



RESULTS RECIPIENT
SEATTLE SPERM BANK
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 NPI: 1306838271
 Report Date: 11/26/2021

MALE
DONOR 12809
 DOB: [REDACTED]
 Ethnicity: Northern European
 Sample Type: EDTA Blood
 Date of Collection: 11/16/2021
 Date Received: 11/18/2021
 Date Tested: 11/24/2021
 Barcode: 11004513015676
 Accession ID:
 CSLK4FAG9WLKWYX
 Indication: Egg or sperm donor

FEMALE
 N/A

Foresight® Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

RESULTS SUMMARY

| Risk Details | DONOR 12809 | Partner |
|---|--|---|
| Panel Information | Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested) | N/A |
| POSITIVE: CARRIER Pompe Disease Reproductive Risk: 1 in 400 Inheritance: Autosomal Recessive | ⊕ CARRIER* NM_000152.3(GAA):c. -32-13T>G(aka IVS1-13T>G) 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 Glycine Encephalopathy, GLDC-related Reproductive Risk: 1 in 620 Inheritance: Autosomal Recessive | ⊕ CARRIER* NM_000170.2(GLDC):c.1652G>T (S551I) 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". |

†Likely to have a negative impact on gene function.

*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 9.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

POSITIVE: CARRIER

Pompe Disease

Reproductive risk: 1 in 400
 Risk before testing: 1 in 40,000

Gene: GAA | Inheritance Pattern: Autosomal Recessive

| Patient | DONOR 12809 | No partner tested |
|-----------------------|---|-------------------|
| Result | ⊕ Carrier | N/A |
| Variant(s) | NM_000152.3(GAA):c.-32-13T>G(aka IVS1-13T>G) heterozygote | N/A |
| Methodology | Sequencing with copy number analysis (v3.1) | N/A |
| Interpretation | This individual is a carrier of Pompe disease. Carriers generally do not experience symptoms. GAA:IVS1-13T>G is seen in 36% to 90% of late-onset Pompe disease and is not associated with the infantile-onset form. | N/A |
| Detection rate | 98% | N/A |
| Exons tested | NM_000152:2-20. | N/A |

What Is Pompe Disease?

Pompe disease also called glycogen storage disease type II, is an inherited disorder where the body fails to produce enough alpha-glucosidase (also called maltase), an enzyme needed to break down a type of sugar called glycogen. Without adequate amounts of alpha-glucosidase, glycogen builds up in the body, particularly in the muscles, and damages cells. Pompe disease is caused by mutations in the *GAA* gene. People with Pompe disease have muscle weakness that progresses over time, mainly in the muscles used for movement and breathing. The heart may also be affected. The level of alpha-glucosidase remaining is correlated to the severity of symptoms, the age of onset, and disease progression.

Pompe disease is separated into two forms, the infantile-onset form and the late-onset form. These forms are described below.

INFANTILE-ONSET FORM

Infantile-onset Pompe disease is the most severe form because alpha-glucosidase function is entirely absent. Muscle weakness and poor muscle tone causes infants to have trouble moving, holding up their heads, and feeding. They have trouble gaining weight and grow at a slower pace. Infants also have trouble breathing, which can worsen with lung infections. They typically have enlarged hearts, livers, and tongues. Disease progression is usually rapid, and the most common causes of death are heart or lung failure.

LATE-ONSET FORM

Late-onset Pompe disease is less severe because some alpha-glucosidase is still present. Symptoms start with muscle weakness and breathing problems. Some individuals with late-onset Pompe disease have heart problems but without an enlarged heart. They may eventually lose the ability to walk and require a wheelchair, and they may need mechanical assistance to breathe. Disease progression is more gradual, and the most common cause of death is lung failure.

How Common Is Pompe Disease?

The incidence of Pompe disease is approximately 1 in 100,000. Infantile-onset Pompe disease is the most common form.



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N/A

How Is Pompe Disease Treated?

The FDA has approved enzyme replacement therapy for both infantile-onset and late-onset Pompe disease. Enzyme replacement therapy can help maintain a healthy heart size and normal heart function and may also help improve muscle tone and strength. Individuals need to follow a protein-rich diet, attend physical therapy, and monitor and treat lung infections.

