



Client/Sending Facility:
Seattle Sperm Bank

4915 25th Ave Ne Ste 204
SEATTLE, WA 98105
Ph: (206)588-1484
Fax: (206) 466-4696 WAB-55

LCLS Specimen Number: 093-129-1758-0

Patient Name: 12314, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: (J18-1355 L

Indications: DONOR

Account Number: [REDACTED]

Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 04/03/2018

Date Received: 04/04/2018

Date Reported: 04/20/2018

Test: Chromosome, Blood, Routine

Cells Counted: 20

Cells Analyzed: 20

Cells Karyotyped: 2

Band Resolution: 500

CYTOGENETIC RESULT: 46,XY

INTERPRETATION: NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Chromosome analysis performed by LabCorp, CLIA 45D0674994. 7207 North Gessner Rd., Houston, TX 77040. Laboratory Director, Venkateswara R Potluri PhD.



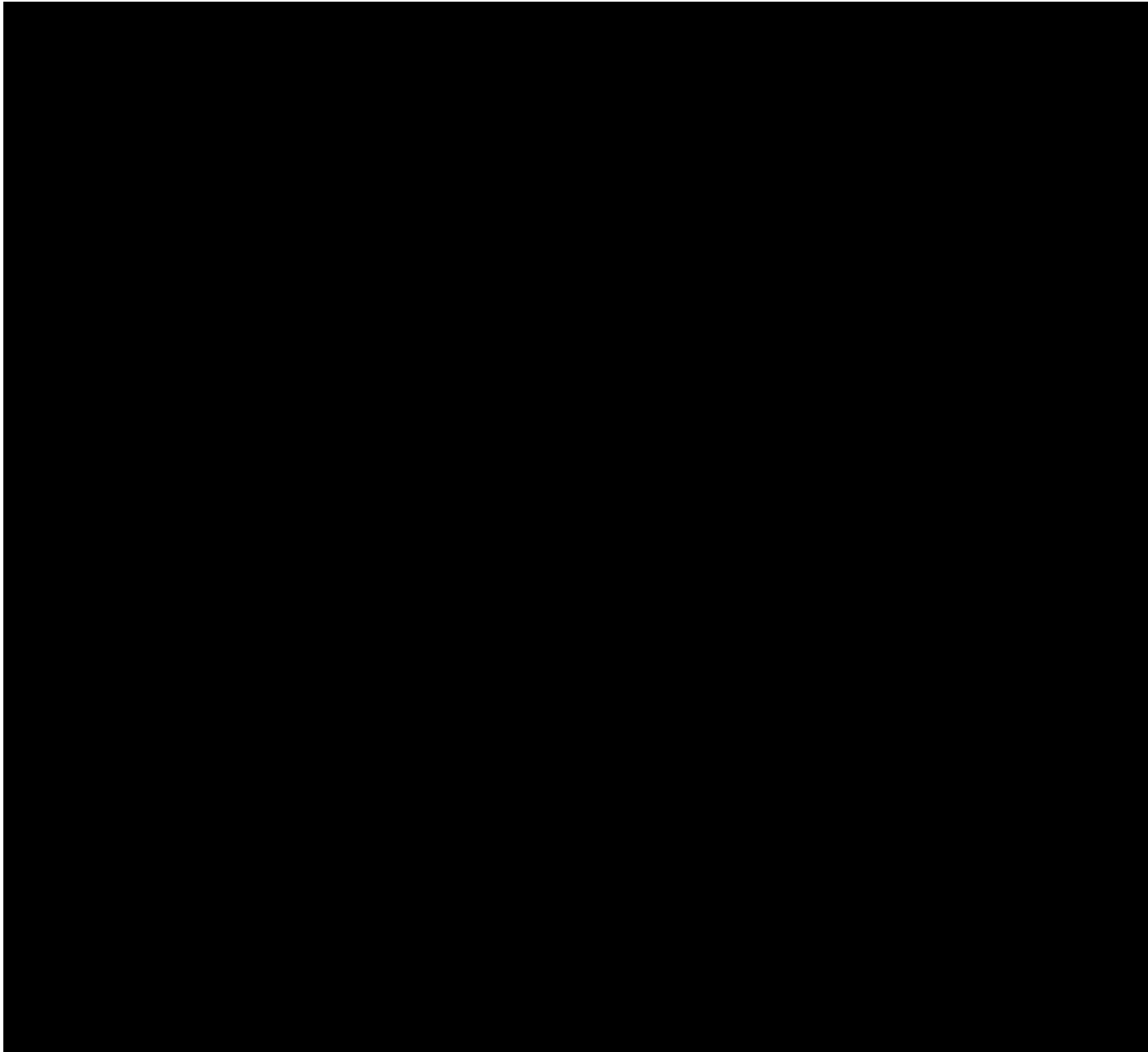
LabCorp Specialty Testing Group

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Hiba Risheg, PhD., FACMG

Patricia Kandalaft, MD
Medical Director
Stuart Schwartz, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
550 17th Ave. Suite 200, SEATTLE, WA, 98122-5789 (800) 676-8033

Professional Component performed by LabCorp/Dynacare CLIA 50D0632667, 550 17th Ave. Suite 200, Seattle WA 98122-5789. Medical Director, Patricia Kandalaft, MD
Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

This document contains private and confidential health information **protected by state and federal law.**



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: 04/10/2018

MALE
DONOR 12314
 DOB: [REDACTED]
 Ethnicity: Mixed or Other
 Caucasian
 Sample Type: EDTA Blood
 Date of Collection:
 Date Received: 04/04/2018
 Date Tested: 04/10/2018
 Barcode: 11004212274861
 Accession ID: CSLUDDRJDF4FYDZ
 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 12314 | Partner |
|--|--|--|
| Panel Information | Foresight Carrier Screen Universal Panel (175 conditions tested) | N/A |
| <p>POSITIVE: CARRIER 21-hydroxylase-deficient Congenital Adrenal Hyperplasia Reproductive Risk: 1 in 230 Inheritance: Autosomal Recessive</p> | <p>CARRIER* NM_000500.7(CYP21A2):c.844G>T (V282L, aka V281L) heterozygote</p> | <p>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".</p> |

