Counsyl

RESULTS RECIPIENT
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**Report Date:** 10/31/2018

MALE
**DONOR 12369**
**DOB:** [redacted]
**Ethnicity:** Southeast Asian
**Sample Type:** EDTA Blood
**Date of Collection:** 10/23/2018
**Date Received:** 10/24/2018
**Date Tested:** 10/29/2018
**Barcode:** 11004212502551
**Accession ID:** CSLX6N3QXYLLJFH
**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 12369 | Partner |
|---|---|---|
| Panel Information | Foresight Carrier Screen Universal Panel **(175 conditions tested)** | N/A |
| All conditions tested<br>A complete list of all conditions tested can be found on page 4. | ⊖ **NEGATIVE**<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 12369 [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 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**.

**LABORATORY DIRECTOR**

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Report content approved by Saurav Guha, PhD, FACMG on Oct 31, 2018

# Conditions Tested

**11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia** - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** Southeast Asian 94%.

**21-hydroxylase-deficient Congenital Adrenal Hyperplasia** - **Gene:** CYP21A2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Southeast Asian 88%.

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

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

**Adenosine Deaminase Deficiency** - **Gene:** ADA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000022:1-12. **Detection Rate:** Southeast Asian >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:** Southeast Asian 90%.

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

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

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

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

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

**Argininemia** - **Gene:** ARG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001244438:1-8. **Detection Rate:** Southeast Asian 97%.

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

**ARSACS** - **Gene:** SACS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014363:2-10. **Detection Rate:** Southeast Asian 99%.

**Aspartylglycosaminuria** - **Gene:** AGA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000027:1-9. **Detection Rate:** Southeast Asian >99%.

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

**Ataxia-telangiectasia** - **Gene:** ATM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000051:2-63. **Detection Rate:** Southeast Asian >99%.

**ATP7A-related Disorders** - **Gene:** ATP7A. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000052:2-23. **Detection Rate:** Southeast Asian 92%.

**Autosomal Recessive Osteopetrosis Type 1** - **Gene:** TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006019:2-20. **Detection Rate:** Southeast Asian >99%.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

**Congenital Disorder of Glycosylation Type Ib** - **Gene:** MPI. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002435:1-8. **Detection Rate:** Southeast Asian >99%.

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

**Congenital Finnish Nephrosis** - **Gene:** NPHS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004646:1-29. **Detection Rate:** Southeast Asian >99%.

