Results Recipient	Male	Female
NW Cryobank Attn: Dr. Edwin Robins 508 West 6th Avenue Suite 801 Spokane, WA 99204 Phone: (509) 232-0132 Fax: (509) 232-0145 NPI: 1588748933 Report Date: 09/18/2013	Name: DONOR 3243 DOB: 01/01/1901 Ethnicity: Middle Eastern Sample Type: OG-510 Saliva Date of Collection: 09/11/2013 Date Received: 09/16/2013 Barcode: 55101211090716 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 3243

DONOR 3243'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/.

Methods and Limitations

DONOR 3243: 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:

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Male	Female
Name: DONOR 3243 DOB: 01/01/1901	Not tested

Diseases Tested

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

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

Alkaptonuria - **Gene:** HGD. **Variants (11):** G161R, G270R, P230S, S47L, V300G, M368V, IVS1-1G>A, IVS5+1G>A, G152fs, R58fs, 1111_1112insC. **Detection rate**: Middle Eastern <10%.

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

Alpha-Mannosidosis - **Gene:** MAN2B1. **Variant (1):** R750W. **Detection rate**: Middle Eastern <10%.

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

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

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

Ataxia With Vitamin E Deficiency - **Gene:** TTPA. **Variant (1):** 744delA. **Detection rate**: Middle Eastern 83%.

Ataxia-Telangiectasia - **Gene:** ATM. **Variants (8):** R35X, Q1970X, 7517del4, 5762ins137, 2546_2548del, 3245ATC>TGAT, K1192K, E1978X. **Detection rate**: Middle Eastern 33%.

Autosomal Recessive Polycystic Kidney Disease - **Gene:** PKHD1. **Variants (4):** Leu1965fs, T36M, R496X, V3471G. **Detection rate**: Middle Eastern <10%.

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

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

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

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

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

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

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

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

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

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

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

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

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

Congenital Disorder of Glycosylation Type Ia - **Gene:** PMM2. **Variants (4):** V231M, F119L, R141H, P113L. **Detection rate**: Middle Eastern <10%.

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

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

Costeff Optic Atrophy Syndrome - **Gene:** OPA3. **Variant (1):** 143-1G>C. **Detection rate**: Middle Eastern >99%.

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**: Middle Eastern 55%.

Cystinosis - **Gene:** CTNS. **Variants (4):** 57 kb deletion, 537del21, W138X, L158P. **Detection rate**: Middle Eastern <10%.

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

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

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

Familial Mediterranean Fever - **Gene:** MEFV. **Variants (4):** M694V, V726A, M680I, M694I. **Detection rate**: Middle Eastern 96%.

Fanconi Anemia Type C - **Gene:** FANCC. **Variants (3):** IVS4+4A>T, 322delG, R548X. **Detection rate**: Middle Eastern <10%.

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

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

GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness - **Gene:** GJB2. **Variants (7):** 35delG, 167delT, 235delC, E120del, W24X, W77R, L90P. **Detection rate**: Middle Eastern 65%.

Glutaric Acidemia Type 1 - **Gene:** GCDH. **Variant (1):** R402W. **Detection rate**: Middle Eastern 12%.

Glycogen Storage Disease Type Ia - **Gene:** G6PC. **Variants (7):** R83C, Q347X, Q27fsdelC, 459insTA, R83H, G188R, Q242X. **Detection rate**: Middle Eastern 30%.

Glycogen Storage Disease Type Ib - **Gene:** SLC37A4. **Variants (2):** 1211delCT, G339C. **Detection rate**: Middle Eastern <10%.

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

Glycogen Storage Disease Type V - **Gene:** PYGM. **Variants (4):** R49X, G204S, 708/709del, W797R. **Detection rate**: Middle Eastern <10%.

GRACILE Syndrome - **Gene:** BCS1L. **Variant (1):** S78G. **Detection rate**: Middle Eastern <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**: Middle Eastern 72%.

Hereditary Fructose Intolerance - **Gene:** ALDOB. **Variants (3):** A149P, N334K, A174D. **Detection rate**: Middle Eastern <10%.

Hereditary Thymine-Uraciluria - **Gene:** DPYD. **Variant (1):** IVS14+1G>A. **Detection rate**: Middle Eastern <10%.

