



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
Attn: Dr. Jeffrey Olliffe  
4915 25th Ave NE, Suite 204W  
Seattle, WA 98105  
Phone: (206) 588-1484  
Fax: (206) 588-1484  
NPI: 1306838271  
Report Date: 07/20/2016

MALE  
**DONOR 10057**  
DOB: [REDACTED]  
Ethnicity: Northern European  
Sample Type: OG-510 Saliva  
Date of Collection: 05/26/2016  
Date Received: 06/02/2016  
Date Tested: 06/10/2016  
Barcode: 55101509201147  
Indication: Egg or sperm donor

FEMALE  
N/A

This is an **amended report**, from the 06/10/2016 original. Panel change requested.

# Family Prep Screen

**NEGATIVE**

## ABOUT THIS TEST

The Counsyl Family Prep Screen (version 2.0) 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 10057	Partner
Panel Information	Family Prep Screen 2.0 Universal Panel (104 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.



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## Methods and Limitations

**DONOR 10057** [Family Prep Screen 2.0]: sequencing, targeted genotyping, copy number analysis, and analysis of homologous regions.

### Sequencing

High-throughput sequencing is 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. These regions are sequenced to high coverage and the sequences are compared to standards and references of normal variation. Mutations may not be detected in areas of lower sequence coverage. On average, more than 99% of all bases in the exons listed for each gene are sequenced at the minimum read depth. Variants discovered in other exons of these genes will also be reported if they meet quality control criteria. Triplet repeats and large deletions and duplications may not be detected. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes are not well analyzed by this method.

High-throughput sequencing detects, on average, 94% of known clinically significant variants. Disease-specific detection rates and residual risks are reported as "greater than (>)" and "less than (<)" the values for targeted genotyping, respectively. More precise values are not currently available, but may become available in the future.

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 "predicted" or "likely" pathogenic are reported. Predicted/likely pathogenic variants are described elsewhere in the report as "predicted/likely to have a negative impact on gene function". In general, predicted pathogenic variants are those which are predicted to be pathogenic based on the nature of the sequence change, while likely pathogenic variants are evaluated by reviewing reports of allele frequencies in cases and controls, functional studies, variant annotation and effect prediction, and segregation studies. Benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Literature citations validating reported variants are available upon request.

### Targeted genotyping

Targeted DNA mutation analysis is used to determine the genotypes of the listed variants in the Conditions Tested section of the report. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

### Copy number analysis

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. In addition, a small percentage of spinal muscular atrophy (SMA) cases are caused by nondeletion mutations in the *SMN1* gene. Thus, a test result of two *SMN1* copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more *SMN1* gene copies. Some SMA cases arise as the result of *de novo* mutation events which will not be detected by carrier testing.

### 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 regions cannot be determined, but are estimated from copy number analysis. 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). In addition, some individuals with four alpha globin genes are carriers with three genes on one chromosome and a deletion on the other chromosome. This and similar carrier states, where complementary changes exist in both the gene and a pseudogene, may not be detected by the assay.



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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.

## 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. 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 Family Prep Screen 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*), and additional Tay-Sachs disease testing can be performed using a biochemical assay (*Gross et al. Genet. Med. 2008;10(1):54-56*).

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**.

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### LAB DIRECTORS

H. Peter Kang, MD, MS, FCAP

Rebecca Mar-Heyming, PhD, DABMG

# Conditions Tested

**21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia** - Gene: CYP21A2. Autosomal Recessive. Analysis of Homologous Regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111VfsX21, I173N, L308FfsX6, P31L, Q319\*, Q319\*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.

**ABCC8-related Hyperinsulinism** - Gene: ABCC8. Autosomal Recessive. Sequencing. Exons: NM\_000352:1-39. **Detection Rate:** Unknown due to rarity of disease.

**Achromatopsia** - Gene: CNGB3. Autosomal Recessive. Sequencing. Exons: NM\_019098:1-18. **Detection Rate:** Northern European > 62%.

