Counsyl

SEATTLE SPERM BANK
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Phone: (206) 588-1484
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NPI: 1306838271
Report Date: 12/18/2017

DONOR 10222
DOB: ▇▇▇▇▇▇▇
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 12/08/2017
Date Received: 12/11/2017
Date Tested: 12/18/2017
Barcode: 11004212319131
Accession ID:
CSL6GCMYLDDYHC9
Indication: Egg or sperm donor

Foresight™ Carrier Screen

POSITIVE: CARRIER

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 10222	Partner
Panel Information	Foresight Carrier Screen Universal Panel Minus X-Linked **(102 conditions tested)**	N/A
POSITIVE: CARRIER **Steroid-resistant Nephrotic Syndrome** Reproductive Risk: 1 in 1,600 Inheritance: Autosomal Recessive	➕ **CARRIER*** NM_014625.2(NPHS2):c.686G>A (R229Q) heterozygote †	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".

†Likely to have a negative impact on gene function.
*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 6.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner, as both parents must be carriers before a child is at high risk of developing the disease.
- Genetic counseling is recommended and patients may wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

Counsyl has renamed its products effective July 19, 2017. The Family Prep Screen is now the Foresight Carrier Screen. The new names now appear on all communications from Counsyl. If you have any questions, please contact Counsyl directly.

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RESULTS RECIPIENT	MALE	FEMALE
SEATTLE SPERM BANK	**DONOR 10222**	N/A
Attn: Dr. Jeffrey Olliffe	**DOB:** ▮▮▮▮▮▮	
NPI: 1306838271	**Ethnicity:** Northern European	
Report Date: 12/18/2017	**Barcode:** 11004212319131	

POSITIVE: CARRIER
Steroid-resistant Nephrotic Syndrome

Reproductive risk: 1 in 1,600
Risk before testing: 1 in 640,000

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

Patient	**DONOR 10222**	**No partner tested**
Result	➕ Carrier	N/A
Variant(s)	NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote [†]	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of steroid-resistant nephrotic syndrome. Carriers generally do not experience symptoms. The pathogenicity of R229Q is dependent on the variant observed on the other chromosome. In homozygous state, R229Q is not disease-causing.	N/A
Detection rate	>99%	N/A
Exons tested	NM_014625:1-8.	N/A

[†]Likely to have a negative impact on gene function.

What is Steroid-Resistant Nephrotic Syndrome?

Steroid-resistant nephrotic syndrome type 2 is a disease that causes significant abnormalities in kidney function, often leading to kidney failure.

The age at which symptoms begin varies; in some cases, symptoms have begun before age 2 while in others, symptoms did not appear until later in childhood.

Symptoms include an excess of protein in the urine, a shortage of protein in the blood, an excess of cholesterol and triglycerides in the blood, and generalized swelling in the body tissues. The water-retention that causes swelling can also cause weight gain and high blood pressure. The disease can cause scar tissue to form in the kidney's glomeruli, which are structures responsible for filtering waste products. This is known as focal segmental glomerulosclerosis.

The disease typically leads to kidney failure, necessitating transplantation in many before the age of 20. Even after receiving a kidney transplant, symptoms of the disease can recur. It is described as "steroid-resistant" because unlike other forms of nephritic syndrome, it does not respond to steroid medications.

The disease is caused by a mutation in the gene that provides the instructions for making podocin, a protein used by the kidney's glomeruli.

How common is Steroid-Resistant Nephrotic Syndrome?

The frequency of steroid-resistant nephritic syndrome type 2 is unknown. Several cases have been reported among Israeli-Arab children, however it has been found in other populations as well.

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How is Steroid-Resistant Nephrotic Syndrome treated?

The goal of treatment is to minimize damage to the kidneys, partially by controlling blood pressure. Medication may also be required for high cholesterol. Often children with steroid-resistant nephritic syndrome require kidney transplants. They many also need medication to control for infection.

What is the prognosis for a person with Steroid-Resistant Nephrotic Syndrome?

The prognosis for a person with steroid-resistant nephritic syndrome type 2 is varied, however with transplantation and careful medical management, these children can live into adulthood.

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Methods and Limitations

DONOR 10222 [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.

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Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. 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

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

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Conditions Tested

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%.