What Is the Prognosis for a Person with Pompe Disease?

In infantile-onset Pompe disease, symptoms may begin at birth but more often begin in the first few months of life. Patients typically die within the first year of life, although enzyme replacement therapy can now prolong life into early childhood. In late-onset Pompe disease, symptoms can begin at any age from childhood to adulthood, and the lifespan depends on how early symptoms begin. The most common cause of death in individuals with Pompe disease is lung failure.

POSITIVE: CARRIER

Glycine Encephalopathy, GLDC-related

Reproductive risk: 1 in 620

Risk before testing: 1 in 95,000

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

| Patient | DONOR 12809 | No partner tested |
|-----------------------|--|-------------------|
| Result | ⊕ Carrier | N/A |
| Variant(s) | NM_000170.2(GLDC):c.1652G>T(S551I) heterozygote † | N/A |
| Methodology | Sequencing with copy number analysis (v3.1) | N/A |
| Interpretation | This individual is a carrier of glycine encephalopathy, GLDC-related. Carriers generally do not experience symptoms. | N/A |
| Detection rate | 94% | N/A |
| Exons tested | NM_000170:1-25. | N/A |

†Likely to have a negative impact on gene function.

What is Glycine Encephalopathy, GLDC-related?

Glycine encephalopathy (GE), GLDC-related, also known as nonketotic hyperglycinemia (NKH), is a disease that impairs the body's ability to breakdown glycine, an amino acid found in proteins. Glycine encephalopathy is caused by harmful genetic changes (mutations) in the *GLDC* gene, which codes for the enzyme glycine decarboxylase (GLDC). Over time, if this enzyme is not working properly, glycine builds up to toxic levels in the brain, organs, and other body tissues. This can lead to lack of energy (lethargy), low muscle tone, breathing difficulties, coma, and often death. Individuals with GE typically have intellectual disability and seizures. The majority of individuals with encephalopathy present in the newborn period, but the age at which symptoms appear can vary. There are two forms of the disease: severe and attenuated.

SEVERE FORM

Approximately 85% of those with symptoms in the newborn period and 50% of those with onset between two weeks to three months will have the severe form of GE. Individuals with the severe form will not make any developmental progress and will have seizures that are difficult to control with medications. Most affected individuals will present in the newborn period with rapidly worsening symptoms. However, individuals with the severe form can also begin to show symptoms between two weeks to three months.

ATTENUATED FORM

Approximately 20% of all children affected with GE will have less severe symptoms (the attenuated form). Individuals who have the attenuated form will have variable progress with their development and may have seizures, but the seizures are typically more easily controlled with medications. While some individuals with the attenuated form will present in the newborn period, others may not show symptoms until after three months of age.

How common is Glycine Encephalopathy, GLDC-related?

Several genes are known to cause GE and approximately 80% of GE is caused by mutations in the *GLDC* gene. The incidence of glycine encephalopathy, GLDC-related is approximately 1/95,000, however, the incidence may be higher in certain populations, such as Finland and British Columbia.

How is Glycine Encephalopathy, GLDC-related treated?

There is no cure for GE. Treatment is aimed at lowering the levels of glycine in the body and controlling seizures. Glycine can be reduced through certain medications (e.g., sodium benzoate) and a low-protein diet. Anticonvulsant medications can be used to control seizures, but they may not be completely effective for all individuals. The health of individuals with GE is usually monitored by many different specialists.

What is the prognosis for an individual with Glycine Encephalopathy, GLDC-related?

Infants with the severe form of GE will not make any progress in major developmental milestones (such holding objects, sitting, crawling, walking, or talking). However, some children may learn to smile and roll. Certain skills may be lost as the child grows (such as bottle feeding and eye contact). Seizures will develop in the first year of life and are very difficult to treat with medications. Death in the first year of life is common.

