



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
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 NPI: 1306838271
 Report Date: 07/29/2020

MALE
DONOR 10478
 FEMALE
 N/A
 DOB: [REDACTED]
 Ethnicity: Southern European
 Sample Type: EDTA Blood
 Date of Collection: 07/17/2020
 Date Received: 07/18/2020
 Date Tested: 07/28/2020
 Barcode: 11004512663142
 Accession ID: CSLQDGEJA4EDPEF
 Indication: Egg or sperm donor

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 10478 | Partner |
|---|--|---|
| Panel Information | Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested) | N/A |
| POSITIVE: CARRIER Nephrotic Syndrome, NPHS2-related Reproductive Risk: 1 in 110,000 Inheritance: Autosomal Recessive | CARRIER* NM_014625.2(NPHS2):c. 686G>A(R229Q) heterozygote | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |

*Carriers generally do not experience symptoms.

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

Nephrotic Syndrome, NPHS2-related

Reproductive risk: 1 in 110,000

Risk before testing: 1 in 310,000

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

| Patient | DONOR 10478 | No partner tested |
|-----------------------|--|-------------------|
| Result | Carrier | N/A |
| Variant(s) | NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote | N/A |
| Methodology | Sequencing with copy number analysis | N/A |
| Interpretation | This individual is a carrier of nephrotic syndrome, NPHS2-related. Carriers generally do not experience symptoms. The pathogenicity of R229Q is dependent on the variant observed on the other chromosome. | N/A |
| Detection rate | >99% | N/A |
| Exons tested | NM_014625:1-8. | N/A |

What Is Nephrotic Syndrome, NPHS2-Related?

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

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

How Common Is Nephrotic Syndrome, NPHS2-Related?

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

How Is Nephrotic Syndrome, NPHS2-Related Treated?

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



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

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

Methods and Limitations

DONOR 10478 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions (DTS v3.1).

Sequencing with copy number analysis

High-throughput sequencing and read depth-based copy number analysis are used to analyze the listed exons, as well as selected intergenic and intronic regions, of the genes in the Conditions Tested section of the report. The region of interest (ROI) of the test comprises these regions, in addition to the 20 intronic bases flanking each exon. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to high coverage and the sequences are compared to standards and references of normal variation (Genome Reference Consortium Human Build 37 (GRCh37)/hg19). More than 99% of all bases in the ROI are sequenced at greater than the minimum read depth. Mutations may not be detected in areas of lower sequence coverage. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes may be addressed by a different method. *CFTR* and *DMD* testing includes analysis for both large (exon-level) deletions and duplications with an average sensitivity of 99%, while other genes are only analyzed for large deletions with a sensitivity of >75%. However, the sensitivity may be higher for selected founder deletions. The breakpoints of copy number variants and exons affected are estimated from probe positions. Only exons known to be included in the copy number variant are provided in the name. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, 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.



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Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. This test is not designed to detect sex chromosome copy number variations. If present, sex chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Residual and reproductive risks provided assume a normal karyotype. Risks for individuals with abnormal karyotypes may be different. The test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet. Gynecol. 2007;109:229-37*).