**Alkaptonuria** - Gene: HGD. Autosomal Recessive. Sequencing. Exons: NM\_000187:1-14. **Detection Rate:** Northern European > 80%.

**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-1 Antitrypsin Deficiency** - Gene: SERPINA1. Autosomal Recessive. Sequencing. Exons: NM\_000295:2-5. **Detection Rate:** Northern European > 95%.

**Alpha-Mannosidosis** - Gene: MAN2B1. Autosomal Recessive. Sequencing. Exons: NM\_000528:1-15,17-24. **Detection Rate:** Northern European > 32%.

**Andermann Syndrome** - Gene: SLC12A6. Autosomal Recessive. Sequencing. Exons: NM\_133647:1-25. **Detection Rate:** Unknown due to rarity of disease.

**ARSACS** - Gene: SACS. Autosomal Recessive. Sequencing. Exons: NM\_014363:2-10. **Detection Rate:** Unknown due to rarity of disease.

**Aspartylglycosaminuria** - Gene: AGA. Autosomal Recessive. Sequencing. Exons: NM\_000027:1-9. **Detection Rate:** Unknown due to rarity of disease.

**Ataxia With Vitamin E Deficiency** - Gene: TTPA. Autosomal Recessive. Sequencing. Exons: NM\_000370:1-5. **Detection Rate:** Northern European > 10%.

**Ataxia-Telangiectasia** - Gene: ATM. Autosomal Recessive. Sequencing. Exons: NM\_000051:2-63. **Detection Rate:** Northern European > 65%.

**Autosomal Recessive Polycystic Kidney Disease** - Gene: PKHD1. Autosomal Recessive. Sequencing. Exons: NM\_138694:2-67. **Detection Rate:** Northern European > 18%.

**Bardet-Biedl Syndrome, BBS1-related** - Gene: BBS1. Autosomal Recessive. Sequencing. Exons: NM\_024649:1-17. **Detection Rate:** Northern European > 79%.

**Bardet-Biedl Syndrome, BBS10-related** - Gene: BBS10. Autosomal Recessive. Sequencing. Exons: NM\_024685:1-2. **Detection Rate:** Northern European > 46%.

**Biotinidase Deficiency** - Gene: BTM. Autosomal Recessive. Sequencing. Exons: NM\_000060:1-4. **Detection Rate:** Northern European > 45%.

**Bloom Syndrome** - Gene: BLM. Autosomal Recessive. Sequencing. Exons: NM\_000057:2-22. **Detection Rate:** Northern European > 10%.

**Canavan Disease** - Gene: ASPA. Autosomal Recessive. Sequencing. Exons: NM\_000049:1-6. **Detection Rate:** Northern European > 53%.

**Carnitine Palmitoyltransferase IA Deficiency** - Gene: CPT1A. Autosomal Recessive. Sequencing. Exons: NM\_001876:2-19. **Detection Rate:** Northern European > 10%.

**Carnitine Palmitoyltransferase II Deficiency** - Gene: CPT2. Autosomal Recessive. Sequencing. Exons: NM\_000098:1-5. **Detection Rate:** Northern European > 80%.

**Cartilage-Hair Hypoplasia** - Gene: RMRP. Autosomal Recessive. Sequencing. Exon: NR\_003051:1. **Detection Rate:** Northern European > 48%.

**Choroideremia** - Gene: CHM. X-linked Recessive. Sequencing. Exons: NM\_000390:1-15. **Detection Rate:** Unknown due to rarity of disease.

**Citrullinemia Type 1** - Gene: ASS1. Autosomal Recessive. Sequencing. Exons: NM\_000050:3-16. **Detection Rate:** Northern European > 20%.

**CLN3-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN3. Autosomal Recessive. Sequencing. Exons: NM\_001042432:2-16. **Detection Rate:** Northern European > 96%.

**CLN5-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN5. Autosomal Recessive. Sequencing. Exons: NM\_006493:1-4. **Detection Rate:** Unknown due to rarity of disease.