Developmental progress varies in individuals with the attenuated form. Some children may learn to sit, walk, and communicate. Communication is most often non-verbal (such as using sign language). The majority of affected individuals will have moderate to severe intellectual disability and attend special-education classes in school. These children may develop movement disorders and behavioral problems. Individuals who have seizures typically respond to treatment. Affected individuals usually start showing symptoms before one year of age. However, for individuals who do start showing symptoms after one year of age, intellectual disability is typically milder and and seizures are uncommon.

Methods and Limitations

DONOR 12809 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

Sequencing with copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to analyze the genes listed in the Conditions Tested section of the report. Except where otherwise noted, the region of interest (ROI) comprises the indicated coding regions and 20 non-coding bases flanking each region. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected non-coding bases are excluded from the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. Select genes or regions for which pseudogenes or other regions of homology impede reliable variant detection may be assayed using alternate technology, or they may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with an average sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel and such deletions are detected with a sensitivity of $\geq 75\%$. Selected founder deletions may be detected at slightly higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from junction reads, where available, or using the positions of affected probes. Only exons known to be included in the region affected by a copy number variant are provided in the variant nomenclature. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed.

Spinal muscular atrophy

Targeted copy number analysis via high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. Other genetic variants may interfere with this analysis. Some individuals with two copies of *SMN1* are "silent" carriers with both *SMN1* genes on one chromosome and no copies of the gene on the other chromosome. This is more likely in individuals who have two copies of the *SMN1* gene and are positive for the g.27134T>G single-nucleotide polymorphism (SNP) (PMID: 9199562, 23788250, and 28676062), which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have two copies of *SMN1*.

Analysis of homologous regions

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

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a pathogenic variant may or may not be a carrier of 21-hydroxylase deficient CAH, depending on the chromosomal location of the variants (phase). Benign *CYP21A2* gene duplications and/or triplications will only be reported in this context. Some individuals with two functional *CYP21A2* gene copies may be "silent" carriers, with two gene copies resulting from a duplication on one chromosome and a gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay. Given that the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are based only on the published incidence for classic CAH. However, the published prevalence of non-classic CAH is highest in individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent. Therefore, the residual and reproductive risks are likely an underestimate for CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to identify sequence variation and functional gene copies within the region of interest (ROI) of *HBA1* and *HBA2*, which includes the listed exons plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. For large deletions or duplications in these genes, the precise breakpoints cannot be determined but are instead estimated from copy number analysis. This assay has been validated to detect up to two additional copies of each alpha globin gene. In rare instances where assay results suggest greater than two additional copies are present, this will be noted but the specific number of gene copies observed will not be provided.

Extensive sequence homology exists between *HBA1* and *HBA2*. This sequence homology can prevent certain variants from being localized to one gene over the other. In these instances, variant nomenclature will be provided for both genes. If follow-up testing is indicated for patients with the nomenclature provided for both genes, both *HBA1* and *HBA2* should be tested. Some individuals with four functional alpha globin gene copies may be "silent" carriers, with three gene copies resulting from triplication on one chromosome and a single gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay.

Interpretation of reported variants

The classification and interpretation of all variants identified in this assay reflects the current state of Myriad's scientific understanding at the time this report was issued. Variants are classified according to internally defined criteria, which are compatible with the ACMG Standards and Guidelines for the Interpretation of Sequence Variants (PMID: 25741868). Variants that have been determined by Myriad to be disease-causing or likely disease-causing (i.e. pathogenic or likely pathogenic) are reported. Benign variants, variants of uncertain clinical significance (VUS), and variants not directly associated with the specified disease phenotype(s) are not reported. Variant classification and interpretation may change for a variety of reasons, including but not limited to, improvements to classification techniques, availability of additional scientific information, and observation of a variant in more patients. If the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If an updated report is issued, the variants reported may change based on their current classification. This can include changes to the variants displayed in gene specific 'variants tested' sections. Healthcare providers may contact Myriad directly to request updated variant classification information specific to this test result.