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

Resources

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

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

SENIOR LABORATORY DIRECTOR

Jack Ji, PhD, FACMG

Report content approved by Lulu Mao, PhD, DABMGG on Jul 29, 2020

Conditions Tested

- 11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia** - Gene: CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000497:1-9. **Detection Rate:** Southern European 94%.
- 6-pyruvoyl-tetrahydropterin Synthase Deficiency** - Gene: PTS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000317:1-6. **Detection Rate:** Southern European >99%.
- ABCC8-related Familial Hyperinsulinism** - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000352:1-39. **Detection Rate:** Southern European >99%.
- Adenosine Deaminase Deficiency** - Gene: ADA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000022:1-12. **Detection Rate:** Southern European >99%.
- Alpha Thalassemia, HBA1/HBA2-related** - Genes: HBA1, HBA2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, anti3.7, anti4.2, del HS-40. **Detection Rate:** Southern European 90%.
- Alpha-mannosidosis** - Gene: MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000528:1-23. **Detection Rate:** Southern European >99%.
- Alpha-sarcoglycanopathy** - Gene: SGCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000023:1-9. **Detection Rate:** Southern European >99%.
- Alstrom Syndrome** - Gene: ALMS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015120:1-23. **Detection Rate:** Southern European >99%.
- AMT-related Glycine Encephalopathy** - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000481:1-9. **Detection Rate:** Southern European >99%.
- Andermann Syndrome** - Gene: SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_133647:1-25. **Detection Rate:** Southern European >99%.
- Argininemia** - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000045:1-8. **Detection Rate:** Southern European 97%.
- Argininosuccinic Aciduria** - Gene: ASL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001024943:1-16. **Detection Rate:** Southern European >99%.
- Aspartylglucosaminuria** - Gene: AGA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000027:1-9. **Detection Rate:** Southern European >99%.
- Ataxia with Vitamin E Deficiency** - Gene: TTPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000370:1-5. **Detection Rate:** Southern European >99%.
- Ataxia-telangiectasia** - Gene: ATM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000051:2-63. **Detection Rate:** Southern European 99%.
- ATP7A-related Disorders** - Gene: ATP7A. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000052:2-23. **Detection Rate:** Southern European 96%.
- Autoimmune Polyglandular Syndrome Type 1** - Gene: AIRE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000383:1-14. **Detection Rate:** Southern European >99%.
- Autosomal Recessive Osteopetrosis Type 1** - Gene: TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006019:2-20. **Detection Rate:** Southern European >99%.
- Autosomal Recessive Polycystic Kidney Disease, PKHD1-related** - Gene: PKHD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_138694 2-67. **Detection Rate:** Southern 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:** Southern European 99%.
- Bardet-Biedl Syndrome, BBS1-related** - Gene: BBS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024649:1-17. **Detection Rate:** Southern European >99%.
- Bardet-Biedl Syndrome, BBS10-related** - Gene: BBS10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024685:1-2. **Detection Rate:** Southern European >99%.
- Bardet-Biedl Syndrome, BBS12-related** - Gene: BBS12. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_152618:2. **Detection Rate:** Southern European >99%.
- Bardet-Biedl Syndrome, BBS2-related** - Gene: BBS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_031885:1-17. **Detection Rate:** Southern European >99%.
- BCS1L-related Disorders** - Gene: BCS1L. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004328:3-9. **Detection Rate:** Southern European >99%.
- Beta-sarcoglycanopathy** - Gene: SGCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000232:1-6. **Detection Rate:** Southern European >99%.
- Biotinidase Deficiency** - Gene: BTD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000060:1-4. **Detection Rate:** Southern European >99%.
- Bloom Syndrome** - Gene: BLM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000057:2-22. **Detection Rate:** Southern European >99%.
- Calpainopathy** - Gene: CAPN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000070:1-24. **Detection Rate:** Southern European >99%.
- Canavan Disease** - Gene: ASPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000049:1-6. **Detection Rate:** Southern European 98%.
- Carbamoylphosphate Synthetase I Deficiency** - Gene: CPS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001875:1-38. **Detection Rate:** Southern European >99%.
- Carnitine Palmitoyltransferase IA Deficiency** - Gene: CPT1A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001876:2-19. **Detection Rate:** Southern European >99%.
- Carnitine Palmitoyltransferase II Deficiency** - Gene: CPT2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000098:1-5. **Detection Rate:** Southern European >99%.
- Cartilage-hair Hypoplasia** - Gene: RMRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NR_003051:1. **Detection Rate:** Southern European >99%.
- Cerebrotendinous Xanthomatosis** - Gene: CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000784:1-9. **Detection Rate:** Southern European >99%.
- Citrullinemia Type 1** - Gene: ASS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000050:3-16. **Detection Rate:** Southern European >99%.
- CLN3-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001042432 2-16. **Detection Rate:** Southern European >99%.
- CLN5-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006493:1-4. **Detection Rate:** Southern European >99%.
- CLN6-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017882:1-7. **Detection Rate:** Southern European >99%.

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

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

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

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

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

Congenital Adrenal Hyperplasia, CYP21A2-related - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. Variants (13): CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Southern European 96%.