**Cohen Syndrome** - Gene: VPS13B. Autosomal Recessive. Sequencing. Exons: NM\_017890:2-62. **Detection Rate:** Unknown due to rarity of disease.

**Congenital Disorder of Glycosylation Type Ia** - Gene: PMM2. Autosomal Recessive. Sequencing. Exons: NM\_000303:1-8. **Detection Rate:** Northern European > 72%.

**Congenital Disorder of Glycosylation Type Ib** - Gene: MPI. Autosomal Recessive. Sequencing. Exons: NM\_002435:1-8. **Detection Rate:** Unknown due to rarity of disease.

**Congenital Finnish Nephrosis** - Gene: NPHS1. Autosomal Recessive. Sequencing. Exons: NM\_004646:2-23,26-27,29. **Detection Rate:** Unknown due to rarity of disease.

**Costeff Optic Atrophy Syndrome** - Gene: OPA3. Autosomal Recessive. Sequencing. Exons: NM\_025136:1-2. **Detection Rate:** Unknown due to rarity of disease.

**Cystic Fibrosis** - Gene: CFTR. Autosomal Recessive. Sequencing. Exons: NM\_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Northern European > 91%.

**Cystinosis** - Gene: CTNS. Autosomal Recessive. Sequencing. Exons: NM\_004937:3-12. **Detection Rate:** Northern European > 67%.

**D-Bifunctional Protein Deficiency** - Gene: HSD17B4. Autosomal Recessive. Sequencing. Exons: NM\_000414:1-24. **Detection Rate:** Northern European > 35%.

**Factor XI Deficiency** - Gene: F11. Autosomal Recessive. Sequencing. Exons: NM\_000128:2-15. **Detection Rate:** Northern European > 10%.

**Familial Dysautonomia** - Gene: IKBKAP. Autosomal Recessive. Sequencing. Exons: NM\_003640:19-20,26. **Detection Rate:** Unknown due to rarity of disease.

**Familial Mediterranean Fever** - Gene: MEFV. Autosomal Recessive. Sequencing. Exons: NM\_000243:1-10. **Detection Rate:** Unknown due to rarity of disease.

**Fanconi Anemia Type C** - Gene: FANCC. Autosomal Recessive. Sequencing. Exons: NM\_000136:2-15. **Detection Rate:** Northern European > 54%.

**Galactosemia** - Gene: GALT. Autosomal Recessive. Sequencing. Exons: NM\_000155:1-11. **Detection Rate:** Northern European > 80%.

**Gaucher Disease** - Gene: GBA. Autosomal Recessive. Targeted Genotyping. **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. Exons: NM\_004004:1-2. **Detection Rate:** Northern European > 79%.

**Glutaric Acidemia Type 1** - Gene: GCDH. Autosomal Recessive. Sequencing. Exons: NM\_000159:2-12. **Detection Rate:** Northern European > 40%.

**Glycogen Storage Disease Type Ia** - Gene: G6PC. Autosomal Recessive. Sequencing. Exons: NM\_000151:1-5. **Detection Rate:** Northern European > 61%.

**Glycogen Storage Disease Type Ib** - Gene: SLC37A4. Autosomal Recessive. Sequencing. Exons: NM\_001164277:3-11. **Detection Rate:** Northern European > 46%.

**Glycogen Storage Disease Type III** - Gene: AGL. Autosomal Recessive. Sequencing. Exons: NM\_000642:2-34. **Detection Rate:** Northern European > 45%.

**Glycogen Storage Disease Type V** - Gene: PYGM. Autosomal Recessive. Sequencing. Exons: NM\_005609:1-20. **Detection Rate:** Northern European > 80%.

**GRACILE Syndrome** - Gene: BCS1L. Autosomal Recessive. Sequencing. Exons: NM\_004328:3-9. **Detection Rate:** Unknown due to rarity of disease.

**Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sick Cell Disease)** - Gene: HBB. Autosomal Recessive. Sequencing. Exons: NM\_000518:1-3. **Detection Rate:** Northern European > 83%.