Limitations

The MWH Foresight Carrier Screen is designed to detect and report germline (constitutional) alterations. Mosaic (somatic) variation may not be detected, and if it is detected, it may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes (phase). This test is not designed to detect sex-chromosome copy number variations. If present, sex-chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Variant interpretation and residual and reproductive risk estimations assume a normal karyotype and may be different for individuals with abnormal karyotypes. The test does not fully address all inherited forms of intellectual disability, birth defects, or heritable diseases. Furthermore, not all forms of genetic variation are detected by this assay (i.e., duplications [except in specified genes], chromosomal rearrangements, structural abnormalities, etc.). Additional testing may be appropriate for some individuals. Pseudogenes and other regions of homology may interfere with this analysis. In an unknown number of cases, other genetic variation may interfere with variant detection. Rare carrier states where complementary changes exist between the chromosomes may not be detected by the assay. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions, and technical or analytical errors.

Detection rates are determined using published scientific literature and/or reputable databases, when available, to estimate the fraction of disease alleles, weighted by frequency, that the methodology is predicted to be able or unable to detect. Detection rates are approximate and only account for analytical sensitivity. Certain variants that have been previously described in the literature may not be reported, if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease specific rates of *de novo* variation.

This test was developed, and its performance characteristics determined by, Myriad Women's Health, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: #05D1102604.



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N/A

Incidental Findings

Unless otherwise indicated, these results and interpretations are limited to the specific disease panel(s) requested by the ordering healthcare provider. In some cases, standard data analyses may identify genetic findings beyond the region(s) of interest specified by the test, and such findings may not be reported. These findings may include genomic abnormalities with major, minor, or no, clinical significance.

If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

Resources

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

Patients can share their reports using research registries such as Genome Connect, an online research registry building a genetics and health knowledge base. Genome Connect provides patients, physicians, and researchers an opportunity to share genetic information to support the study of the impact of genetic variation on health conditions.

SENIOR LABORATORY DIRECTOR

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

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Heather Labreche, PhD, FACMG, CGMBS on Nov 26, 2021

Conditions Tested

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

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

Alpha Thalassemia, HBA1/HBA2-related - Genes: HBA1, HBA2. Autosomal Recessive. Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis. Exons: NM_000517:1-3; NM_000558:1-3. Variants (16): -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40. **Detection Rate:** Not calculated due to rarity of disease in this individual's reported ethnicity.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Congenital Adrenal Hyperplasia, CYP21A2-related - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Gaucher Disease - Gene: GBA. Autosomal Recessive. Analysis of homologous regions. **Variants (10):** D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. **Detection Rate:** Northern European 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:** Northern European >99%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Hydrolethalus Syndrome - Gene: HYL51. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_145014:4. **Detection Rate:** Northern European >99%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Mucopolidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032520:1-11. **Detection Rate:** Northern European 98%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000152:2-20. **Detection Rate:** Northern European 98%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. **Detection Rate:** Northern European 95%.

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

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

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

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

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

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

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

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

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



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 11/26/2021

MALE
DONOR 12809
DOB: [REDACTED]
Ethnicity: Northern European
Barcode: 11004513015676

FEMALE
N/A

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

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

X-linked Adrenal Hypoplasia Congenita - Gene: NR0B1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. **Detection Rate:** Northern European 97%.

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

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

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

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

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

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

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

Risk Calculations

Below are the risk calculations for all conditions tested. Negative results do not rule out the possibility of being a carrier. Residual risk is an estimate of each patient's post-test likelihood of being a carrier, while the reproductive risk represents an estimated likelihood that the patients' future children could inherit each disease. These risks are inherent to all carrier-screening tests, may vary by ethnicity, are predicated on a negative family history, and are present even given a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. In addition, average carrier rates are estimated using incidence or prevalence data from published scientific literature and/or reputable databases, where available, and are incorporated into residual risk calculations for each population/ethnicity. When population-specific data is not available for a condition, average worldwide incidence or prevalence is used. Further, incidence and prevalence data are only collected for the specified phenotypes (which include primarily the classic or severe forms of disease) and may not include alternate or milder disease manifestations associated with the gene. Actual incidence rates, prevalence rates, and carrier rates, and therefore actual residual risks, may be higher or lower than the estimates provided. Carrier rates, incidence/prevalence, and/or residual risks are not provided for some genes with biological or heritable properties that would make these estimates inaccurate. A '+' symbol indicates a positive result. See the full clinical report for interpretation and details. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