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

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

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

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

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

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

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

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

Dysferlinopathy - Gene: DYSF. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003494:1-55. **Detection Rate:** Southern 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:** Southern European >99%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. **Detection Rate:** Southern European 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:** Southern 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:** Southern European >99%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 07/29/2020

MALE
DONOR 10478
DOB: [REDACTED]
Ethnicity: Southern European
Barcode: 11004512663142

FEMALE
N/A

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

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

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

X-linked Congenital Adrenal Hypoplasia - Gene: NR0B1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. **Detection Rate:** Southern European 99%.

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

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

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

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

Xeroderma Pigmentosum Group C - Gene: XPC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004628:1-16. **Detection Rate:** Southern European 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 10478 Residual Risk | Reproductive Risk |
|--|------------------------------|-------------------|
| 11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia | 1 in 3,800 | < 1 in 1,000,000 |
| 6-pyruvoyl-tetrahydropterin Synthase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| ABCC8-related Familial Hyperinsulinism | 1 in 17,000 | < 1 in 1,000,000 |
| Adenosine Deaminase Deficiency | 1 in 39,000 | < 1 in 1,000,000 |
| Alpha Thalassemia, HBA1/HBA2-related | Alpha globin status: aa/aa. | Not calculated |
| Alpha-mannosidosis | 1 in 35,000 | < 1 in 1,000,000 |
| Alpha-sarcoglycanopathy | 1 in 29,000 | < 1 in 1,000,000 |
| Alstrom Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| AMT-related Glycine Encephalopathy | 1 in 22,000 | < 1 in 1,000,000 |
| Andermann Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Argininemia | < 1 in 17,000 | < 1 in 1,000,000 |
| Argininosuccinic Aciduria | 1 in 13,000 | < 1 in 1,000,000 |
| Aspartylglucosaminuria | < 1 in 50,000 | < 1 in 1,000,000 |
| Ataxia with Vitamin E Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Ataxia-telangiectasia | 1 in 18,000 | < 1 in 1,000,000 |
| ATP7A-related Disorders | < 1 in 1,000,000 | 1 in 600,000 |
| Autoimmune Polyglandular Syndrome Type 1 | 1 in 18,000 | < 1 in 1,000,000 |
| Autosomal Recessive Osteopetrosis Type 1 | 1 in 35,000 | < 1 in 1,000,000 |
| Autosomal Recessive Polycystic Kidney Disease, PKHD1-related | 1 in 8,100 | < 1 in 1,000,000 |
| Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay | < 1 in 44,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS1-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Bardet-Biedl Syndrome, BBS10-related | < 1 in 50,000 | < 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 17,000 | 1 in 990,000 |
| 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 20,000 | < 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 28,000 | < 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 |
| 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 6,200 | < 1 in 1,000,000 |
| COL4A4-related Alport Syndrome | 1 in 13,000 | < 1 in 1,000,000 |
| Combined Pituitary Hormone Deficiency, PROP1-related | 1 in 6,100 | < 1 in 1,000,000 |
| Congenital Adrenal Hyperplasia, CYP21A2-related | 1 in 1,600 | 1 in 430,000 |
| Congenital Disorder of Glycosylation Type Ia | 1 in 16,000 | < 1 in 1,000,000 |
| Congenital Disorder of Glycosylation Type Ic | < 1 in 50,000 | < 1 in 1,000,000 |
| Congenital Disorder of Glycosylation, MPI-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Costeff Optic Atrophy Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 07/29/2020

MALE
DONOR 10478
 DOB: [REDACTED]
 Ethnicity: Southern European
 Barcode: 11004512663142