**Hereditary Fructose Intolerance** - Gene: ALDOB. Autosomal Recessive. Sequencing. Exons: NM\_000035:2-9. **Detection Rate:** Northern European > 75%.

**Hereditary Thymine-Uraciluria** - Gene: DPYD. Autosomal Recessive. Sequencing. Exons: NM\_000110:1-23. **Detection Rate:** Northern European > 52%.

**Herlitz Junctional Epidermolysis Bullosa, LAMA3-related** - Gene: LAMA3. Autosomal Recessive. Sequencing. Exons: NM\_000227:1-16,18-38. **Detection Rate:** Northern European > 10%.

**Herlitz Junctional Epidermolysis Bullosa, LAMB3-related** - Gene: LAMB3. Autosomal Recessive. Sequencing. Exons: NM\_000228:2-23. **Detection Rate:** Northern European > 48%.

**Herlitz Junctional Epidermolysis Bullosa, LAMC2-related** - Gene: LAMC2. Autosomal Recessive. Sequencing. Exons: NM\_005562:1-23. **Detection Rate:** Unknown due to rarity of disease.

**Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** - Gene: HEXA. Autosomal Recessive. Sequencing. Exons: NM\_000520:1-14. **Detection Rate:** Northern European > 23%.



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**Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency** - Gene: CBS. Autosomal Recessive. Sequencing. Exons: NM\_000071:3-17. **Detection Rate:** Northern European > 14%.

**Hurler Syndrome** - Gene: IDUA. Autosomal Recessive. Targeted Genotyping. Variants (2): Q70\*, W402\*. **Detection Rate:** Northern European 67%.

**Hypophosphatasia, Autosomal Recessive** - Gene: ALPL. Autosomal Recessive. Sequencing. Exons: NM\_000478:2-12. **Detection Rate:** Northern European > 30%.

**Inclusion Body Myopathy 2** - Gene: GNE. Autosomal Recessive. Sequencing. Exons: NM\_001128227:3-12. **Detection Rate:** Unknown due to rarity of disease.

**Isovaleric Acidemia** - Gene: IVD. Autosomal Recessive. Sequencing. Exons: NM\_002225:1-12. **Detection Rate:** Northern European > 47%.

**Joubert Syndrome 2** - Gene: TMEM216. Autosomal Recessive. Sequencing. Exons: NM\_001173990:1-5. **Detection Rate:** Unknown due to rarity of disease.

**Krabbe Disease** - Gene: GALC. Autosomal Recessive. Sequencing. Exons: NM\_000153:1-17. **Detection Rate:** Northern European > 58%.

**Limb-Girdle Muscular Dystrophy Type 2D** - Gene: SGCA. Autosomal Recessive. Sequencing. Exons: NM\_000023:1-9. **Detection Rate:** Northern European > 32%.

**Limb-Girdle Muscular Dystrophy Type 2E** - Gene: SGCB. Autosomal Recessive. Sequencing. Exons: NM\_000232:1-6. **Detection Rate:** Northern European > 12%.

**Lipoamide Dehydrogenase Deficiency** - Gene: DLD. Autosomal Recessive. Sequencing. Exons: NM\_000108:1-14. **Detection Rate:** Unknown due to rarity of disease.

**Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency** - Gene: HADHA. Autosomal Recessive. Sequencing. Exons: NM\_000182:1-20. **Detection Rate:** Northern European > 87%.

**Maple Syrup Urine Disease Type 1B** - Gene: BCKDHB. Autosomal Recessive. Sequencing. Exons: NM\_183050:1-10. **Detection Rate:** Unknown due to rarity of disease.

**Medium Chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADM. Autosomal Recessive. Sequencing. Exons: NM\_000016:1-12. **Detection Rate:** Northern European > 78%.

**Megalencephalic Leukoencephalopathy With Subcortical Cysts** - Gene: MLC1. Autosomal Recessive. Sequencing. Exons: NM\_015166:2-12. **Detection Rate:** Northern European > 13%.

**Metachromatic Leukodystrophy** - Gene: ARSA. Autosomal Recessive. Sequencing. Exons: NM\_000487:1-8. **Detection Rate:** Northern European > 53%.

