420,000
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 Type 1	1 in 10,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 18,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 32,000	< 1 in 1,000,000
GRACILE Syndrome	< 1 in 50,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 15,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 5,000	1 in 990,000
Hereditary Fructose Intolerance	1 in 8,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 33,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency	1 in 25,000	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia, Autosomal Recessive	1 in 16,000	< 1 in 1,000,000
Inclusion Body Myopathy 2	< 1 in 50,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 25,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
KCNJ11-related Familial Hyperinsulinism	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 15,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	1 in 34,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 18,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 25,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 42,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 13,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 5,900	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 20,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	1 in 48,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucopolipidosis III Gamma	< 1 in 50,000	< 1 in 1,000,000
Mucopolipidosis IV	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10280 Residual Risk	Reproductive Risk
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	1 in 600,000	1 in 150,000
Mucopolysaccharidosis Type IIIA	1 in 12,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 25,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 37,000	< 1 in 1,000,000
Muscle-eye-brain Disease	< 1 in 12,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 6,700	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 1 in 1,000,000
Niemann-Pick Disease Type C	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-associated	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Northern Epilepsy	< 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 5,300	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	< 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
PEX1-related Zellweger Syndrome Spectrum	1 in 11,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 5,000	1 in 990,000
PKHD1-related Autosomal Recessive Polycystic Kidney Disease	1 in 6,100	< 1 in 1,000,000
Polyglandular Autoimmune Syndrome Type 1	1 in 14,000	< 1 in 1,000,000
Pompe Disease	1 in 6,300	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 11,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 35,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	< 1 in 1,000,000
Pycnodysostosis	< 1 in 50,000	< 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
Salla Disease	< 1 in 30,000	< 1 in 1,000,000
Sandhoff Disease	1 in 32,000	< 1 in 1,000,000
Segawa Syndrome	< 1 in 50,000	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	1 in 16,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	1 in 9,100	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 4,900	1 in 970,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
Sulfate Transporter-related Osteochondrodysplasia	1 in 11,000	< 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
Tyrosinemia Type I	1 in 17,000	< 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 50,000	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	1 in 8,800	< 1 in 1,000,000
Wilson Disease	1 in 8,600	< 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: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 09/10/2018

MALE
DONOR 10280
DOB: [REDACTED]
Ethnicity: Mixed or Other
Caucasian
Barcode: 11004212489042

FEMALE
N/A

Disease	DONOR 10280 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 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