FEMALE
 N/A

| Disease | DONOR 10478 Residual Risk | Reproductive Risk |
|---|------------------------------|-------------------|
| Cystic Fibrosis | 1 in 2,700 | 1 in 290,000 |
| Cystinosis | 1 in 22,000 | < 1 in 1,000,000 |
| D-bifunctional Protein Deficiency | 1 in 9,000 | < 1 in 1,000,000 |
| Delta-sarcoglycanopathy | < 1 in 40,000 | < 1 in 1,000,000 |
| Dihydrofolate 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 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 10,000 | < 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 19,000 | < 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 31,000 | < 1 in 1,000,000 |
| Galactosemia | 1 in 11,000 | < 1 in 1,000,000 |
| Gamma-sarcoglycanopathy | 1 in 3,000 | < 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 19,000 | < 1 in 1,000,000 |
| GLDC-related Glycine Encephalopathy | 1 in 2,800 | < 1 in 1,000,000 |
| Glutaric Acidemia, GCDH-related | 1 in 14,000 | < 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 |
| GNE Myopathy | < 1 in 50,000 | < 1 in 1,000,000 |
| GNPTAB-related Disorders | 1 in 32,000 | < 1 in 1,000,000 |
| HADHA-related Disorders | 1 in 25,000 | < 1 in 1,000,000 |
| Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) | 1 in 1,300 | 1 in 74,000 |
| Hereditary Fructose Intolerance | 1 in 7,900 | < 1 in 1,000,000 |
| Herlitz Junctional Epidermolysis Bullosa, LAMB3-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Hexosaminidase A Deficiency (Including Tay-Sachs Disease) | 1 in 30,000 | < 1 in 1,000,000 |
| HMG-CoA Lyase Deficiency | 1 in 10,000 | < 1 in 1,000,000 |
| Holocarboxylase Synthetase Deficiency | 1 in 15,000 | < 1 in 1,000,000 |
| Homocystinuria, CBS-related | 1 in 17,000 | < 1 in 1,000,000 |
| Hydroletharus Syndrome | < 1 in 50,000 | < 1 in 1,000,000 |
| Hypophosphatasia | 1 in 27,000 | < 1 in 1,000,000 |
| Isovaleric Acidemia | 1 in 22,000 | < 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, LAMC2-related | < 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 14,000 | < 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 |
| 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 Ia | 1 in 16,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type Ib | 1 in 21,000 | < 1 in 1,000,000 |
| Maple Syrup Urine Disease Type II | 1 in 11,000 | < 1 in 1,000,000 |
| Medium Chain Acyl-CoA Dehydrogenase Deficiency | 1 in 6,100 | < 1 in 1,000,000 |
| Megalencephalic Leukoencephalopathy with Subcortical Cysts | < 1 in 50,000 | < 1 in 1,000,000 |
| Metachromatic Leukodystrophy | 1 in 16,000 | < 1 in 1,000,000 |
| Methylmalonic Acidemia, cblA Type | < 1 in 50,000 | < 1 in 1,000,000 |
| Methylmalonic Acidemia, cblB Type | < 1 in 50,000 | < 1 in 1,000,000 |
| Methylmalonic Aciduria and Homocystinuria, cblC Type | 1 in 16,000 | < 1 in 1,000,000 |