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

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

**D-bifunctional Protein Deficiency** - **Gene:** HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000414:1-24. **Detection Rate:** Southeast Asian 98%.
**Delta-sarcoglycanopathy** - **Gene:** SGCD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000337:2-9. **Detection Rate:** Southeast Asian 99%.
**Dysferlinopathy** - **Gene:** DYSF. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001130987:1-56. **Detection Rate:** Southeast Asian 98%.
**Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)** - **Gene:** DMD. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_004006:1-79. **Detection Rate:** Southeast Asian >99%.
**ERCC6-related Disorders** - **Gene:** ERCC6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000124:2-21. **Detection Rate:** Southeast Asian 99%.
**ERCC8-related Disorders** - **Gene:** ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000082:1-12. **Detection Rate:** Southeast Asian 95%.
**EVC-related Ellis-van Creveld Syndrome** - **Gene:** EVC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153717:1-21. **Detection Rate:** Southeast Asian 96%.
**EVC2-related Ellis-van Creveld Syndrome** - **Gene:** EVC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_147127:1-22. **Detection Rate:** Southeast Asian >99%.
**Fabry Disease** - **Gene:** GLA. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000169:1-7. **Detection Rate:** Southeast Asian 98%.
**Familial Dysautonomia** - **Gene:** IKBKAP. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003640:2-37. **Detection Rate:** Southeast Asian >99%.
**Familial Mediterranean Fever** - **Gene:** MEFV. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000243:1-10. **Detection Rate:** Southeast Asian >99%.
**Fanconi Anemia Complementation Group A** - **Gene:** FANCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000135:1-43. **Detection Rate:** Southeast Asian 92%.
**Fanconi Anemia Type C** - **Gene:** FANCC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000136:2-15. **Detection Rate:** Southeast Asian >99%.
**FKRP-related Disorders** - **Gene:** FKRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_024301:4. **Detection Rate:** Southeast Asian >99%.
**FKTN-related Disorders** - **Gene:** FKTN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001079802:3-11. **Detection Rate:** Southeast Asian >99%.
**Galactokinase Deficiency** - **Gene:** GALK1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000154:1-8. **Detection Rate:** Southeast Asian >99%.
**Galactosemia** - **Gene:** GALT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000155:1-11. **Detection Rate:** Southeast Asian >99%.
**Gamma-sarcoglycanopathy** - **Gene:** SGCG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000231:2-8. **Detection Rate:** Southeast Asian 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:** Southeast Asian 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:** Southeast Asian >99%.
**GLB1-related Disorders** - **Gene:** GLB1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000404:1-16. **Detection Rate:** Southeast Asian >99%.
**GLDC-related Glycine Encephalopathy** - **Gene:** GLDC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000170:1-25. **Detection Rate:** Southeast Asian 94%.
**Glutaric Acidemia Type 1** - **Gene:** GCDH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000159:2-12. **Detection Rate:** Southeast Asian >99%.
**Glycogen Storage Disease Type Ia** - **Gene:** G6PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000151:1-5. **Detection Rate:** Southeast Asian >99%.
**Glycogen Storage Disease Type Ib** - **Gene:** SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001164277:3-11. **Detection Rate:** Southeast Asian >99%.
**Glycogen Storage Disease Type III** - **Gene:** AGL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000642:2-34. **Detection Rate:** Southeast Asian >99%.
**GNPTAB-related Disorders** - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024312:1-21. **Detection Rate:** Southeast Asian >99%.
**GRACILE Syndrome** - **Gene:** BCS1L. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004328:3-9. **Detection Rate:** Southeast Asian >99%.
**HADHA-related Disorders** - **Gene:** HADHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000182:1-20. **Detection Rate:** Southeast Asian >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:** Southeast Asian >99%.
**Hereditary Fructose Intolerance** - **Gene:** ALDOB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000035:2-9. **Detection Rate:** Southeast Asian >99%.
**Herlitz Junctional Epidermolysis Bullosa, LAMA3-related** - **Gene:** LAMA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000227:1-38. **Detection Rate:** Southeast Asian >99%.
**Herlitz Junctional Epidermolysis Bullosa, LAMB3-related** - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228:2-23. **Detection Rate:** Southeast Asian >99%.
**Herlitz Junctional Epidermolysis Bullosa, LAMC2-related** - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** Southeast Asian >99%.
**Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** - **Gene:** HEXA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000520:1-14. **Detection Rate:** Southeast Asian >99%.
**HMG-CoA Lyase Deficiency** - **Gene:** HMGCL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000191:1-9. **Detection Rate:** Southeast Asian 98%.
**Holocarboxylase Synthetase Deficiency** - **Gene:** HLCS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000411:4-12. **Detection Rate:** Southeast Asian >99%.
**Homocystinuria Caused by Cystathionine Beta-synthase Deficiency** - **Gene:** CBS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000071:3-17. **Detection Rate:** Southeast Asian >99%.
**Hydrolethalus Syndrome** - **Gene:** HYLS1. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_001134793:3. **Detection Rate:** Southeast Asian >99%.
**Hypophosphatasia, Autosomal Recessive** - **Gene:** ALPL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000478:2-12. **Detection Rate:** Southeast Asian >99%.
**Inclusion Body Myopathy 2** - **Gene:** GNE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001128227:1-12. **Detection Rate:** Southeast Asian >99%.
**Isovaleric Acidemia** - **Gene:** IVD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002225:1-12. **Detection Rate:** Southeast Asian >99%.
**Joubert Syndrome 2** - **Gene:** TMEM216. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001173990:1-5. **Detection Rate:** Southeast Asian >99%.
**KCNJ11-related Familial Hyperinsulinism** - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000525:1. **Detection Rate:** Southeast Asian >99%.
**Krabbe Disease** - **Gene:** GALC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000153:1-17. **Detection Rate:** Southeast Asian >99%.
**LAMA2-related Muscular Dystrophy** - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-65. **Detection Rate:** Southeast Asian >99%.
**Leigh Syndrome, French-Canadian Type** - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133259:1-38. **Detection Rate:** Southeast Asian >99%.
**Lipoamide Dehydrogenase Deficiency** - **Gene:** DLD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000108:1-14. **Detection Rate:** Southeast Asian >99%.
**Lipoid Congenital Adrenal Hyperplasia** - **Gene:** STAR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000349:1-7. **Detection Rate:** Southeast Asian >99%.