| Disease | DONOR 12809 Residual Risk | Reproductive Risk |
|--|------------------------------|-------------------|
| 6-pyruvoyl-tetrahydropterin Synthase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Adenosine Deaminase Deficiency | 1 in 22,000 | < 1 in 1,000,000 |
| Alpha Thalassemia, HBA1/HBA2-related | Alpha globin status: aa/aa. | Not calculated |
| Alpha-mannosidosis | 1 in 35,000 | < 1 in 1,000,000 |
| Alpha-sarcoglycanopathy | < 1 in 50,000 | < 1 in 1,000,000 |
| Alstrom Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Andermann Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Argininemia | 1 in 12,000 | < 1 in 1,000,000 |
| Argininosuccinic Aciduria | 1 in 15,000 | < 1 in 1,000,000 |
| Aspartylglucosaminuria | < 1 in 50,000 | < 1 in 1,000,000 |
| Ataxia with Vitamin E Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Ataxia-telangiectasia | 1 in 4,200 | < 1 in 1,000,000 |
| ATP7A-related Disorders | < 1 in 1,000,000 | 1 in 250,000 |
| Autoimmune Polyglandular Syndrome Type 1 | 1 in 15,000 | < 1 in 1,000,000 |
| Autosomal Recessive Osteopetrosis Type 1 | 1 in 8,900 | < 1 in 1,000,000 |
| Autosomal Recessive Polycystic Kidney Disease, PKHD1-related | 1 in 8,100 | < 1 in 1,000,000 |
| Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay | < 1 in 44,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS1-related | 1 in 32,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS10-related | 1 in 42,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS12-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS2-related | < 1 in 50,000 | < 1 in 1,000,000 |
| BCS1L-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| Beta-sarcoglycanopathy | 1 in 39,000 | < 1 in 1,000,000 |
| Biotinidase Deficiency | 1 in 13,000 | 1 in 650,000 |
| Bloom Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Calpainopathy | 1 in 13,000 | < 1 in 1,000,000 |
| Canavan Disease | 1 in 9,700 | < 1 in 1,000,000 |
| Carbamoylphosphate Synthetase I Deficiency | < 1 in 57,000 | < 1 in 1,000,000 |
| Carnitine Palmitoyltransferase IA Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Carnitine Palmitoyltransferase II Deficiency | 1 in 25,000 | < 1 in 1,000,000 |
| Cartilage-hair Hypoplasia | < 1 in 50,000 | < 1 in 1,000,000 |
| Cerebrotendinous Xanthomatosis | 1 in 11,000 | < 1 in 1,000,000 |
| Citrullinemia Type 1 | 1 in 14,000 | < 1 in 1,000,000 |
| CLN3-related Neuronal Ceroid Lipofuscinosis | 1 in 8,600 | < 1 in 1,000,000 |
| CLN5-related Neuronal Ceroid Lipofuscinosis | < 1 in 50,000 | < 1 in 1,000,000 |
| CLN8-related Neuronal Ceroid Lipofuscinosis | < 1 in 50,000 | < 1 in 1,000,000 |
| Cohen Syndrome | < 1 in 15,000 | < 1 in 1,000,000 |
| COL4A3-related Alport Syndrome | 1 in 3,400 | < 1 in 1,000,000 |
| COL4A4-related Alport Syndrome | 1 in 35,000 | < 1 in 1,000,000 |
| Combined Pituitary Hormone Deficiency, PROP1-related | 1 in 6,100 | < 1 in 1,000,000 |
| Congenital Adrenal Hyperplasia, CYP11B1-related | 1 in 8,400 | < 1 in 1,000,000 |
| Congenital Adrenal Hyperplasia, CYP21A2-related | 1 in 1,300 | 1 in 280,000 |
| Congenital Disorder of Glycosylation Type Ia | 1 in 16,000 | < 1 in 1,000,000 |
| Congenital Disorder of Glycosylation Type Ic | < 1 in 50,000 | < 1 in 1,000,000 |
| Congenital Disorder of Glycosylation, MPI-related | < 1 in 50,000 | < 1 in 1,000,000 |