*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 6.

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

21-hydroxylase-deficient Congenital Adrenal Hyperplasia

Reproductive risk: 1 in 230
 Risk before testing: 1 in 13,000

Gene: CYP21A2 | Inheritance Pattern: Autosomal Recessive

| | | |
|------------------------|---|--------------------------|
| Patient | DONOR 12314 | No partner tested |
| Result | + Carrier | N/A |
| Variants | NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) heterozygote | N/A |
| Methodology | Analysis of homologous regions | N/A |
| Interpretation | This individual is a carrier of 21-hydroxylase-deficient congenital adrenal hyperplasia. Carriers generally do not experience symptoms. NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) is a non-classic 21-hydroxylase-deficient congenital adrenal hyperplasia mutation. | N/A |
| Detection rate | 96% | N/A |
| Variants tested | CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. | N/A |

What is 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia?

Congenital adrenal hyperplasia (CAH) refers to a group of genetic disorders that affect the body's adrenal glands. The adrenal glands are located above each kidney and regulate essential functions in the body, including the production of several important hormones. CAH occurs when the adrenal glands are unable to produce these hormones properly, resulting in a hormone imbalance.

More than 90% of of CAH cases are caused by deficiency of the 21-hydroxylase enzyme. When this enzyme is missing or present at low levels, the adrenal glands are unable to produce two critical hormones, cortisol and aldosterone. The body responds to this deficiency by producing an excess of male sex hormones, called androgens. Collectively, the excess androgen production and hormone deficiencies can lead to a variety of medical problems, which vary in severity depending on the form of CAH.

There are two major forms of 21-hydroxylase-deficient CAH: classic CAH and non-classic CAH.