**Lysosomal Acid Lipase Deficiency** - **Gene:** LIPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000235:2-10. **Detection Rate:** Southeast Asian >99%.
**Maple Syrup Urine Disease Type 1B** - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_183050:1-10. **Detection Rate:** Southeast Asian >99%.
**Maple Syrup Urine Disease Type Ia** - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000709:1-9. **Detection Rate:** Southeast Asian >99%.
**Maple Syrup Urine Disease Type II** - **Gene:** DBT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001918:1-11. **Detection Rate:** Southeast Asian 96%.
**Medium Chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000016:1-12. **Detection Rate:** Southeast Asian >99%.
**Megalencephalic Leukoencephalopathy with Subcortical Cysts** - **Gene:** MLC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015166:2-12. **Detection Rate:** Southeast Asian >99%.
**Metachromatic Leukodystrophy** - **Gene:** ARSA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000487:1-8. **Detection Rate:** Southeast Asian >99%.
**Methylmalonic Acidemia, cblA Type** - **Gene:** MMAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_172250:2-7. **Detection Rate:** Southeast Asian >99%.
**Methylmalonic Acidemia, cblB Type** - **Gene:** MMAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_052845:1-9. **Detection Rate:** Southeast Asian >99%.
**Methylmalonic Aciduria and Homocystinuria, cblC Type** - **Gene:** MMACHC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015506:1-4. **Detection Rate:** Southeast Asian >99%.
**MKS1-related Disorders** - **Gene:** MKS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017777:1-18. **Detection Rate:** Southeast Asian >99%.
**Mucolipidosis III Gamma** - **Gene:** GNPTG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032520:1-11. **Detection Rate:** Southeast Asian >99%.
**Mucolipidosis IV** - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_020533:1-14. **Detection Rate:** Southeast Asian >99%.
**Mucopolysaccharidosis Type I** - **Gene:** IDUA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000203:1-14. **Detection Rate:** Southeast Asian >99%.
**Mucopolysaccharidosis Type II** - **Gene:** IDS. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000202:1-9. **Detection Rate:** Southeast Asian 88%.
**Mucopolysaccharidosis Type IIIA** - **Gene:** SGSH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000199:1-8. **Detection Rate:** Southeast Asian >99%.
**Mucopolysaccharidosis Type IIIB** - **Gene:** NAGLU. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000263:1-6. **Detection Rate:** Southeast Asian >99%.
**Mucopolysaccharidosis Type IIIC** - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_152419:1-18. **Detection Rate:** Southeast Asian >99%.
**Muscle-eye-brain Disease** - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017739:2-22. **Detection Rate:** Southeast Asian 96%.
**MUT-related Methylmalonic Acidemia** - **Gene:** MUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** Southeast Asian >99%.
**MYO7A-related Disorders** - **Gene:** MYO7A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000260:2-49. **Detection Rate:** Southeast Asian >99%.
**NEB-related Nemaline Myopathy** - **Gene:** NEB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** Southeast Asian 92%.
**Nephrotic Syndrome, NPHS2-related** - **Gene:** NPHS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014625:1-8. **Detection Rate:** Southeast Asian >99%.
**Niemann-Pick Disease Type C** - **Gene:** NPC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000271:1-25. **Detection Rate:** Southeast Asian >99%.
**Niemann-Pick Disease Type C2** - **Gene:** NPC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006432:1-5. **Detection Rate:** Southeast Asian >99%.
