Results Recipient	Male	Female
Seattle Sperm Bank Attn: Dr. Jeffrey Olliffe 4915 25th Ave E, Suite 204W Seattle, WA 98105 Phone: (206) 588-1484 Fax: (206) 588-1484 NPI: 1306838271 Report Date: 11/20/2013	Name: DONOR 9684 DOB: [redacted] Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 11/16/2013 Date Received: 11/19/2013 Barcode: 11004211305579 Indication: Egg or Sperm Donor Test Type: The Counsyl Test	Not tested

Counsyl Test Results Summary (Egg or Sperm Donor)

The Counsyl test (**Universal Panel**) uses targeted genotyping and copy number analysis as described in the methods section on page 2 to determine carrier status associated with **101 diseases**. Please refer to page 3 for a complete list of diseases and genes included in this panel.

DONOR 9684

DONOR 9684's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Reproductive Risk Summary

No increased reproductive risks to highlight. Please refer to the following pages for detailed information about the results.

Clinical Notes

- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complimentary appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

Male	Female
Name: DONOR 9684 DOB: [redacted]	Not tested

Methods and Limitations

DONOR 9684: The Counsyl Test - targeted genotyping and copy number analysis.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 398 variants associated with 100 diseases. 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 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.

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. The Counsyl 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*) 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 workup. CLIA Number: **#05D1102604**.

Lab Directors:

H. Peter Kang, MD, MS, FCAP

Jelena Brezo, PhD, FACMG

Male	Female
Name: DONOR 9684 DOB: [redacted]	Not tested

Diseases Tested

ABCC8-Related Hyperinsulinism - **Gene:** ABCC8. **Variants (3):** F1388del, V187D, 3992-9G>A. **Detection rate**: Northern European <10%.

Achromatopsia - **Gene:** CNGB3. **Variants (3):** R403Q, 819_826del8, T383fs. **Detection rate**: Northern European 62%.

Alkaptonuria - **Gene:** HGD. **Variants (11):** G161R, G270R, P230S, S47L, V300G, M368V, IVS1-1G>A, IVS5+1G>A, G152fs, R58fs, 1111_1112insC. **Detection rate**: Northern European 80%.

Alpha-1 Antitrypsin Deficiency - **Gene:** SERPINA1. **Variant (1):** Z allele. **Detection rate**: Northern European 95%.

Alpha-Mannosidosis - **Gene:** MAN2B1. **Variant (1):** R750W. **Detection rate**: Northern European 32%.

Andermann Syndrome - **Gene:** SLC12A6. **Variants (2):** Thr813fsX813, R1011X. **Detection rate**: Northern European <10%.

ARSACS - **Gene:** SACS. **Variants (2):** 6594delT, 5254C>T. **Detection rate**: Northern European <10%.

Aspartylglycosaminuria - **Gene:** AGA. **Variant (1):** C163S. **Detection rate**: Northern European <10%.

Ataxia With Vitamin E Deficiency - **Gene:** TTPA. **Variant (1):** 744delA. **Detection rate**: Northern European <10%.

Ataxia-Telangiectasia - **Gene:** ATM. **Variants (8):** R35X, Q1970X, 7517del4, 5762ins137, 2546_2548del, 3245ATC>TGAT, K1192K, E1978X. **Detection rate**: Northern European 65%.

Autosomal Recessive Polycystic Kidney Disease - **Gene:** PKHD1. **Variants (4):** Leu1965fs, T36M, R496X, V3471G. **Detection rate**: Northern European 18%.

Bardet-Biedl Syndrome, BBS1-Related - **Gene:** BBS1. **Variant (1):** M390R. **Detection rate**: Northern European 79%.

Bardet-Biedl Syndrome, BBS10-Related - **Gene:** BBS10. **Variant (1):** C91fs. **Detection rate**: Northern European 46%.

Biotinidase Deficiency - **Gene:** BTD. **Variants (4):** G98:d7i3, D252G, Q456H, R538C. **Detection rate**: Northern European 45%.

Bloom Syndrome - **Gene:** BLM. **Variant (1):** 2281del6ins7. **Detection rate**: Northern European <10%.

Canavan Disease - **Gene:** ASPA. **Variants (4):** E285A, Y231X, A305E, IVS2-2A>G. **Detection rate**: Northern European 53%.

