



RESULTS RECEIPT  
**SEATTLE SPERM BANK**  
 Attn: Dr. Jeffrey Olliffe  
 4915 25th Ave NE, Suite 204W  
 Seattle, WA 98105  
 Phone: (206) 588-1484  
 Fax: (206) 466-4696  
 NPI: 1306838271  
 Report Date: 01/24/2018

MATERNAL  
**DONOR 12280**  
 DOB: [REDACTED]  
 Ethnicity: Northern European  
 Sample Type: EDTA Blood  
 Date of Collection:  
 Date Received: 01/17/2018  
 Date Tested: 01/24/2018  
 Barcode: 11004212320531  
 Accession ID: CSLRJFYERJFZY2V  
 Indication: Egg or sperm donor

FEMALE  
 N/A

# Foresight™ Carrier Screen

**NEGATIVE**

## ABOUT THIS TEST

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

## RESULTS SUMMARY

Risk Details	DONOR 12280	Partner
Panel Information	Foresight Carrier Screen Universal Panel (175 conditions tested)	N/A
<b>All conditions tested</b> A complete list of all conditions tested can be found on page 4.	<input type="checkbox"/> <b>NEGATIVE</b> 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 12280** [Foresight Carrier Screen]: sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions.

### Sequencing with copy number analysis

High-throughput sequencing and read depth-based copy number analysis are used to analyze the listed exons, as well as selected intergenic and intronic regions, of the genes in the Conditions Tested section of the report. The region of interest (ROI) of the test comprises these regions, in addition to the 20 intronic bases flanking each exon. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to high coverage and the sequences are compared to standards and references of normal variation. More than 99% of all bases in the ROI are sequenced at greater than the minimum read depth. Mutations may not be detected in areas of lower sequence coverage. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes may be addressed by a different method. *CFTR* and *DMD* testing includes analysis for both large (exon-level) deletions and duplications with an average sensitivity of 99%, while other genes are only analyzed for large deletions with a sensitivity of >75%. However, the sensitivity may be higher for selected founder deletions. If *GJB2* is tested, two large upstream deletions which overlap *GJB6* and affect the expression of *GJB2*, *del(GJB6-D13S1830)* and *del(GJB6-D13S1854)*, are also analyzed. Mosaicism or somatic variants present at low levels may not be detected. If detected, these may not be reported.

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

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

### Spinal muscular atrophy

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

### Analysis of homologous regions

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

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

## Limitations

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

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

---

### LAB DIRECTORS



H. Peter Kang, MD, MS, FCAP

# 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:** Northern European 94%.

**21-hydroxylase-deficient Congenital Adrenal Hyperplasia** - Gene: CYP21A2. Autosomal Recessive. Analysis of Homologous Regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs\*21, I173N, L308FfsX6, P31L, Q319\*, Q319\*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.

**6-pyruvoyl-tetrahydropterin Synthase Deficiency** - Gene: PTS. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000317:1-6. **Detection Rate:** Northern European >99%.

**ABCC8-related Hyperinsulinism** - Gene: ABCC8. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000352:1-39. **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 >99%.

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

**Alpha-mannosidosis** - Gene: MAN2B1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000528:1-23. **Detection Rate:** 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%.

**AMT-related Glycine Encephalopathy** - Gene: AMT. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000481:1-9. **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\_001244438:1-8. **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%.

**ARSACS** - Gene: SACS. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_014363:2-10. **Detection Rate:** Northern European 99%.

**Aspartylglycosaminuria** - 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 98%.

**ATP7A-related Disorders** - Gene: ATP7A. X-linked Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000052:2-23. **Detection Rate:** Northern European 96%.

**Autosomal Recessive Osteopetrosis Type 1** - Gene: TCIRG1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_006019:2-20. **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%.

**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%.

**CLN6-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN6. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_017882:1-7. **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 97%.

**COL4A4-related Alport Syndrome** - Gene: COL4A4. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000092:2-48. **Detection Rate:** Northern European 98%.

**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 Ib** - Gene: MPI. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_002435:1-8. **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 Finnish Nephrosis** - Gene: NPHS1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_004646:1-29. **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 99%.

**Dysferlinopathy** - Gene: DYSF. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_001130987:1-56. **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 99%.