**Mucopolysaccharidosis IV** - Gene: MCOLN1. Autosomal Recessive. Sequencing. Exons: NM\_020533:1-14. **Detection Rate:** Northern European > 10%.

**Muscle-Eye-Brain Disease** - Gene: POMGNT1. Autosomal Recessive. Sequencing. Exons: NM\_017739:2-22. **Detection Rate:** Northern European > 75%.

**NEB-related Nemaline Myopathy** - Gene: NEB. Autosomal Recessive. Sequencing. Exons: NM\_004543:7-8,18,25,28,33,36,45,48,54-55,58,61,71,73-74,91,94,101,111-112,114,118-119,122-123,127,129,132-135,138,140,143,146-147. **Detection Rate:** Unknown due to rarity of disease.

**Niemann-Pick Disease Type C** - Gene: NPC1. Autosomal Recessive. Sequencing. Exons: NM\_000271:1-25. **Detection Rate:** Northern European > 17%.

**Niemann-Pick Disease, SMPD1-associated** - Gene: SMPD1. Autosomal Recessive. Sequencing. Exons: NM\_000543:1-6. **Detection Rate:** Northern European > 38%.

**Nijmegen Breakage Syndrome** - Gene: NBN. Autosomal Recessive. Sequencing. Exons: NM\_002485:1-16. **Detection Rate:** Northern European > 78%.

**Northern Epilepsy** - Gene: CLN8. Autosomal Recessive. Sequencing. Exons: NM\_018941:2-3. **Detection Rate:** Unknown due to rarity of disease.

**Pendred Syndrome** - Gene: SLC26A4. Autosomal Recessive. Sequencing. Exons: NM\_000441:2-21. **Detection Rate:** Northern European > 69%.

**PEX1-related Zellweger Syndrome Spectrum** - Gene: PEX1. Autosomal Recessive. Sequencing. Exons: NM\_000466:1-24. **Detection Rate:** Northern European > 68%.

**Phenylalanine Hydroxylase Deficiency** - Gene: PAH. Autosomal Recessive. Sequencing. Exons: NM\_000277:1-13. **Detection Rate:** Northern European > 43%.

**Polyglandular Autoimmune Syndrome Type 1** - Gene: AIRE. Autosomal Recessive. Sequencing. Exons: NM\_000383:1-14. **Detection Rate:** Northern European > 65%.

**Pompe Disease** - Gene: GAA. Autosomal Recessive. Sequencing. Exons: NM\_000152:2-20. **Detection Rate:** Northern European > 67%.

**PPT1-related Neuronal Ceroid Lipofuscinosis** - Gene: PPT1. Autosomal Recessive. Sequencing. Exons: NM\_000310:1-9. **Detection Rate:** Northern European > 53%.

**Primary Carnitine Deficiency** - Gene: SLC22A5. Autosomal Recessive. Sequencing. Exons: NM\_003060:1-10. **Detection Rate:** Unknown due to rarity of disease.

**Primary Hyperoxaluria Type 1** - Gene: AGXT. Autosomal Recessive. Sequencing. Exons: NM\_000030:1-11. **Detection Rate:** Northern European > 42%.

**Primary Hyperoxaluria Type 2** - Gene: GRHPR. Autosomal Recessive. Sequencing. Exons: NM\_012203:1-9. **Detection Rate:** Northern European > 37%.

**PROP1-related Combined Pituitary Hormone Deficiency** - Gene: PROP1. Autosomal Recessive. Sequencing. Exons: NM\_006261:1-3. **Detection Rate:** Northern European > 55%.

**Pseudocholinesterase Deficiency** - Gene: BCHE. Autosomal Recessive. Sequencing. Exons: NM\_000055:2-4. **Detection Rate:** Northern European > 83%.

**Pycnodysostosis** - Gene: CTSK. Autosomal Recessive. Sequencing. Exons: NM\_000396:2-8. **Detection Rate:** Northern European > 10%.