Carnitine Palmitoyltransferase IA Deficiency - **Gene:** CPT1A. **Variant (1):** G710E. **Detection rate**: Northern European <10%.

Carnitine Palmitoyltransferase II Deficiency - **Gene:** CPT2. **Variants (3):** Q413fs, S113L, R124X. **Detection rate**: Northern European 80%.

Cartilage-Hair Hypoplasia - **Gene:** RMRP. **Variant (1):** g.70A>G. **Detection rate**: Northern European 48%.

Choroideremia - **Gene:** CHM. **Variant (1):** IVS13+2dupT. **Detection rate**: Northern European <10%.

Citrullinemia Type 1 - **Gene:** ASS1. **Variants (2):** IVS6-2A>G, G390R. **Detection rate**: Northern European 20%.

CLN3-Related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN3. **Variant (1):** 461_677del. **Detection rate**: Northern European 96%.

CLN5-Related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN5. **Variant (1):** 2467AT. **Detection rate**: Northern European <10%.

Cohen Syndrome - **Gene:** VPS13B. **Variant (1):** 3348_3349delCT. **Detection rate**: Northern European <10%.

Congenital Disorder of Glycosylation Type Ia - **Gene:** PMM2. **Variants (4):** V231M, F119L, R141H, P113L. **Detection rate**: Northern European 72%.

Congenital Disorder of Glycosylation Type Ib - **Gene:** MPI. **Variant (1):** R295H. **Detection rate**: Northern European <10%.

Congenital Finnish Nephrosis - **Gene:** NPHS1. **Variants (2):** 121_122del, R1109X. **Detection rate**: Northern European <10%.

Costeff Optic Atrophy Syndrome - **Gene:** OPA3. **Variant (1):** 143-1G>C. **Detection rate**: Northern European <10%.

Cystic Fibrosis - **Gene:** CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, E92X, Y122X, G178R, R347H, Q493X, V520F, S549N, P574H, M1101K, D1152H, 2143delT, 394delTT, 444delA, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), R117C, L206W, G330X, T338I, R352Q, S364P, G480C, C524X, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, R764X, Q890X, R1066C, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, 3199del6, 574delA, 663delT, 935delA, 936delTA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667del4, 3791delC, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, 3849+4A>G, Q359K/T360K. **Detection rate**: Northern European 91%.

Cystinosis - **Gene:** CTNS. **Variants (4):** 57 kb deletion, 537del21, W138X, L158P. **Detection rate**: Northern European 67%.

D-Bifunctional Protein Deficiency - **Gene:** HSD17B4. **Variants (2):** G16S, N457Y. **Detection rate**: Northern European 35%.

Factor XI Deficiency - **Gene:** F11. **Variants (4):** E117X, F283L, IVS14+1G>A, IVS14del14. **Detection rate**: Northern European <10%.

Familial Dysautonomia - **Gene:** IKBKAP. **Variants (2):** IVS20+6T>C, R696P. **Detection rate**: Northern European <10%.

Familial Mediterranean Fever - **Gene:** MEFV. **Variants (4):** M694V, V726A, M680I, M694I. **Detection rate**: Northern European <10%.

Fanconi Anemia Type C - **Gene:** FANCC. **Variants (3):** IVS4+4A>T, 322delG, R548X. **Detection rate**: Northern European 54%.

Galactosemia - **Gene:** GALT. **Variants (8):** S135L, Q188R, F171S, L195P, K285N, IVS2-2A>G, T138M, Y209C. **Detection rate**: Northern European 80%.

Gaucher Disease - **Gene:** GBA. **Variants (10):** N370S, L444P, 84GG, IVS2+1G>A, V394L, R496H, D409H, D409V, R463C, R463H. **Detection rate**: Northern European 60%.

GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness - **Gene:** GJB2. **Variants (7):** 35delG, 167delT, 235delC, E120del, W24X, W77R, L90P. **Detection rate**: Northern European 79%.

Glutaric Acidemia Type 1 - **Gene:** GCDH. **Variant (1):** R402W. **Detection rate**: Northern European 40%.