| Disease | DONOR 12809 Residual Risk | Reproductive Risk |
|---|---|-------------------|
| Costeff Optic Atrophy Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Cystic Fibrosis | 1 in 3,000 | 1 in 360,000 |
| Cystinosis | 1 in 22,000 | < 1 in 1,000,000 |
| D-bifunctional Protein Deficiency | 1 in 9,000 | < 1 in 1,000,000 |
| Delta-sarcoglycanopathy | < 1 in 13,000 | < 1 in 1,000,000 |
| Dihydroipoamide Dehydrogenase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Dysferlinopathy | 1 in 11,000 | < 1 in 1,000,000 |
| Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) | Not calculated | Not calculated |
| ERCC6-related Disorders | 1 in 8,500 | < 1 in 1,000,000 |
| ERCC8-related Disorders | < 1 in 16,000 | < 1 in 1,000,000 |
| EVC-related Ellis-van Creveld Syndrome | 1 in 7,800 | < 1 in 1,000,000 |
| EVC2-related Ellis-van Creveld Syndrome | 1 in 9,800 | < 1 in 1,000,000 |
| Fabry Disease | < 1 in 1,000,000 | 1 in 220,000 |
| Familial Dysautonomia | < 1 in 50,000 | < 1 in 1,000,000 |
| Familial Hyperinsulinism, ABCC8-related | 1 in 17,000 | < 1 in 1,000,000 |
| Familial Hyperinsulinism, KCNJ11-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Familial Mediterranean Fever | 1 in 11,000 | < 1 in 1,000,000 |
| Fanconi Anemia Complementation Group A | 1 in 2,800 | < 1 in 1,000,000 |
| Fanconi Anemia, FANCC-related | < 1 in 50,000 | < 1 in 1,000,000 |
| FKRP-related Disorders | 1 in 16,000 | < 1 in 1,000,000 |
| FKTN-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| Free Sialic Acid Storage Disorders | < 1 in 30,000 | < 1 in 1,000,000 |
| Galactokinase Deficiency | 1 in 37,000 | < 1 in 1,000,000 |
| Galactosemia | 1 in 8,600 | < 1 in 1,000,000 |
| Gamma-sarcoglycanopathy | 1 in 3,300 | < 1 in 1,000,000 |
| Gaucher Disease | 1 in 260 | 1 in 110,000 |
| GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness | 1 in 2,500 | 1 in 260,000 |
| GLB1-related Disorders | 1 in 17,000 | < 1 in 1,000,000 |
| Glutaric Acidemia, GCDH-related | 1 in 16,000 | < 1 in 1,000,000 |
| Glycine Encephalopathy, AMT-related | 1 in 26,000 | < 1 in 1,000,000 |
| Glycine Encephalopathy, GLDC-related | NM_000170.2(GLDC):c.1652G>T(S551I) heterozygote † | 1 in 620 |
| Glycogen Storage Disease Type Ia | 1 in 8,700 | < 1 in 1,000,000 |
| Glycogen Storage Disease Type Ib | 1 in 35,000 | < 1 in 1,000,000 |
| Glycogen Storage Disease Type III | 1 in 16,000 | < 1 in 1,000,000 |
| GNE Myopathy | 1 in 23,000 | < 1 in 1,000,000 |
| GNPTAB-related Disorders | 1 in 20,000 | < 1 in 1,000,000 |
| HADHA-related Disorders | 1 in 20,000 | < 1 in 1,000,000 |
| Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) | 1 in 3,700 | 1 in 560,000 |
| Hereditary Fructose Intolerance | 1 in 7,900 | < 1 in 1,000,000 |
| Hexosaminidase A Deficiency (Including Tay-Sachs Disease) | 1 in 30,000 | < 1 in 1,000,000 |
| HMG-CoA Lyase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Holocarboxylase Synthetase Deficiency | 1 in 15,000 | < 1 in 1,000,000 |
| Homocystinuria, CBS-related | 1 in 9,400 | < 1 in 1,000,000 |
| Hydrolethals Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Hypophosphatasia | 1 in 30,000 | < 1 in 1,000,000 |
| Isovaleric Acidemia | 1 in 32,000 | < 1 in 1,000,000 |
| Joubert Syndrome 2 | < 1 in 50,000 | < 1 in 1,000,000 |
| Junctional Epidermolysis Bullosa, LAMA3-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Junctional Epidermolysis Bullosa, LAMB3-related | 1 in 32,000 | < 1 in 1,000,000 |
| Junctional Epidermolysis Bullosa, LAMC2-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Krabbe Disease | 1 in 14,000 | < 1 in 1,000,000 |
| Leigh Syndrome, French-Canadian Type | < 1 in 50,000 | < 1 in 1,000,000 |
| Lipoid Congenital Adrenal Hyperplasia | < 1 in 50,000 | < 1 in 1,000,000 |
| Lysosomal Acid Lipase Deficiency | 1 in 14,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type Ia | 1 in 39,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type Ib | 1 in 39,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type II | 1 in 16,000 | < 1 in 1,000,000 |
| Medium Chain Acyl-CoA Dehydrogenase Deficiency | 1 in 4,400 | 1 in 790,000 |
| Megalencephalic Leukoencephalopathy with Subcortical Cysts | < 1 in 50,000 | < 1 in 1,000,000 |
| Metachromatic Leukodystrophy | 1 in 16,000 | < 1 in 1,000,000 |
| Methylmalonic Acidemia, cblA Type | < 1 in 50,000 | < 1 in 1,000,000 |