**Rhizomelic Chondrodysplasia Punctata Type 1** - Gene: PEX7. Autosomal Recessive. Sequencing. Exons: NM\_000288:1-10. **Detection Rate:** Northern European > 70%.

**Salla Disease** - Gene: SLC17A5. Autosomal Recessive. Sequencing. Exons: NM\_012434:1-11. **Detection Rate:** Unknown due to rarity of disease.

**Segawa Syndrome** - Gene: TH. Autosomal Recessive. Sequencing. Exons: NM\_000360:1-13. **Detection Rate:** Northern European > 10%.

**Short Chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADS. Autosomal Recessive. Sequencing. Exons: NM\_000017:1-10. **Detection Rate:** Unknown due to rarity of disease.

**Sjogren-Larsson Syndrome** - Gene: ALDH3A2. Autosomal Recessive. Sequencing. Exons: NM\_000382:1-10. **Detection Rate:** Northern European > 24%.

**Smith-Lemli-Opitz Syndrome** - Gene: DHCR7. Autosomal Recessive. Sequencing. Exons: NM\_001360:3-9. **Detection Rate:** Northern European > 69%.

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

**Steroid-Resistant Nephrotic Syndrome** - Gene: NPHS2. Autosomal Recessive. Sequencing. Exons: NM\_014625:1-8. **Detection Rate:** Northern European > 33%.

**Sulfate Transporter-Related Osteochondrodysplasia** - Gene: SLC26A2. Autosomal Recessive. Sequencing. Exons: NM\_000112:2-3. **Detection Rate:** Northern European > 75%.

**TPP1-related Neuronal Ceroid Lipofuscinosis** - Gene: TPP1. Autosomal Recessive. Sequencing. Exons: NM\_000391:1-13. **Detection Rate:** Northern European > 60%.

**Tyrosinemia Type I** - Gene: FAH. Autosomal Recessive. Sequencing. Exons: NM\_000137:1-14. **Detection Rate:** Northern European > 50%.

**Usher Syndrome Type 1F** - Gene: PCDH15. Autosomal Recessive. Sequencing. Exons: NM\_033056:2-33. **Detection Rate:** Unknown due to rarity of disease.

**Usher Syndrome Type 3** - Gene: CLRN1. Autosomal Recessive. Sequencing. Exons: NM\_174878:1-3. **Detection Rate:** Unknown due to rarity of disease.

**Very Long Chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADVL. Autosomal Recessive. Sequencing. Exons: NM\_000018:1-20. **Detection Rate:** Northern European > 20%.

**Walker-Warburg Syndrome** - Gene: FKTN. Autosomal Recessive. Sequencing. Exons: NM\_001079802:3-11. **Detection Rate:** Unknown due to rarity of disease.

**Wilson Disease** - Gene: ATP7B. Autosomal Recessive. Sequencing. Exons: NM\_000053:1-21. **Detection Rate:** Northern European > 40%.

**X-Linked Juvenile Retinoschisis** - Gene: RS1. X-linked Recessive. Sequencing. Exons: NM\_000330:1-6. **Detection Rate:** Northern European > 20%.