Glycogen Storage Disease Type Ia - **Gene:** G6PC. **Variants (7):** R83C, Q347X, Q27fsdelC, 459insTA, R83H, G188R, Q242X. **Detection rate**: Northern European 61%.

Glycogen Storage Disease Type Ib - **Gene:** SLC37A4. **Variants (2):** 1211delCT, G339C. **Detection rate**: Northern European 46%.

Glycogen Storage Disease Type III - **Gene:** AGL. **Variants (3):** 1484delT, Q6X, 17delAG. **Detection rate**: Northern European 45%.

Glycogen Storage Disease Type V - **Gene:** PYGM. **Variants (4):** R49X, G204S, 708/709del, W797R. **Detection rate**: Northern European 80%.

GRACILE Syndrome - **Gene:** BCS1L. **Variant (1):** S78G. **Detection rate**: Northern European <10%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - **Gene:** HBB. **Variants (28):** Hb S, K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab. **Detection rate**: Northern European 83%.

Hereditary Fructose Intolerance - **Gene:** ALDOB. **Variants (3):** A149P, N334K, A174D. **Detection rate**: Northern European 75%.

Hereditary Thymine-Uraciluria - **Gene:** DPYD. **Variant (1):** IVS14+1G>A. **Detection rate**: Northern European 52%.

Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related - **Gene:** LAMA3. **Variant (1):** R650X. **Detection rate**: Northern European <10%.

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related - **Gene:** LAMB3. **Variants (3):** R42X, Q243X, R635X. **Detection rate**: Northern European 48%.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related - **Gene:** LAMC2. **Variant (1):** R95X. **Detection rate**: Northern European <10%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. **Variants (9):** 1278insTATC, IVS12+1G>C, G269S, IVS9+1G>A, R178H, IVS7+1G>A, 7.6kb del, G250D, R170W. **Detection rate**: Northern European 23%.

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency - **Gene:** CBS. **Variant (1):** G307S. **Detection rate**: Northern European 14%.

Hurler Syndrome - **Gene:** IDUA. **Variants (2):** W402X, Q70X. **Detection rate**: Northern European 67%.

Male	Female
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Hypophosphatasia, Autosomal Recessive - **Gene:** ALPL. **Variants (4):** 1559delT, F310L, D361V, E174K. **Detection rate**: Northern European 30%.

Inclusion Body Myopathy 2 - **Gene:** GNE. **Variants (2):** M712T, V572L. **Detection rate**: Northern European <10%.

Isovaleric Acidemia - **Gene:** IVD. **Variant (1):** A311V. **Detection rate**: Northern European 47%.

Joubert Syndrome 2 - **Gene:** TMEM216. **Variant (1):** 35G>T. **Detection rate**: Northern European <10%.

Krabbe Disease - **Gene:** GALC. **Variants (2):** Ex11-17del, T513M. **Detection rate**: Northern European 58%.

Limb-Girdle Muscular Dystrophy Type 2D - **Gene:** SGCA. **Variant (1):** R77C. **Detection rate**: Northern European 32%.

Limb-Girdle Muscular Dystrophy Type 2E - **Gene:** SGCB. **Variant (1):** S114F. **Detection rate**: Northern European 12%.

Lipoamide Dehydrogenase Deficiency - **Gene:** DLD. **Variants (2):** 105insA, G229C. **Detection rate**: Northern European <10%.

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency - **Gene:** HADHA. **Variant (1):** E474Q. **Detection rate**: Northern European 87%.

Maple Syrup Urine Disease Type 1B - **Gene:** BCKDHB. **Variants (3):** R183P, G278S, E372X. **Detection rate**: Northern European <10%.

Medium Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. **Variants (2):** K304E, Y42H. **Detection rate**: Northern European 78%.

Megalencephalic Leukoencephalopathy With Subcortical Cysts - **Gene:** MLC1. **Variants (4):** 135insC, c.176G>A, c.278C>T, IVS2-10T>A. **Detection rate**: Northern European 13%.

Metachromatic Leukodystrophy - **Gene:** ARSA. **Variants (5):** P426L, IVS2+1G>A, c.1204+1G>A, I179S, p.Thr409Ile. **Detection rate**: Northern European 53%.

Mucolipidosis IV - **Gene:** MCOLN1. **Variants (2):** 511_6944del, IVS3-2A>G. **Detection rate**: Northern European <10%.