**ERCC8-related Disorders** - Gene: ERCC8. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000082:1-12. **Detection Rate:** Northern European 95%.

**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 >99%.

**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: IKBKAP. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_003640:2-37. **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 Type C** - 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%.

**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 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:** 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 Type 1** - Gene: GCDH. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000159:2-12. **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%.

**GNPTAB-related Disorders** - Gene: GNPTAB. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_024312:1-21. **Detection Rate:** Northern European >99%.

**GRACILE Syndrome** - Gene: BCS1L. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_004328:3-9. **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%.

**Herlitz Junctional Epidermolysis Bullosa, LAMA3-related** - Gene: LAMA3. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000227:1-38. **Detection Rate:** Northern European >99%.

**Herlitz Junctional Epidermolysis Bullosa, LAMB3-related** - Gene: LAMB3. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000228:2-23. **Detection Rate:** Northern European >99%.

**Herlitz Junctional Epidermolysis Bullosa, LAMC2-related** - Gene: LAMC2. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_005562:1-23. **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 98%.

**Holocarboxylase Synthetase Deficiency** - Gene: HLCS. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000411:4-12. **Detection Rate:** Northern European >99%.

**Homocystinuria Caused by Cystathionine Beta-synthase Deficiency** - Gene: CBS. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000071:3-17. **Detection Rate:** Northern European >99%.

**Hydroletharus Syndrome** - Gene: HYL51. Autosomal Recessive. Sequencing with Copy Number Analysis. Exon: NM\_001134793:3. **Detection Rate:** Northern European >99%.

**Hypophosphatase, Autosomal Recessive** - Gene: ALPL. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000478:2-12. **Detection Rate:** Northern European >99%.

**Inclusion Body Myopathy 2** - Gene: GNE. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_001128227:1-12. **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%.

**KCNJ11-related Familial Hyperinsulinism** - Gene: KCNJ11. Autosomal Recessive. Sequencing with Copy Number Analysis. Exon: NM\_000525:1. 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%.

**LAMA2-related Muscular Dystrophy** - Gene: LAMA2. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000426:1-65. 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%.

**Lipoamide Dehydrogenase Deficiency** - Gene: DLD. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000108:1-14. 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 >99%.

**Maple Syrup Urine Disease Type 1B** - Gene: BCKDHB. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_183050:1-10. Detection Rate: Northern European >99%.

**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 II** - Gene: DBT. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_001918:1-11. Detection Rate: Northern European 96%.

**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%.

**Mucopolysaccharidosis III Gamma** - Gene: GNPTG. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_032520:1-11. Detection Rate: Northern European >99%.

**Mucopolysaccharidosis 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 88%.

**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%.

**Muscle-eye-brain Disease** - Gene: POMGNT1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_017739:2-22. Detection Rate: Northern European 96%.

**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%.

**Niemann-Pick Disease Type C** - 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-associated** - 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%.

**Northern Epilepsy** - Gene: CLN8. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_018941:2-3. 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\_001178014:1-16. 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 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%.

**PEX1-related Zellweger Syndrome Spectrum** - Gene: PEX1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000466:1-24. 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%.

**PKHD1-related Autosomal Recessive Polycystic Kidney Disease** - Gene: PKHD1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_138694:2-67. Detection Rate: Northern European >99%.

**Polyglandular Autoimmune Syndrome Type 1** - Gene: AIRE. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000383:1-14. Detection Rate: Northern European >99%.

**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%.

**PROP1-related Combined Pituitary Hormone Deficiency** - Gene: PROP1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_006261:1-3. **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\_022172:2-21. **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%.

**Salla Disease** - Gene: SLC17A5. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_012434:1-11. **Detection Rate:** Northern European 98%.

**Sandhoff Disease** - Gene: HEXB. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000521:1-14. **Detection Rate:** Northern European >99%.

**Segawa Syndrome** - Gene: TH. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000360:1-13. **Detection Rate:** Northern European >99%.

**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 97%.

**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%.

**Steroid-resistant Nephrotic Syndrome** - Gene: NPHS2. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_014625:1-8. **Detection Rate:** Northern European >99%.

**Sulfate Transporter-related Osteochondrodysplasia** - Gene: SLC26A2. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000112:2-3. **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%.

