



RESULTS RECIPIENT  
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 Report Date: 03/19/2021

MALE  
**DONOR 14223**  
 DOB:  
 Ethnicity: Northern European  
 Sample Type: EDTA Blood  
 Date of Collection: 03/08/2021  
 Date Received: 03/11/2021  
 Date Tested: 03/19/2021  
 Barcode: 11004512824944  
 Accession ID: CSL642FJY96LEUA  
 Indication: Egg or sperm donor

FEMALE  
 N/A

# Foresight® Carrier Screen

**NEGATIVE**

## 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 14223  | Partner |
|--|--|---------|
| Panel Information  | Foresight Carrier Screen<br>Universal Panel<br>Fundamental Plus Panel<br>Fundamental Panel<br><b>(175 conditions tested)</b> | N/A     |
| <b>All conditions tested</b><br>A complete list of all conditions tested can be found on page 5. | <input type="checkbox"/> <b>NEGATIVE</b><br>No disease-causing mutations were detected.                                      | N/A     |

## CLINICAL NOTES

- None

## NEXT STEPS

- If necessary, patients can discuss residual risks with their physician or a genetic counselor.

## Methods and Limitations

**DONOR 14223 [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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DOB:  
Ethnicity: Northern European  
Barcode: 11004512824944

FEMALE  
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.

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### SENIOR LABORATORY DIRECTOR

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

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Lulu Mao, PhD, DABMGG on Mar 19, 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%.

**Dihydroipoamide 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%.

**GLDC-related Glycine Encephalopathy** - Gene: GLDC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000170:1-25. **Detection Rate:** Northern European 94%.

**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%.

**Glycogen Storage Disease Type Ia** - Gene: G6PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000151:1-5. **Detection Rate:** Northern European >99%.

**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: HYLS1. 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 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%.

**Mucopolipidosis III Gamma** - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_032520:1-11. **Detection Rate:** Northern European 98%.

**Mucopolipidosis 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%.

**MUT-related Methylmalonic Acidemia** - Gene: MUT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000255:2-13. **Detection Rate:** Northern European >99%.

**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: 03/19/2021

MALE  
DONOR 14223  
DOB:  
Ethnicity: Northern European  
Barcode: 11004512824944

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 14223<br>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  |



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
 Attn: Jeffrey Olliffe  
 NPI: 1306838271  
 Report Date: 03/19/2021

MALE  
**DONOR 14223**  
 DOB:  
 Ethnicity: Northern European  
 Barcode: 11004512824944

FEMALE  
 N/A

| Disease   | DONOR 14223<br>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  |
| GLDC-related Glycine Encephalopathy   | 1 in 2,800                   | < 1 in 1,000,000  |
| Glutaric Acidemia, GCDH-related   | 1 in 16,000                  | < 1 in 1,000,000  |
| Glycine Encephalopathy, AMT-related   | 1 in 26,000                  | < 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 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  |



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
 Attn: Jeffrey Olliffe  
 NPI: 1306838271  
 Report Date: 03/19/2021

MALE  
**DONOR 14223**  
 DOB:  
 Ethnicity: Northern European  
 Barcode: 11004512824944

FEMALE  
 N/A

| Disease  | DONOR 14223<br>Residual Risk  | Reproductive Risk |
|--|-------------------------------|-------------------|
| Methylmalonic Acidemia, cblB Type                      | 1 in 48,000                   | < 1 in 1,000,000  |
| Methylmalonic Aciduria and Homocystinuria, cblC Type   | 1 in 16,000                   | < 1 in 1,000,000  |
| MKS1-related Disorders                                 | < 1 in 50,000                 | < 1 in 1,000,000  |
| Mucopolipidosis III Gamma                              | < 1 in 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  |
| MUT-related Methylmalonic Acidemia                     | 1 in 26,000                   | < 1 in 1,000,000  |
| MYO7A-related Disorders                                | 1 in 15,000                   | < 1 in 1,000,000  |
| NEB-related Nermaline 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  | 1 in 4,000                    | < 1 in 1,000,000  |
| PPT1-related Neuronal Ceroid Lipofuscinosis            | 1 in 7,700                    | < 1 in 1,000,000  |
| Primary Carnitine Deficiency                           | 1 in 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                                | SMN1: 3+ copies<br>1 in 4,800 | 1 in 670,000      |
| 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  |
| Wilson Disease   | 1 in 6,500                    | < 1 in 1,000,000  |



RESULTS RECIPIENT  
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MALE  
**DONOR 14223**  
 DOB:  
 Ethnicity: Northern European  
 Barcode: 11004512824944

FEMALE  
 N/A

| Disease                                   | DONOR 14223<br>Residual Risk | Reproductive Risk |
|---|------------------------------|-------------------|
| 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  |