Muscle-Eye-Brain Disease - **Gene:** POMGNT1. **Variant (1):** IVS17+1G>A. **Detection rate**: Northern European 75%.

NEB-Related Nemaline Myopathy - **Gene:** NEB. **Variant (1):** R2478_D2512del. **Detection rate**: Northern European <10%.

Niemann-Pick Disease Type C - **Gene:** NPC1. **Variant (1):** I1061T. **Detection rate**: Northern European 17%.

Niemann-Pick Disease, SMPD1-Associated - **Gene:** SMPD1. **Variants (4):** fsP330, L302P, R496L, p.R608del. **Detection rate**: Northern European 38%.

Nijmegen Breakage Syndrome - **Gene:** NBN. **Variant (1):** 657del5. **Detection rate**: Northern European 78%.

Northern Epilepsy - **Gene:** CLN8. **Variant (1):** R24G. **Detection rate**: Northern European <10%.

Pendred Syndrome - **Gene:** SLC26A4. **Variants (5):** IVS8+1G>A, L236P, E384G, T416P, H723R. **Detection rate**: Northern European 69%.

PEX1-Related Zellweger Syndrome Spectrum - **Gene:** PEX1. **Variants (2):** 2097_2098insT, G843D. **Detection rate**: Northern European 68%.

Phenylalanine Hydroxylase Deficiency - **Gene:** PAH. **Variants (13):** IVS-10int-546, I65T, R261Q, R408W, IVS12+1G>A, R408Q, Y414C, L48S, R158Q, G272X, P281L, E280K, S349P. **Detection rate**: Northern European 43%.

Polyglandular Autoimmune Syndrome Type 1 - **Gene:** AIRE. **Variants (2):** Y85C, R257X. **Detection rate**: Northern European 65%.

Pompe Disease - **Gene:** GAA. **Variants (4):** D645E, R854X, IVS1-13T>G, 525delT. **Detection rate**: Northern European 67%.

PPT1-Related Neuronal Ceroid Lipofuscinosis - **Gene:** PPT1. **Variants (3):** T75P, R122W, R151X. **Detection rate**: Northern European 53%.

Primary Carnitine Deficiency - **Gene:** SLC22A5. **Variant (1):** 760C>T. **Detection rate**: Northern European <10%.

Primary Hyperoxaluria Type 1 - **Gene:** AGXT. **Variants (2):** G170R, I244T. **Detection rate**: Northern European 42%.

Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. **Variants (2):** 103delG, c.403_405+2delAAGT. **Detection rate**: Northern European 37%.

PROP1-Related Combined Pituitary Hormone Deficiency - **Gene:** PROP1. **Variant (1):** Ser101fs. **Detection rate**: Northern European 55%.

Pseudocholinesterase Deficiency - **Gene:** BCHE. **Variant (1):** D70G. **Detection rate**: Northern European 83%.

Pycnodysostosis - **Gene:** CTSK. **Variant (1):** X330W. **Detection rate**: Northern European <10%.

Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. **Variants (4):** G217R, A218V, L292X, IVS9+1G>C. **Detection rate**: Northern European 70%.

Salla Disease - **Gene:** SLC17A5. **Variant (1):** R39C. **Detection rate**: Northern European <10%.

Segawa Syndrome - **Gene:** TH. **Variant (1):** R233H. **Detection rate**: Northern European <10%.

Short Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADS. **Variant (1):** R107C. **Detection rate**: Northern European <10%.

Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. **Variant (1):** P315S. **Detection rate**: Northern European 24%.

Smith-Lemli-Opitz Syndrome - **Gene:** DHCR7. **Variants (13):** IVS8-1G>C, T93M, W151X(c.452G>A), V326L, R352Q, R352W, R404C, S169L, R242C, R242H, F302L, G410S, E448L. **Detection rate**: Northern European 69%.

Spinal Muscular Atrophy (copy number analysis only) - **Gene:** SMN1. **Variant (1):** SMN1 copy number. **Detection rate**: Northern European 95%.

Steroid-Resistant Nephrotic Syndrome - **Gene:** NPHS2. **Variants (2):** R138Q, R138X. **Detection rate**: Northern European 33%.