**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\_153676:1-27. **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 94%.

**Usher Syndrome Type 3** - Gene: CLRN1. Autosomal Recessive. Sequencing with Copy Number Analysis. Exons: NM\_174878:1-3. **Detection Rate:** Northern 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:** 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 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 95%.

**X-linked Congenital Adrenal Hypoplasia** - Gene: NR0B1. X-linked Recessive. Sequencing with Copy Number Analysis. Exons: NM\_000475:1-2. **Detection Rate:** Northern European 99%.

**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 98%.

**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. 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.

Disease	DONOR 12280 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,800	< 1 in 1,000,000
21-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 310,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Hyperinsulinism	1 in 11,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
AMT-related Glycine Encephalopathy	1 in 22,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	< 1 in 17,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 13,000	< 1 in 1,000,000
ARSACS	< 1 in 44,000	< 1 in 1,000,000
Aspartylglycosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 8,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 650,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	< 1 in 31,000	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	< 1 in 50,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 12,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 22,000	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN6-related Neuronal Ceroid Lipofuscinosis	1 in 43,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 6,200	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 12,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 2,700	1 in 290,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 40,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000

Disease	DONOR 12280 Residual Risk	Reproductive Risk
<b>Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)</b>	Not calculated	Not calculated
<b>ERCC6-related Disorders</b>	1 in 26,000	< 1 in 1,000,000
<b>ERCC8-related Disorders</b>	< 1 in 9,900	< 1 in 1,000,000
<b>EVC-related Ellis-van Creveld Syndrome</b>	1 in 7,500	< 1 in 1,000,000
<b>EVC2-related Ellis-van Creveld Syndrome</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Fabry Disease</b>	< 1 in 1,000,000	1 in 80,000
<b>Familial Dysautonomia</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Familial Mediterranean Fever</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Fanconi Anemia Complementation Group A</b>	1 in 2,800	< 1 in 1,000,000
<b>Fanconi Anemia Type C</b>	1 in 16,000	< 1 in 1,000,000
<b>FKRP-related Disorders</b>	1 in 16,000	< 1 in 1,000,000
<b>FKTN-related Disorders</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Galactokinase Deficiency</b>	1 in 10,000	< 1 in 1,000,000
<b>Galactosemia</b>	1 in 8,600	< 1 in 1,000,000
<b>Gamma-sarcoglycanopathy</b>	1 in 3,000	< 1 in 1,000,000
<b>Gaucher Disease</b>	1 in 280	1 in 120,000
<b>GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness</b>	1 in 3,200	1 in 420,000
<b>GLB1-related Disorders</b>	1 in 19,000	< 1 in 1,000,000
<b>GLDC-related Glycine Encephalopathy</b>	1 in 2,800	< 1 in 1,000,000
<b>Glutaric Acidemia Type 1</b>	1 in 10,000	< 1 in 1,000,000
<b>Glycogen Storage Disease Type Ia</b>	1 in 18,000	< 1 in 1,000,000
<b>Glycogen Storage Disease Type Ib</b>	1 in 35,000	< 1 in 1,000,000
<b>Glycogen Storage Disease Type III</b>	1 in 16,000	< 1 in 1,000,000
<b>GNPTAB-related Disorders</b>	1 in 32,000	< 1 in 1,000,000
<b>GRACILE Syndrome</b>	< 1 in 50,000	< 1 in 1,000,000
<b>HADHA-related Disorders</b>	1 in 15,000	< 1 in 1,000,000
<b>Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)</b>	1 in 5,000	1 in 990,000
<b>Hereditary Fructose Intolerance</b>	1 in 8,000	< 1 in 1,000,000
<b>Herlitz Junctional Epidermolysis Bullosa, LAMA3-related</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Herlitz Junctional Epidermolysis Bullosa, LAMB3-related</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Herlitz Junctional Epidermolysis Bullosa, LAMC2-related</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Hexosaminidase A Deficiency (Including Tay-Sachs Disease)</b>	1 in 30,000	< 1 in 1,000,000
<b>HMG-CoA Lyase Deficiency</b>	< 1 in 33,000	< 1 in 1,000,000
<b>Holocarboxylase Synthetase Deficiency</b>	1 in 15,000	< 1 in 1,000,000
<b>Homocystinuria Caused by Cystathionine Beta-synthase Deficiency</b>	1 in 25,000	< 1 in 1,000,000
<b>Hydrolethalus Syndrome</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Hypophosphatasia, Autosomal Recessive</b>	1 in 16,000	< 1 in 1,000,000
<b>Inclusion Body Myopathy 2</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Isovaleric Acidemia</b>	1 in 25,000	< 1 in 1,000,000
<b>Joubert Syndrome 2</b>	< 1 in 50,000	< 1 in 1,000,000
<b>KCNJ11-related Familial Hyperinsulinism</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Krabbe Disease</b>	1 in 15,000	< 1 in 1,000,000
<b>LAMA2-related Muscular Dystrophy</b>	1 in 34,000	< 1 in 1,000,000
<b>Leigh Syndrome, French-Canadian Type</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Lipoamide Dehydrogenase Deficiency</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Lipoid Congenital Adrenal Hyperplasia</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Lysosomal Acid Lipase Deficiency</b>	1 in 18,000	< 1 in 1,000,000
<b>Maple Syrup Urine Disease Type 1B</b>	1 in 25,000	< 1 in 1,000,000
<b>Maple Syrup Urine Disease Type Ia</b>	1 in 42,000	< 1 in 1,000,000
<b>Maple Syrup Urine Disease Type II</b>	1 in 13,000	< 1 in 1,000,000
<b>Medium Chain Acyl-CoA Dehydrogenase Deficiency</b>	1 in 5,900	< 1 in 1,000,000
<b>Megalencephalic Leukoencephalopathy with Subcortical Cysts</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Metachromatic Leukodystrophy</b>	1 in 20,000	< 1 in 1,000,000
<b>Methylmalonic Acidemia, cblA Type</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Methylmalonic Acidemia, cblB Type</b>	1 in 48,000	< 1 in 1,000,000
<b>Methylmalonic Aciduria and Homocystinuria, cblC Type</b>	1 in 16,000	< 1 in 1,000,000
<b>MKS1-related Disorders</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Mucopolysaccharidosis III Gamma</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Mucopolysaccharidosis IV</b>	< 1 in 50,000	< 1 in 1,000,000
<b>Mucopolysaccharidosis Type I</b>	1 in 16,000	< 1 in 1,000,000
<b>Mucopolysaccharidosis Type II</b>	1 in 600,000	1 in 150,000