**Niemann-Pick Disease, SMPD1-associated** - **Gene:** SMPD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000543:1-6. **Detection Rate:** Southeast Asian >99%.
**Nijmegen Breakage Syndrome** - **Gene:** NBN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002485:1-16. **Detection Rate:** Southeast Asian >99%.
**Northern Epilepsy** - **Gene:** CLN8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_018941:2-3. **Detection Rate:** Southeast Asian >99%.
**Ornithine Transcarbamylase Deficiency** - **Gene:** OTC. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000531:1-10. **Detection Rate:** Southeast Asian 97%.
**PCCA-related Propionic Acidemia** - **Gene:** PCCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000282:1-24. **Detection Rate:** Southeast Asian 95%.
**PCCB-related Propionic Acidemia** - **Gene:** PCCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001178014:1-16. **Detection Rate:** Southeast Asian >99%.
**PCDH15-related Disorders** - **Gene:** PCDH15. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_033056:2-33. **Detection Rate:** Southeast Asian 93%.
**Pendred Syndrome** - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000441:2-21. **Detection Rate:** Southeast Asian >99%.
**Peroxisome Biogenesis Disorder Type 3** - **Gene:** PEX12. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000286:1-3. **Detection Rate:** Southeast Asian >99%.
**Peroxisome Biogenesis Disorder Type 4** - **Gene:** PEX6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000287:1-17. **Detection Rate:** Southeast Asian 97%.
**Peroxisome Biogenesis Disorder Type 5** - **Gene:** PEX2. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000318:4. **Detection Rate:** Southeast Asian >99%.
**Peroxisome Biogenesis Disorder Type 6** - **Gene:** PEX10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153818:1-6. **Detection Rate:** Southeast Asian >99%.
**PEX1-related Zellweger Syndrome Spectrum** - **Gene:** PEX1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000466:1-24. **Detection Rate:** Southeast Asian >99%.
**Phenylalanine Hydroxylase Deficiency** - **Gene:** PAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000277:1-13. **Detection Rate:** Southeast Asian >99%.
**PKHD1-related Autosomal Recessive Polycystic Kidney Disease** - **Gene:** PKHD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138694:2-67. **Detection Rate:** Southeast Asian >99%.
**Polyglandular Autoimmune Syndrome Type 1** - **Gene:** AIRE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000383:1-14. **Detection Rate:** Southeast Asian >99%.
**Pompe Disease** - **Gene:** GAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000152:2-20. **Detection Rate:** Southeast Asian >99%.
**PPT1-related Neuronal Ceroid Lipofuscinosis** - **Gene:** PPT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000310:1-9. **Detection Rate:** Southeast Asian >99%.
**Primary Carnitine Deficiency** - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003060:1-10. **Detection Rate:** Southeast Asian >99%.
**Primary Hyperoxaluria Type 1** - **Gene:** AGXT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000030:1-11. **Detection Rate:** Southeast Asian >99%.
**Primary Hyperoxaluria Type 2** - **Gene:** GRHPR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012203:1-9. **Detection Rate:** Southeast Asian >99%.
**Primary Hyperoxaluria Type 3** - **Gene:** HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** Southeast Asian >99%.
**PROP1-related Combined Pituitary Hormone Deficiency** - **Gene:** PROP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006261:1-3. **Detection Rate:** Southeast Asian >99%.