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## 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 10057 Residual Risk	Reproductive Risk
<b>21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia</b>	1 in 1,400	1 in 310,000
<b>ABCC8-related Hyperinsulinism</b>	< 1 in 110	< 1 in 50,000
<b>Achromatopsia</b>	< 1 in 230	< 1 in 79,000
<b>Alkaptonuria</b>	< 1 in 500	< 1 in 1,000,000
<b>Alpha Thalassemia</b>	Alpha globin status: aa/aa.	Not calculated
<b>Alpha-1 Antitrypsin Deficiency</b>	< 1 in 680	< 1 in 93,000
<b>Alpha-Mannosidosis</b>	< 1 in 520	< 1 in 730,000
<b>Andermann Syndrome</b>	< 1 in 500	< 1 in 1,000,000
<b>ARSACS</b>	< 1 in 500	< 1 in 1,000,000
<b>Aspartylglycosaminuria</b>	< 1 in 500	< 1 in 1,000,000
<b>Ataxia With Vitamin E Deficiency</b>	< 1 in 500	< 1 in 1,000,000
<b>Ataxia-Telangiectasia</b>	< 1 in 450	< 1 in 290,000
<b>Autosomal Recessive Polycystic Kidney Disease</b>	< 1 in 75	< 1 in 18,000
<b>Bardet-Biedl Syndrome, BBS1-related</b>	< 1 in 750	< 1 in 480,000
<b>Bardet-Biedl Syndrome, BBS10-related</b>	< 1 in 290	< 1 in 180,000
<b>Biotinidase Deficiency</b>	< 1 in 220	< 1 in 110,000
<b>Bloom Syndrome</b>	< 1 in 500	< 1 in 1,000,000
<b>Canavan Disease</b>	< 1 in 500	< 1 in 1,000,000
<b>Carnitine Palmitoyltransferase IA Deficiency</b>	< 1 in 500	< 1 in 1,000,000
<b>Carnitine Palmitoyltransferase II Deficiency</b>	< 1 in 500	< 1 in 1,000,000
<b>Cartilage-Hair Hypoplasia</b>	< 1 in 500	< 1 in 1,000,000
<b>Choroideremia</b>	< 1 in 500	< 1 in 100,000
<b>Citrullinemia Type 1</b>	< 1 in 150	< 1 in 70,000
<b>CLN3-related Neuronal Ceroid Lipofuscinosis</b>	< 1 in 5,600	< 1 in 1,000,000
<b>CLN5-related Neuronal Ceroid Lipofuscinosis</b>	< 1 in 500	< 1 in 1,000,000
<b>Cohen Syndrome</b>	< 1 in 500	< 1 in 1,000,000
<b>Congenital Disorder of Glycosylation Type Ia</b>	< 1 in 560	< 1 in 360,000
<b>Congenital Disorder of Glycosylation Type Ib</b>	< 1 in 500	< 1 in 1,000,000
<b>Congenital Finnish Nephrosis</b>	< 1 in 500	< 1 in 1,000,000
<b>Costeff Optic Atrophy Syndrome</b>	< 1 in 500	< 1 in 1,000,000
<b>Cystic Fibrosis</b>	< 1 in 300	< 1 in 33,000
<b>Cystinosis</b>	< 1 in 670	< 1 in 600,000
<b>D-Bifunctional Protein Deficiency</b>	< 1 in 500	< 1 in 1,000,000
<b>Factor XI Deficiency</b>	< 1 in 500	< 1 in 1,000,000
<b>Familial Dysautonomia</b>	< 1 in 500	< 1 in 1,000,000
<b>Familial Mediterranean Fever</b>	< 1 in 500	< 1 in 1,000,000
<b>Fanconi Anemia Type C</b>	< 1 in 340	< 1 in 220,000
<b>Galactosemia</b>	< 1 in 430	< 1 in 150,000
<b>Gaucher Disease</b>	1 in 280	1 in 120,000
<b>GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness</b>	< 1 in 160	< 1 in 20,000
<b>Glutaric Acidemia Type 1</b>	< 1 in 170	< 1 in 67,000
<b>Glycogen Storage Disease Type Ia</b>	< 1 in 450	< 1 in 320,000
<b>Glycogen Storage Disease Type Ib</b>	< 1 in 660	< 1 in 930,000
<b>Glycogen Storage Disease Type III</b>	< 1 in 290	< 1 in 180,000
<b>Glycogen Storage Disease Type V</b>	< 1 in 790	< 1 in 500,000
<b>GRACILE Syndrome</b>	< 1 in 500	< 1 in 1,000,000
<b>Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)</b>	< 1 in 290	< 1 in 58,000
<b>Hereditary Fructose Intolerance</b>	< 1 in 320	< 1 in 100,000
<b>Hereditary Thymine-Uraciluria</b>	< 1 in 210	< 1 in 83,000



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
 Attn: Dr. Jeffrey Olliffe  
 NPI: 1306838271  
 Report Date: 07/20/2016