| Disease | DONOR 10478 Residual Risk | Reproductive Risk |
|--|---|-------------------|
| MKS1-related Disorders | < 1 in 50,000 | < 1 in 1,000,000 |
| Mucopolidosis III Gamma | < 1 in 50,000 | < 1 in 1,000,000 |
| Mucopolidosis IV | < 1 in 50,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type I | 1 in 16,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type II | < 1 in 1,000,000 | 1 in 300,000 |
| Mucopolysaccharidosis Type IIIA | 1 in 16,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type IIIB | 1 in 18,000 | < 1 in 1,000,000 |
| Mucopolysaccharidosis Type IIIC | 1 in 43,000 | < 1 in 1,000,000 |
| MUT-related Methylmalonic Acidemia | 1 in 18,000 | < 1 in 1,000,000 |
| MYO7A-related Disorders | 1 in 15,000 | < 1 in 1,000,000 |
| NEB-related Nemaline Myopathy | 1 in 1,200 | 1 in 400,000 |
| Nephrotic Syndrome, NPHS1-related | < 1 in 50,000 | < 1 in 1,000,000 |
| Nephrotic Syndrome, NPHS2-related | NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote † | 1 in 110,000 |
| Niemann-Pick Disease Type C1 | 1 in 11,000 | < 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 15,000 | < 1 in 1,000,000 |
| Ornithine Transcarbamylase Deficiency | < 1 in 1,000,000 | 1 in 140,000 |
| PCCA-related Propionic Acidemia | 1 in 4,200 | < 1 in 1,000,000 |
| PCCB-related Propionic Acidemia | 1 in 22,000 | < 1 in 1,000,000 |
| PCDH15-related Disorders | 1 in 3,300 | < 1 in 1,000,000 |
| Pendred Syndrome | 1 in 6,400 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 1 | 1 in 16,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 3 | 1 in 44,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 4 | 1 in 9,300 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 5 | < 1 in 71,000 | < 1 in 1,000,000 |
| Peroxisome Biogenesis Disorder Type 6 | < 1 in 50,000 | < 1 in 1,000,000 |
| Phenylalanine Hydroxylase Deficiency | 1 in 4,100 | 1 in 690,000 |
| POMGNT-related Disorders | < 1 in 12,000 | < 1 in 1,000,000 |
| Pompe Disease | 1 in 15,000 | < 1 in 1,000,000 |
| PPT1-related Neuronal Ceroid Lipofuscinosis | 1 in 7,700 | < 1 in 1,000,000 |
| Primary Carnitine Deficiency | 1 in 16,000 | < 1 in 1,000,000 |
| Primary Hyperoxaluria Type 1 | 1 in 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 32,000 | < 1 in 1,000,000 |
| Short-chain Acyl-CoA Dehydrogenase Deficiency | 1 in 9,700 | < 1 in 1,000,000 |
| Sjogren-Larsson Syndrome | < 1 in 12,000 | < 1 in 1,000,000 |
| SLC26A2-related Disorders | 1 in 16,000 | < 1 in 1,000,000 |
| Smith-Lemli-Opitz Syndrome | 1 in 9,400 | < 1 in 1,000,000 |
| Spastic Paraplegia Type 15 | < 1 in 50,000 | < 1 in 1,000,000 |
| Spinal Muscular Atrophy | Negative for g.27134T>G SNP SMN1: 2 copies 1 in 890 | 1 in 200,000 |
| Spondylothoracic Dysostosis | < 1 in 50,000 | < 1 in 1,000,000 |
| TGM1-related Autosomal Recessive Congenital Ichthyosis | 1 in 22,000 | < 1 in 1,000,000 |
| TPP1-related Neuronal Ceroid Lipofuscinosis | 1 in 30,000 | < 1 in 1,000,000 |
| Tyrosine Hydroxylase Deficiency | < 1 in 50,000 | < 1 in 1,000,000 |
| Tyrosinemia Type I | 1 in 16,000 | < 1 in 1,000,000 |
| Tyrosinemia Type II | 1 in 25,000 | < 1 in 1,000,000 |
| USH1C-related Disorders | 1 in 35,000 | < 1 in 1,000,000 |
| USH2A-related Disorders | 1 in 2,200 | < 1 in 1,000,000 |
| Usher Syndrome Type 3 | 1 in 41,000 | < 1 in 1,000,000 |
| Very-long-chain Acyl-CoA Dehydrogenase Deficiency | 1 in 20,000 | < 1 in 1,000,000 |
| Wilson Disease | 1 in 9,000 | < 1 in 1,000,000 |
| X-linked Adrenoleukodystrophy | 1 in 120,000 | 1 in 56,000 |
| X-linked Alport Syndrome | Not calculated | Not calculated |



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 07/29/2020

MALE
DONOR 10478
DOB: [REDACTED]
Ethnicity: Southern European
Barcode: 11004512663142

FEMALE
N/A

| Disease | DONOR 10478 Residual Risk | Reproductive Risk |
|---|------------------------------|-------------------|
| X-linked Congenital Adrenal Hypoplasia | < 1 in 1,000,000 | < 1 in 1,000,000 |
| X-linked Juvenile Retinoschisis | < 1 in 1,000,000 | 1 in 40,000 |
| X-linked Myotubular Myopathy | Not calculated | Not calculated |
| X-linked Severe Combined Immunodeficiency | < 1 in 1,000,000 | 1 in 200,000 |
| Xeroderma Pigmentosum Group A | < 1 in 50,000 | < 1 in 1,000,000 |
| Xeroderma Pigmentosum Group C | 1 in 7,300 | < 1 in 1,000,000 |