Disease	DONOR 12280 Residual Risk	Reproductive Risk
Mucopolysaccharidosis Type IIIA	1 in 12,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 25,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 37,000	< 1 in 1,000,000
Muscle-eye-brain Disease	< 1 in 12,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 6,700	< 1 in 1,000,000
Niemann-Pick Disease Type C	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-associated	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Northern Epilepsy	< 1 in 50,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	1 in 140,000
PCCA-related Propionic Acidemia	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 5,300	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
PEX1-related Zellweger Syndrome Spectrum	1 in 11,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 5,000	1 in 990,000
PKHD1-related Autosomal Recessive Polycystic Kidney Disease	1 in 6,100	< 1 in 1,000,000
Polyglandular Autoimmune Syndrome Type 1	1 in 14,000	< 1 in 1,000,000
Pompe Disease	1 in 6,300	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Primary Carnitine Deficiency	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 35,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	< 1 in 1,000,000
Pycnodysostosis	< 1 in 50,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Salla Disease	< 1 in 30,000	< 1 in 1,000,000
Sandhoff Disease	1 in 32,000	< 1 in 1,000,000
Segawa Syndrome	< 1 in 50,000	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	1 in 16,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	1 in 9,100	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 4,900	1 in 970,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
Steroid-resistant Nephrotic Syndrome	1 in 40,000	< 1 in 1,000,000
Sulfate Transporter-related Osteochondrodysplasia	1 in 11,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 17,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 35,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 2,200	< 1 in 1,000,000
Usher Syndrome Type 3	< 1 in 50,000	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	1 in 8,800	< 1 in 1,000,000
Wilson Disease	1 in 8,600	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Congenital Adrenal Hypoplasia	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 50,000

Disease	DONOR 12280 Residual Risk	Reproductive Risk
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