| Disease | DONOR 12809 Residual Risk | Reproductive Risk |
|--|---|-------------------|
| Methylmalonic Acidemia, cblB Type | 1 in 48,000 | < 1 in 1,000,000 |
| Methylmalonic Acidemia, MMUT-related | 1 in 26,000 | < 1 in 1,000,000 |
| Methylmalonic Aciduria and Homocystinuria, cblC Type | 1 in 16,000 | < 1 in 1,000,000 |
| MKS1-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| Mucopolipidosis III Gamma | < 1 in 20,000 | < 1 in 1,000,000 |
| Mucopolipidosis IV | < 1 in 50,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type I | 1 in 16,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type II | < 1 in 1,000,000 | 1 in 300,000 |
| Mucopolysaccharidosis Type IIIA | 1 in 19,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type IIIB | 1 in 27,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type IIIC | < 1 in 50,000 | < 1 in 1,000,000 |
| Muscular Dystrophy, LAMA2-related | 1 in 5,700 | < 1 in 1,000,000 |
| MYO7A-related Disorders | 1 in 15,000 | < 1 in 1,000,000 |
| NEB-related Nemaline Myopathy | 1 in 1,200 | 1 in 400,000 |
| Nephrotic Syndrome, NPHS1-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Nephrotic Syndrome, NPHS2-related | 1 in 35,000 | < 1 in 1,000,000 |
| Neuronal Ceroid Lipofuscinosis, CLN6-related | 1 in 20,000 | < 1 in 1,000,000 |
| Niemann-Pick Disease Type C1 | 1 in 19,000 | < 1 in 1,000,000 |
| Niemann-Pick Disease Type C2 | < 1 in 50,000 | < 1 in 1,000,000 |
| Niemann-Pick Disease, SMPD1-related | 1 in 25,000 | < 1 in 1,000,000 |
| Nijmegen Breakage Syndrome | 1 in 16,000 | < 1 in 1,000,000 |
| Ornithine Transcarbamylase Deficiency | < 1 in 1,000,000 | 1 in 140,000 |
| PCCA-related Propionic Acidemia | 1 in 4,200 | < 1 in 1,000,000 |
| PCCB-related Propionic Acidemia | 1 in 22,000 | < 1 in 1,000,000 |
| PCDH15-related Disorders | 1 in 3,300 | < 1 in 1,000,000 |
| Pendred Syndrome | 1 in 8,200 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 1 | 1 in 16,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 3 | 1 in 44,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 4 | 1 in 9,300 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 5 | < 1 in 71,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 6 | < 1 in 50,000 | < 1 in 1,000,000 |
| Phenylalanine Hydroxylase Deficiency | 1 in 4,800 | 1 in 940,000 |
| POMGNT-related Disorders | < 1 in 12,000 | < 1 in 1,000,000 |
| Pompe Disease | IVS1-13T>G heterozygote † | 1 in 400 |
| PPT1-related Neuronal Ceroid Lipofuscinosis | 1 in 7,700 | < 1 in 1,000,000 |