**Pycnodysostosis** - **Gene:** CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000396:2-8. **Detection Rate:** Southeast Asian >99%.
**Pyruvate Carboxylase Deficiency** - **Gene:** PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_022172:2-21. **Detection Rate:** Southeast Asian >99%.
**Rhizomelic Chondrodysplasia Punctata Type 1** - **Gene:** PEX7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000288:1-10. **Detection Rate:** Southeast Asian >99%.
**RTEL1-related Disorders** - **Gene:** RTEL1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032957:2-35. **Detection Rate:** Southeast Asian >99%.
**Salla Disease** - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012434:1-11. **Detection Rate:** Southeast Asian 98%.
**Sandhoff Disease** - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** Southeast Asian 99%.
**Segawa Syndrome** - **Gene:** TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000360:1-13. **Detection Rate:** Southeast Asian >99%.
**Short Chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000017:1-10. **Detection Rate:** Southeast Asian >99%.
**Sjogren-Larsson Syndrome** - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** Southeast Asian 97%.
**Smith-Lemli-Opitz Syndrome** - **Gene:** DHCR7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001360:3-9. **Detection Rate:** Southeast Asian >99%.
**Spastic Paraplegia Type 15** - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015346:2-42. **Detection Rate:** Southeast Asian >99%.
**Spinal Muscular Atrophy** - **Gene:** SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** Southeast Asian 93%.
**Spondylothoracic Dysostosis** - **Gene:** MESP2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** Southeast Asian >99%.
**Sulfate Transporter-related Osteochondrodysplasia** - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000112:2-3. **Detection Rate:** Southeast Asian >99%.
**TGM1-related Autosomal Recessive Congenital Ichthyosis** - **Gene:** TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000359:2-15. **Detection Rate:** Southeast Asian >99%.
**TPP1-related Neuronal Ceroid Lipofuscinosis** - **Gene:** TPP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000391:1-13. **Detection Rate:** Southeast Asian >99%.
**Tyrosinemia Type I** - **Gene:** FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** Southeast Asian >99%.
**Tyrosinemia Type II** - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** Southeast Asian >99%.
**USH1C-related Disorders** - **Gene:** USH1C. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153676:1-27. **Detection Rate:** Southeast Asian >99%.
**USH2A-related Disorders** - **Gene:** USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_206933:2-72. **Detection Rate:** Southeast Asian 94%.
**Usher Syndrome Type 3** - **Gene:** CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** Southeast Asian >99%.
**Very Long Chain Acyl-CoA Dehydrogenase Deficiency** - **Gene:** ACADVL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000018:1-20. **Detection Rate:** Southeast Asian >99%.
**Wilson Disease** - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** Southeast Asian >99%.
**X-linked Adrenoleukodystrophy** - **Gene:** ABCD1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000033:1-6. **Detection Rate:** Southeast Asian 77%.
**X-linked Alport Syndrome** - **Gene:** COL4A5. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000495:1-51. **Detection Rate:** Southeast Asian 95%.
**X-linked Congenital Adrenal Hypoplasia** - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** Southeast Asian 99%.
**X-linked Juvenile Retinoschisis** - **Gene:** RS1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000330:1-6. **Detection Rate:** Southeast Asian 98%.
**X-linked Myotubular Myopathy** - **Gene:** MTM1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000252:2-15. **Detection Rate:** Southeast Asian 98%.
**X-linked Severe Combined Immunodeficiency** - **Gene:** IL2RG. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000206:1-8. **Detection Rate:** Southeast Asian >99%.
**Xeroderma Pigmentosum Group A** - **Gene:** XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** Southeast Asian >99%.
**Xeroderma Pigmentosum Group C** - **Gene:** XPC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004628:1-16. **Detection Rate:** Southeast Asian 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 12369 Residual Risk | Reproductive Risk |
|---|---|---|
| **11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia** | 1 in 3,300 | < 1 in 1,000,000 |
| **21-hydroxylase-deficient Congenital Adrenal Hyperplasia** | 1 in 480 | 1 in 110,000 |
| **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 39,000 | < 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 16,000 | < 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 18,000 | < 1 in 1,000,000 |
| **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 50,000 | < 1 in 1,000,000 |