ABCC8-related Hyperinsulinism - **Gene:** ABCC8. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000352:1-39. **Detection Rate:** Northern European >99%.

Alkaptonuria - **Gene:** HGD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000187:1-14. **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-1 Antitrypsin Deficiency - **Gene:** SERPINA1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000295:2-5. **Detection Rate:** Northern European >99%.

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%.

Andermann Syndrome - **Gene:** SLC12A6. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_133647:1-25. **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%.

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%.

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%.

Canavan Disease - **Gene:** ASPA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000049:1-6. **Detection Rate:** Northern European 98%.

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%.

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%.

CNGB3-related Achromatopsia - **Gene:** CNGB3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_019098:1-18. **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%.

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 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%.

Dihydropyrimidine Dehydrogenase Deficiency - **Gene:** DPYD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000110:1-23. **Detection Rate:** Northern European 98%.

Factor XI Deficiency - **Gene:** F11. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000128:2-15. **Detection Rate:** Northern European >99%.

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 Type C - **Gene:** FANCC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000136:2-15. **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%.

Galactosemia - **Gene:** GALT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000155:1-11. **Detection Rate:** Northern European >99%.

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%.

Glutaric Acidemia Type 1 - **Gene:** GCDH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000159:2-12. **Detection Rate:** Northern European >99%.

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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%.

Glycogen Storage Disease Type V - **Gene:** PYGM. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_005609:1-20. **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%.

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%.

Hypophosphatasia, 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%.

Krabbe Disease - **Gene:** GALC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000153:1-17. **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%.

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%.

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%.

Mucolipidosis 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%.

Muscle-eye-brain Disease - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017739:2-22. **Detection Rate:** Northern European 96%.

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, 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%.

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%.

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%.

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%.

Pseudocholinesterase Deficiency - **Gene:** BCHE. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000055:2-4. **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%.

Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000288:1-10. **Detection Rate:** Northern European >99%.

Counsyl

RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 12/18/2017

MALE
DONOR 10222
DOB: ▮▮▮▮▮▮
Ethnicity: Northern European
Barcode: 11004212319131

FEMALE
N/A

Salla Disease - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_012434:1-11. **Detection Rate:** Northern European 98%.

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%.

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

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%.

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%.

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%.

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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.

†Indicates a positive result. See the full clinical report for interpretation and details.

Disease	DONOR 10222 Residual Risk	Reproductive Risk
21-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 310,000
ABCC8-related Hyperinsulinism	1 in 11,000	< 1 in 1,000,000
Alkaptonuria	1 in 6,800	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-1 Antitrypsin Deficiency	1 in 2,700	1 in 300,000
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 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
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
Beta-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 670,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Canavan Disease	< 1 in 31,000	< 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
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
CNGB3-related Achromatopsia	1 in 11,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 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 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
Dihydropyrimidine Dehydrogenase Deficiency	< 1 in 29,000	< 1 in 1,000,000
Factor XI Deficiency	< 1 in 50,000	< 1 in 1,000,000
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 Type C	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 3,200	1 in 420,000
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
Glycogen Storage Disease Type V	1 in 16,000	< 1 in 1,000,000
GRACILE Syndrome	< 1 in 50,000	< 1 in 1,000,000

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Disease	DONOR 10222 Residual Risk	Reproductive Risk
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 5,000	1 in 990,000
Hereditary Fructose Intolerance	1 in 8,000	< 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
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency	1 in 25,000	< 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
Krabbe Disease	1 in 15,000	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 25,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 5,900	< 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
Mucolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Muscle-eye-brain Disease	< 1 in 12,000	< 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, 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
PCDH15-related Disorders	1 in 5,300	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	< 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
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	< 1 in 1,000,000
Pseudocholinesterase Deficiency (Mild Condition)	1 in 2,700	1 in 300,000
Pycnodysostosis	< 1 in 50,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
Salla 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 4,900	1 in 970,000
Spinal Muscular Atrophy	SMN1: 3+ copies 1 in 4,800	1 in 670,000
Steroid-resistant Nephrotic Syndrome	NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote †	1 in 1,600
Sulfate Transporter-related Osteochondrodysplasia	1 in 11,000	< 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
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