CLASSIC

The most severe form, referred to as classic CAH, can be divided into two different subtypes: the salt-wasting type and the simple virilizing type (non salt-wasting type). The classic salt-wasting type is associated with near to complete deficiency of the enzyme, 21-hydroxylase, resulting in the complete inability to produce the hormones, cortisol and aldosterone. In this type, the body cannot retain enough sodium (salt). When too much salt is lost in the urine, it may lead to dehydration, vomiting, diarrhea, failure to thrive, heart rhythm abnormalities (arrhythmias), and shock; if not properly treated, death may occur in some cases. In addition, female newborns often have external genitalia that do not clearly appear either male or female (ambiguous genitalia), whereas male newborns may present with enlarged genitalia. Signs of early puberty

(virilization) occur in both males and females with CAH. These symptoms may include: rapid growth and development in early childhood, but shorter than average height in adulthood, abnormal menstruation cycles for females, excess facial hair for females, early facial hair growth for males, severe acne, and infertility in both men and women.

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

NON-CLASSIC

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

How common is 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia?

The incidence of CAH varies by type and is more prevalent in certain ethnicities. Classic CAH occurs in approximately 1 in 15,000 births worldwide, while non-classic CAH is much more common, occurring in approximately 1 in 1,000 births. In some populations, namely individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent, the prevalence of the non-classic CAH can reach as high as 3-4 percent.

How is 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia treated?

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

Newborn females with ambiguous genitalia may need surgery to correct the function and appearance of the external genitalia. Surgery, if needed, is most often performed during infancy, but can be performed later in life.

Treatments provided during pregnancy may reduce the degree of virilization in female fetuses. However, because the long term safety of prenatal treatment is unknown, these therapies are considered experimental and are not recommended by professional guidelines.

What is the prognosis for a person with 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia?

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

Methods and Limitations

DONOR 12314 [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.



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 04/10/2018

MALE
DONOR 12314
DOB: [REDACTED]
Ethnicity: Mixed or Other
Caucasian
Barcode: 11004212274861

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. 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 Counsyl, 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

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Report content approved by Bethany Buckley, PhD, FACMG on Apr 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%.
- Arginemia** - 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 >99%.

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

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

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

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

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

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

Glutaric Acidemia 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%.

Hydrolethalus 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%.

Mucopolipidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM_032520:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.

Mucopolipidosis 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: NR0B1. X-linked Recessive. Sequencing with Copy Number Analysis. Exons: NM_000475:1-2. **Detection Rate:** Mixed or Other Caucasian 99%.

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

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

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

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

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

Risk Calculations

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

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

| Disease | DONOR 12314 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 | NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) heterozygote † | 1 in 230 |
| 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 | 1 in 13,000 | 1 in 650,000 |
| Bloom Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Calpainopathy | 1 in 13,000 | < 1 in 1,000,000 |
| Canavan Disease | < 1 in 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 |

| Disease | DONOR 12314 Residual Risk | Reproductive Risk |
|---|------------------------------|----------------------|
| D-bifunctional Protein Deficiency | 1 in 9,000 | < 1 in 1,000,000 |
| 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 | < 1 in 50,000 | < 1 in 1,000,000 |
| Fanconi Anemia Complementation Group A | 1 in 2,800 | < 1 in 1,000,000 |
| Fanconi Anemia 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 | 1 in 280 | 1 in 120,000 |
| 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, cbIA Type | < 1 in 50,000 | < 1 in 1,000,000 |
| Methylmalonic Acidemia, cbIB Type | 1 in 48,000 | < 1 in 1,000,000 |
| Methylmalonic Aciduria and Homocystinuria, cbIC 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 |