| **Cohen Syndrome** | < 1 in 15,000 | < 1 in 1,000,000 |
| **COL4A3-related Alport Syndrome** | 1 in 11,000 | < 1 in 1,000,000 |
| **COL4A4-related Alport Syndrome** | 1 in 21,000 | < 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 8,600 | < 1 in 1,000,000 |
| **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 12369 Residual Risk | Reproductive Risk |
|---|---|---|
| **Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)** | Not calculated | Not calculated |
| **ERCC6-related Disorders** | 1 in 19,000 | < 1 in 1,000,000 |
| **ERCC8-related Disorders** | 1 in 7,300 | < 1 in 1,000,000 |
| **EVC-related Ellis-van Creveld Syndrome** | 1 in 7,500 | < 1 in 1,000,000 |
| **EVC2-related Ellis-van Creveld Syndrome** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Fabry Disease** | < 1 in 1,000,000 | 1 in 80,000 |
| **Familial Dysautonomia** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Familial Mediterranean Fever** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Fanconi Anemia Complementation Group A** | 1 in 3,100 | < 1 in 1,000,000 |
| **Fanconi Anemia Type C** | 1 in 16,000 | < 1 in 1,000,000 |
| **FKRP-related Disorders** | < 1 in 50,000 | < 1 in 1,000,000 |
| **FKTN-related Disorders** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Galactokinase Deficiency** | 1 in 35,000 | < 1 in 1,000,000 |
| **Galactosemia** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Gamma-sarcoglycanopathy** | 1 in 3,000 | < 1 in 1,000,000 |
| **Gaucher Disease** | 1 in 310 | 1 in 150,000 |
| **GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness** | 1 in 10,000 | < 1 in 1,000,000 |
| **GLB1-related Disorders** | 1 in 19,000 | < 1 in 1,000,000 |
| **GLDC-related Glycine Encephalopathy** | 1 in 2,800 | < 1 in 1,000,000 |
| **Glutaric Acidemia Type 1** | 1 in 10,000 | < 1 in 1,000,000 |
| **Glycogen Storage Disease Type Ia** | 1 in 18,000 | < 1 in 1,000,000 |
| **Glycogen Storage Disease Type Ib** | 1 in 35,000 | < 1 in 1,000,000 |
| **Glycogen Storage Disease Type III** | 1 in 16,000 | < 1 in 1,000,000 |
| **GNPTAB-related Disorders** | 1 in 32,000 | < 1 in 1,000,000 |
| **GRACILE Syndrome** | < 1 in 50,000 | < 1 in 1,000,000 |
| **HADHA-related Disorders** | 1 in 15,000 | < 1 in 1,000,000 |
| **Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)** | 1 in 2,400 | 1 in 240,000 |
| **Hereditary Fructose Intolerance** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Herlitz Junctional Epidermolysis Bullosa, LAMA3-related** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Herlitz Junctional Epidermolysis Bullosa, LAMB3-related** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Herlitz Junctional Epidermolysis Bullosa, LAMC2-related** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** | 1 in 30,000 | < 1 in 1,000,000 |
| **HMG-CoA Lyase Deficiency** | < 1 in 33,000 | < 1 in 1,000,000 |
| **Holocarboxylase Synthetase Deficiency** | 1 in 15,000 | < 1 in 1,000,000 |
| **Homocystinuria Caused by Cystathionine Beta-synthase Deficiency** | 1 in 25,000 | < 1 in 1,000,000 |
| **Hydrolethalus Syndrome** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Hypophosphatasia, Autosomal Recessive** | 1 in 16,000 | < 1 in 1,000,000 |
| **Inclusion Body Myopathy 2** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Isovaleric Acidemia** | 1 in 25,000 | < 1 in 1,000,000 |
| **Joubert Syndrome 2** | < 1 in 50,000 | < 1 in 1,000,000 |
| **KCNJ11-related Familial Hyperinsulinism** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Krabbe Disease** | 1 in 15,000 | < 1 in 1,000,000 |
| **LAMA2-related Muscular Dystrophy** | 1 in 17,000 | < 1 in 1,000,000 |
| **Leigh Syndrome, French-Canadian Type** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Lipoamide Dehydrogenase Deficiency** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Lipoid Congenital Adrenal Hyperplasia** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Lysosomal Acid Lipase Deficiency** | 1 in 30,000 | < 1 in 1,000,000 |
| **Maple Syrup Urine Disease Type 1B** | 1 in 25,000 | < 1 in 1,000,000 |
| **Maple Syrup Urine Disease Type Ia** | 1 in 19,000 | < 1 in 1,000,000 |
| **Maple Syrup Urine Disease Type II** | 1 in 7,600 | < 1 in 1,000,000 |
| **Medium Chain Acyl-CoA Dehydrogenase Deficiency** | 1 in 11,000 | < 1 in 1,000,000 |
| **Megalencephalic Leukoencephalopathy with Subcortical Cysts** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Metachromatic Leukodystrophy** | 1 in 20,000 | < 1 in 1,000,000 |
| **Methylmalonic Acidemia, cblA Type** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Methylmalonic Acidemia, cblB Type** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Methylmalonic Aciduria and Homocystinuria, cblC Type** | 1 in 16,000 | < 1 in 1,000,000 |
| **MKS1-related Disorders** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Mucolipidosis III Gamma** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Mucolipidosis 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 |