| Primary Carnitine Deficiency | 1 in 11,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 1 | 1 in 17,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 2 | < 1 in 50,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 3 | 1 in 13,000 | < 1 in 1,000,000 |
| Pycnodysostosis | 1 in 43,000 | < 1 in 1,000,000 |
| Pyruvate Carboxylase Deficiency | 1 in 25,000 | < 1 in 1,000,000 |
| Rhizomelic Chondrodysplasia Punctata Type 1 | 1 in 16,000 | < 1 in 1,000,000 |
| RTEL1-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| Sandhoff Disease | 1 in 18,000 | < 1 in 1,000,000 |
| Short-chain Acyl-CoA Dehydrogenase Deficiency | 1 in 11,000 | < 1 in 1,000,000 |
| Sjogren-Larsson Syndrome | < 1 in 12,000 | < 1 in 1,000,000 |
| SLC26A2-related Disorders | 1 in 16,000 | < 1 in 1,000,000 |
| Smith-Lemli-Opitz Syndrome | 1 in 9,400 | < 1 in 1,000,000 |
| Spastic Paraplegia Type 15 | < 1 in 50,000 | < 1 in 1,000,000 |
| Spinal Muscular Atrophy | Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770 | 1 in 110,000 |
| Spondylothoracic Dysostosis | < 1 in 50,000 | < 1 in 1,000,000 |
| TGM1-related Autosomal Recessive Congenital Ichthyosis | 1 in 22,000 | < 1 in 1,000,000 |
| TPP1-related Neuronal Ceroid Lipofuscinosis | 1 in 30,000 | < 1 in 1,000,000 |
| Tyrosine Hydroxylase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Tyrosinemia Type I | 1 in 16,000 | < 1 in 1,000,000 |
| Tyrosinemia Type II | 1 in 25,000 | < 1 in 1,000,000 |
| USH1C-related Disorders | 1 in 30,000 | < 1 in 1,000,000 |
| USH2A-related Disorders | 1 in 4,100 | < 1 in 1,000,000 |
| Usher Syndrome Type 3 | 1 in 41,000 | < 1 in 1,000,000 |
| Very-long-chain Acyl-CoA Dehydrogenase Deficiency | 1 in 18,000 | < 1 in 1,000,000 |



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 11/26/2021

MALE
DONOR 12809
 DOB: [REDACTED]
 Ethnicity: Northern European
 Barcode: 11004513015676

FEMALE
 N/A

| Disease | DONOR 12809 Residual Risk | Reproductive Risk |
|---|------------------------------|-------------------|
| Wilson Disease | 1 in 6,500 | < 1 in 1,000,000 |
| X-linked Adrenal Hypoplasia Congenita | < 1 in 1,000,000 | < 1 in 1,000,000 |
| X-linked Adrenoleukodystrophy | 1 in 90,000 | 1 in 42,000 |
| X-linked Alport Syndrome | Not calculated | Not calculated |
| X-linked Juvenile Retinoschisis | < 1 in 1,000,000 | 1 in 40,000 |
| X-linked Myotubular Myopathy | Not calculated | Not calculated |
| X-linked Severe Combined Immunodeficiency | < 1 in 1,000,000 | 1 in 200,000 |
| Xeroderma Pigmentosum Group A | < 1 in 50,000 | < 1 in 1,000,000 |
| Xeroderma Pigmentosum Group C | 1 in 7,300 | < 1 in 1,000,000 |