| Disease | DONOR 12314 Residual Risk | Reproductive Risk |
|---|--|----------------------------------|
| Mucopolysaccharidosis Type I | < 1 in 50,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type II | 1 in 16,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type IIIA | 1 in 600,000 | 1 in 150,000 |
| Mucopolysaccharidosis Type IIIB | 1 in 12,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type IIIC | 1 in 25,000 | < 1 in 1,000,000 |
| Muscle-eye-brain Disease | 1 in 37,000 | < 1 in 1,000,000 |
| MUT-related Methylmalonic Acidemia | < 1 in 12,000 | < 1 in 1,000,000 |
| MYO7A-related Disorders | 1 in 26,000 | < 1 in 1,000,000 |
| NEB-related Nemaline Myopathy | 1 in 15,000 | < 1 in 1,000,000 |
| Nephrotic Syndrome, NPHS2-related | < 1 in 6,700 | < 1 in 1,000,000 |
| Niemann-Pick Disease Type C | 1 in 35,000 | < 1 in 1,000,000 |
| Niemann-Pick Disease Type C2 | 1 in 19,000 | < 1 in 1,000,000 |
| Niemann-Pick Disease, SMPD1-associated | < 1 in 50,000 | < 1 in 1,000,000 |
| Nijmegen Breakage Syndrome | 1 in 25,000 | < 1 in 1,000,000 |
| Northern Epilepsy | 1 in 16,000 | < 1 in 1,000,000 |
| Ornithine Transcarbamylase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| PCCA-related Propionic Acidemia | < 1 in 1,000,000 | 1 in 140,000 |
| PCCB-related Propionic Acidemia | 1 in 4,200 | < 1 in 1,000,000 |
| PCDH15-related Disorders | 1 in 22,000 | < 1 in 1,000,000 |
| Pendred Syndrome | 1 in 5,300 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 3 | 1 in 7,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 4 | 1 in 44,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 5 | 1 in 9,300 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 6 | < 1 in 71,000 | < 1 in 1,000,000 |
| PEX1-related Zellweger Syndrome Spectrum | < 1 in 50,000 | < 1 in 1,000,000 |
| Phenylalanine Hydroxylase Deficiency | 1 in 11,000 | < 1 in 1,000,000 |
| PKHD1-related Autosomal Recessive Polycystic Kidney Disease | 1 in 5,000 | 1 in 990,000 |
| Polyglandular Autoimmune Syndrome Type 1 | 1 in 6,100 | < 1 in 1,000,000 |
| Pompe Disease | 1 in 14,000 | < 1 in 1,000,000 |
| PPT1-related Neuronal Ceroid Lipofuscinosis | 1 in 6,300 | < 1 in 1,000,000 |
| Primary Carnitine Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 1 | < 1 in 50,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 2 | 1 in 35,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 3 | < 1 in 50,000 | < 1 in 1,000,000 |
| PROP1-related Combined Pituitary Hormone Deficiency | 1 in 13,000 | < 1 in 1,000,000 |
| Pycnodysostosis | 1 in 11,000 | < 1 in 1,000,000 |
| Pyruvate Carboxylase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Rhizomelic Chondrodysplasia Punctata Type 1 | 1 in 25,000 | < 1 in 1,000,000 |
| RTEL1-related Disorders | 1 in 16,000 | < 1 in 1,000,000 |
| Salla Disease | < 1 in 50,000 | < 1 in 1,000,000 |
| Sandhoff Disease | < 1 in 30,000 | < 1 in 1,000,000 |
| Segawa Syndrome | 1 in 32,000 | < 1 in 1,000,000 |
| Short Chain Acyl-CoA Dehydrogenase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Sjogren-Larsson Syndrome | 1 in 16,000 | < 1 in 1,000,000 |
| Smith-Lemli-Opitz Syndrome | 1 in 9,100 | < 1 in 1,000,000 |
| Spastic Paraplegia Type 15 | 1 in 4,900 | 1 in 970,000 |
| Spinal Muscular Atrophy | < 1 in 50,000 Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770 | < 1 in 1,000,000 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 |



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 04/10/2018

MALE
DONOR 12314
DOB: [REDACTED]
Ethnicity: Mixed or Other
Caucasian
Barcode: 11004212274861

FEMALE
N/A

Disease

- X-linked Alport Syndrome
- X-linked Congenital Adrenal Hypoplasia
- X-linked Juvenile Retinoschisis
- X-linked Myotubular Myopathy
- X-linked Severe Combined Immunodeficiency
- Xeroderma Pigmentosum Group A
- Xeroderma Pigmentosum Group C

**DONOR 12314
Residual Risk**

- Not calculated
- < 1 in 1,000,000
- < 1 in 1,000,000
- Not calculated
- < 1 in 1,000,000
- < 1 in 50,000
- 1 in 7,300

**Reproductive
Risk**

- Not calculated
- < 1 in 1,000,000
- 1 in 50,000
- Not calculated
- 1 in 200,000
- < 1 in 1,000,000
- < 1 in 1,000,000