| Disease | DONOR 12369 Residual Risk | Reproductive Risk |
|---|---|---|
| **Mucopolysaccharidosis Type IIIA** | 1 in 16,000 | < 1 in 1,000,000 |
| **Mucopolysaccharidosis Type IIIB** | 1 in 31,000 | < 1 in 1,000,000 |
| **Mucopolysaccharidosis Type IIIC** | 1 in 43,000 | < 1 in 1,000,000 |
| **Muscle-eye-brain Disease** | < 1 in 12,000 | < 1 in 1,000,000 |
| **MUT-related Methylmalonic Acidemia** | 1 in 5,300 | < 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 |
| **Nephrotic Syndrome, NPHS2-related** | 1 in 35,000 | < 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 35,000 | < 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 50,000 | < 1 in 1,000,000 |
| **Polyglandular Autoimmune Syndrome Type 1** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Pompe Disease** | 1 in 11,000 | < 1 in 1,000,000 |
| **PPT1-related Neuronal Ceroid Lipofuscinosis** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Primary Carnitine Deficiency** | 1 in 16,000 | < 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 20,000 | < 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 30,000 | < 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 50,000 | < 1 in 1,000,000 |
| **Spastic Paraplegia Type 15** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Spinal Muscular Atrophy** | Negative for g.27134T>G SNP<br>SMN1: 2 copies<br>1 in 700 | 1 in 150,000 |
| **Spondylothoracic Dysostosis** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Sulfate Transporter-related Osteochondrodysplasia** | 1 in 11,000 | < 1 in 1,000,000 |
| **TGM1-related Autosomal Recessive Congenital Ichthyosis** | 1 in 22,000 | < 1 in 1,000,000 |
| **TPP1-related Neuronal Ceroid Lipofuscinosis** | 1 in 30,000 | < 1 in 1,000,000 |
| **Tyrosinemia Type I** | 1 in 17,000 | < 1 in 1,000,000 |
| **Tyrosinemia Type II** | 1 in 25,000 | < 1 in 1,000,000 |
| **USH1C-related Disorders** | 1 in 35,000 | < 1 in 1,000,000 |
| **USH2A-related Disorders** | 1 in 2,200 | < 1 in 1,000,000 |
| **Usher Syndrome Type 3** | < 1 in 50,000 | < 1 in 1,000,000 |
| **Very Long Chain Acyl-CoA Dehydrogenase Deficiency** | 1 in 8,800 | < 1 in 1,000,000 |
| **Wilson Disease** | 1 in 5,000 | 1 in 990,000 |
| **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 |

Counsyl

RESULTS RECIPIENT
**SEATTLE SPERM BANK**
**Attn:** Dr. Jeffrey Olliffe
**NPI:** 1306838271
**Report Date:** 10/31/2018

MALE
**DONOR 12369**
**DOB:** [redacted]
**Ethnicity:** Southeast Asian
**Barcode:** 11004212502551

FEMALE
N/A

| Disease | DONOR 12369 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 |