Sulfate Transporter-Related Osteochondrodysplasia - **Gene:** SLC26A2. **Variants (4):** C653S, R178X, R279W, IVS1+2T>C. **Detection rate**: Northern European 75%.

TPP1-Related Neuronal Ceroid Lipofuscinosis - **Gene:** TPP1. **Variants (3):** G284V, R208X, IVS5-1G>C. **Detection rate**: Northern European 60%.

Tyrosinemia Type I - **Gene:** FAH. **Variants (6):** IVS12+5G>A, Q64H, P261L, W262X, E357X, IVS6-1G>T. **Detection rate**: Northern European 50%.

Usher Syndrome Type 1F - **Gene:** PCDH15. **Variant (1):** R245X. **Detection rate**: Northern European <10%.

Usher Syndrome Type 3 - **Gene:** CLRN1. **Variant (1):** N48K. **Detection rate**: Northern European <10%.

Very Long Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADVL. **Variant (1):** V283A. **Detection rate**: Northern European 20%.

Wilson Disease - **Gene:** ATP7B. **Variants (2):** H1069Q, R778L. **Detection rate**: Northern European 40%.

X-Linked Juvenile Retinoschisis - **Gene:** RS1. **Variants (3):** E72K, G74V, G109R. **Detection rate**: Northern European 20%.

Male	Female
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Risk Calculations

Below are the risk calculations for all diseases 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.

Disease	DONOR 9684 Residual Risk	Reproductive Risk
ABCC8-Related Hyperinsulinism	1 in 110	1 in 50,000
Achromatopsia	1 in 230	1 in 79,000
Alkaptonuria	< 1 in 500	< 1 in 1,000,000
Alpha-1 Antitrypsin Deficiency	1 in 680	1 in 93,000
Alpha-Mannosidosis	1 in 520	1 in 730,000
Andermann Syndrome	< 1 in 500	< 1 in 1,000,000
ARSACS	< 1 in 500	< 1 in 1,000,000
Aspartylglycosaminuria	< 1 in 500	< 1 in 1,000,000
Ataxia With Vitamin E Deficiency	< 1 in 500	< 1 in 1,000,000
Ataxia-Telangiectasia	1 in 450	1 in 290,000
Autosomal Recessive Polycystic Kidney Disease	1 in 75	1 in 18,000
Bardet-Biedl Syndrome, BBS1-Related	1 in 750	1 in 480,000
Bardet-Biedl Syndrome, BBS10-Related	1 in 290	1 in 180,000
Biotinidase Deficiency	1 in 220	1 in 110,000
Bloom Syndrome	< 1 in 500	< 1 in 1,000,000
Canavan Disease	< 1 in 500	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 500	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	< 1 in 500	< 1 in 1,000,000
Cartilage-Hair Hypoplasia	< 1 in 500	< 1 in 1,000,000
Choroideremia	< 1 in 500	1 in 100,000
Citrullinemia Type 1	1 in 150	1 in 70,000
CLN3-Related Neuronal Ceroid Lipofuscinosis	1 in 5,600	< 1 in 1,000,000
CLN5-Related Neuronal Ceroid Lipofuscinosis	< 1 in 500	< 1 in 1,000,000
Cohen Syndrome	< 1 in 500	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	1 in 560	1 in 360,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 500	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 500	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 500	< 1 in 1,000,000
Cystic Fibrosis	1 in 300	1 in 33,000
Cystinosis	1 in 670	1 in 600,000
D-Bifunctional Protein Deficiency	< 1 in 500	< 1 in 1,000,000
Factor XI Deficiency	< 1 in 500	< 1 in 1,000,000
Familial Dysautonomia	< 1 in 500	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 500	< 1 in 1,000,000
Fanconi Anemia Type C	1 in 340	1 in 220,000
Galactosemia	1 in 430	1 in 150,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 160	1 in 20,000
Glutaric Acidemia Type 1	1 in 170	1 in 67,000
Glycogen Storage Disease Type Ia	1 in 450	1 in 320,000
Glycogen Storage Disease Type Ib	1 in 660	1 in 930,000
Glycogen Storage Disease Type III	1 in 290	1 in 180,000
Glycogen Storage Disease Type V	1 in 790	1 in 500,000
GRACILE Syndrome	< 1 in 500	< 1 in 1,000,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Hereditary Fructose Intolerance	1 in 320	1 in 100,000
Hereditary Thymine-Uraciluria	1 in 210	1 in 83,000
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