MALE  
**DONOR 10057**  
 DOB: [REDACTED]  
 Ethnicity: Northern European  
 Barcode: 55101509201147

FEMALE  
 N/A

Disease	DONOR 10057 Residual Risk	Reproductive Risk
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 500	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 500	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 500	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	< 1 in 390	< 1 in 470,000
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	< 1 in 290	< 1 in 290,000
Hurler Syndrome	1 in 480	1 in 300,000
Hypophosphatasia, Autosomal Recessive	< 1 in 230	< 1 in 140,000
Inclusion Body Myopathy 2	< 1 in 500	< 1 in 1,000,000
Isovaleric Acidemia	< 1 in 470	< 1 in 470,000
Joubert Syndrome 2	< 1 in 500	< 1 in 1,000,000
Krabbe Disease	< 1 in 360	< 1 in 210,000
Limb-Girdle Muscular Dystrophy Type 2D	< 1 in 660	< 1 in 1,000,000
Limb-Girdle Muscular Dystrophy Type 2E	< 1 in 500	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 500	< 1 in 1,000,000
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	< 1 in 1,200	< 1 in 690,000
Maple Syrup Urine Disease Type 1B	< 1 in 250	< 1 in 250,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 270	< 1 in 63,000
Megalencephalic Leukoencephalopathy With Subcortical Cysts	< 1 in 500	< 1 in 1,000,000
Metachromatic Leukodystrophy	< 1 in 430	< 1 in 340,000
Mucopolidosis IV	< 1 in 500	< 1 in 1,000,000
Muscle-Eye-Brain Disease	< 1 in 500	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 500	< 1 in 1,000,000
Niemann-Pick Disease Type C	< 1 in 230	< 1 in 180,000
Niemann-Pick Disease, SMPD1-associated	< 1 in 400	< 1 in 400,000
Nijmegen Breakage Syndrome	< 1 in 720	< 1 in 450,000
Northern Epilepsy	< 1 in 500	< 1 in 1,000,000
Pendred Syndrome	< 1 in 220	< 1 in 63,000
PEX1-related Zellweger Syndrome Spectrum	< 1 in 350	< 1 in 160,000
Phenylalanine Hydroxylase Deficiency	< 1 in 88	< 1 in 17,000
Polyglandular Autoimmune Syndrome Type 1	< 1 in 400	< 1 in 230,000
Pompe Disease	< 1 in 480	< 1 in 300,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 500	< 1 in 1,000,000
Primary Carnitine Deficiency	< 1 in 500	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	< 1 in 600	< 1 in 850,000
Primary Hyperoxaluria Type 2	< 1 in 500	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	< 1 in 250	< 1 in 110,000
Pseudocholinesterase Deficiency	< 1 in 160	< 1 in 18,000
Pycnodysostosis	< 1 in 500	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	< 1 in 530	< 1 in 330,000
Salla Disease	< 1 in 500	< 1 in 1,000,000
Segawa Syndrome	< 1 in 500	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 160	< 1 in 100,000
Sjogren-Larsson Syndrome	< 1 in 330	< 1 in 330,000
Smith-Lemli-Opitz Syndrome	< 1 in 160	< 1 in 32,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000
Steroid-Resistant Nephrotic Syndrome	< 1 in 600	< 1 in 950,000
Sulfate Transporter-Related Osteochondrodysplasia	< 1 in 420	< 1 in 180,000
TPP1-related Neuronal Ceroid Lipofuscinosis	< 1 in 740	< 1 in 870,000
Tyrosinemia Type I	< 1 in 350	< 1 in 240,000
Usher Syndrome Type 1F	< 1 in 190	< 1 in 150,000
Usher Syndrome Type 3	< 1 in 500	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 110	< 1 in 39,000
Walker-Warburg Syndrome	< 1 in 500	< 1 in 1,000,000
Wilson Disease	< 1 in 140	< 1 in 50,000
X-Linked Juvenile Retinoschisis	< 1 in 500	< 1 in 50,000