Male	Female
Name: DONOR 9684 DOB: [redacted]	Not tested

Disease	DONOR 9684 Residual Risk	Reproductive Risk
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
Mucolipidosis 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
Wilson Disease	1 in 140	1 in 50,000
X-Linked Juvenile Retinoschisis	< 1 in 500	1 in 50,000

Results Recipient	Male	Female
Seattle Sperm Bank Attn: Dr. Jeffrey Olliffe 4915 25th Ave E, Suite 204W Seattle, WA 98105 Phone: (206) 588-1484 Fax: (206) 588-1484 NPI: 1306838271 Report Date: 06/13/2014	Name: DONOR 9684 DOB: [redacted] Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 05/28/2014 Date Received: 05/30/2014 Date Tested: 06/13/2014 Barcode: 11004211381214 Indication: Egg or Sperm Donor Test Type: The Counsyl Test 2.0	Not tested

Counsyl Test Results Summary (Egg or Sperm Donor)

The Counsyl test (**Cystic Fibrosis Panel**) uses sequencing and targeted genotyping as described in the methods section on page 2 to determine carrier status associated with **1 disease**. Please refer to page 3 for a complete list of diseases and genes included in this panel.

DONOR 9684

DONOR 9684's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Reproductive Risk Summary

No increased reproductive risks to highlight. Please refer to the following pages for detailed information about the results.

Clinical Notes

- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complimentary appointment to speak with a clinical expert about these results, please visit counsyl.com/my/consults/.

Methods and Limitations

DONOR 9684: The Counsyl Test 2.0 - sequencing and targeted genotyping.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 99 variants associated with 1 disease. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

Sequencing: High-throughput sequencing is used to analyze 27 exons in 1 gene, as well as selected intergenic and intronic regions. 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.

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 Counsyl 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.

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 workup. CLIA Number: **#05D1102604**.

Lab Directors:

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Jelena Brezo

Jelena Brezo, PhD, FACMG

Male	Female
Name: DONOR 9684 DOB: [redacted]	Not tested

Diseases Tested

Cystic Fibrosis - **Gene:** CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542*, G551D, R553*, R560T, R1162*, W1282*, N1303K, c.1521_1523delCTT, c.1519_1521delATC, c.2052delA, c.3528delC, c.489+1G>T, c.579+1G>T, c.1585-1G>A, c.1766+1G>A, 2789+5G>A, c.2988+1G>A, 3849+10kbC>T, E60*, R75*, E92*, Y122*, G178R, R347H, Q493*, V520F, S549N, P574H, M1101K, D1152H, c.2012delT, c.262_263delTT, c.313delA, c.948delT, c.3744delA, c.3773dupT, c.1680-1G>A, 3272-26A>G, c.2051_2052delAAinsG, S549R, R117C, L206W, G330*, T338I, R352Q, S364P, G480C, C524*, S549R, Q552*, A559T, G622D, R709*, K710*, R764*, Q890*, R1066C, W1089*, Y1092X, R1158*, S1196*, W1204*, Q1238*, S1251N, S1255*, c.3067_3072del6, c.442delA, c.531delT, c.803delA, c.805_806delAT, c.1545_1546delTA, 1949del84, c.1911delG, c.1923_1931del9ins1, c.1976delA, c.3039delC, c.3536_3539delCCAA, c.3659delC, c.1155_1156dupTA, c.2052dupA, c.2175dupA, c.2738insG, 296+12T>C, c.273+1G>A, 405+3A>C, c.274-1G>A, 711+5G>A, c.580-1G>T, c.1766+1G>T, 1898+5G>T, Q996, c.325_327delTATinsG, 3849+4A>G, c.1075_1079del5ins5. **Exons:** NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection rate**: Northern European > 91%.

Male	Female
Name: DONOR 9684 DOB: [redacted]	Not tested

Risk Calculations

Below are the risk calculations for all diseases 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.

Disease	DONOR 9684 Residual Risk	Reproductive Risk
Cystic Fibrosis	< 1 in 300	< 1 in 33,000