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

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

Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related - **Gene:** LAMC2. **Variant (1):** R95X. **Detection rate**: Middle Eastern <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**: Middle Eastern 23%.

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

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

Male	Female
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Hypophosphatasia, Autosomal Recessive - **Gene:** ALPL. **Variants (4):** 1559delT, F310L, D361V, E174K. **Detection rate**: Middle Eastern <10%.

Inclusion Body Myopathy 2 - **Gene:** GNE. **Variants (2):** M712T, V572L. **Detection rate**: Middle Eastern 99%.

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

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

Krabbe Disease - **Gene:** GALC. **Variants (2):** Ex11-17del, T513M. **Detection rate**: Middle Eastern <10%.

Limb-Girdle Muscular Dystrophy Type 2D - **Gene:** SGCA. **Variant (1):** R77C. **Detection rate**: Middle Eastern <10%.

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

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

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency - **Gene:** HADHA. **Variant (1):** E474Q. **Detection rate**: Middle Eastern <10%.

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

Medium Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. **Variants (2):** K304E, Y42H. **Detection rate**: Middle Eastern <10%.

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

Metachromatic Leukodystrophy - **Gene:** ARSA. **Variants (5):** P426L, IVS2+1G>A, c.1204+1G>A, I179S, p.Thr409Ile. **Detection rate**: Middle Eastern <10%.

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

Muscle-Eye-Brain Disease - **Gene:** POMGNT1. **Variant (1):** IVS17+1G>A. **Detection rate**: Middle Eastern 15%.

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

Niemann-Pick Disease Type C - **Gene:** NPC1. **Variant (1):** I1061T. **Detection rate**: Middle Eastern 15%.

Niemann-Pick Disease, SMPD1-Associated - **Gene:** SMPD1. **Variants (4):** fsP330, L302P, R496L, p.R608del. **Detection rate**: Middle Eastern <10%.

Nijmegen Breakage Syndrome - **Gene:** NBN. **Variant (1):** 657del5. **Detection rate**: Middle Eastern <10%.

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

Pendred Syndrome - **Gene:** SLC26A4. **Variants (5):** IVS8+1G>A, L236P, E384G, T416P, H723R. **Detection rate**: Middle Eastern <10%.

PEX1-Related Zellweger Syndrome Spectrum - **Gene:** PEX1. **Variants (2):** 2097_2098insT, G843D. **Detection rate**: Middle Eastern 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**: Middle Eastern 43%.

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

Pompe Disease - **Gene:** GAA. **Variants (4):** D645E, R854X, IVS1-13T>G, 525delT. **Detection rate**: Middle Eastern <10%.

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

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

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

Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. **Variants (2):** 103delG, c.403_405+2delAAGT. **Detection rate**: Middle Eastern <10%.

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

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

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

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

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

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

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

Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. **Variant (1):** P315S. **Detection rate**: Middle Eastern <10%.

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**: Middle Eastern 69%.

Spinal Muscular Atrophy (copy number analysis only) - **Gene:** SMN1. **Variant (1):** SMN1 copy number. **Detection rate**: Middle Eastern 92%.

Steroid-Resistant Nephrotic Syndrome - **Gene:** NPHS2. **Variants (2):** R138Q, R138X. **Detection rate**: Middle Eastern 55%.

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

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

Tyrosinemia Type I - **Gene:** FAH. **Variants (6):** IVS12+5G>A, Q64H, P261L, W262X, E357X, IVS6-1G>T. **Detection rate**: Middle Eastern <10%.

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

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

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

Wilson Disease - **Gene:** ATP7B. **Variants (2):** H1069Q, R778L. **Detection rate**: Middle Eastern <10%.

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

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 3243 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 620	1 in 79,000
Alpha-Mannosidosis	1 in 350	1 in 500,000
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 930	1 in 590,000
Ataxia-Telangiectasia	1 in 240	1 in 150,000
Autosomal Recessive Polycystic Kidney Disease	1 in 65	1 in 16,000
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 160	1 in 100,000
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 9,000	1 in 340,000
Cystic Fibrosis	1 in 190	1 in 66,000
Cystinosis	1 in 220	1 in 200,000
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 350	1 in 22,000
Fanconi Anemia Type C	1 in 160	1 in 100,000
Galactosemia	< 1 in 500	< 1 in 1,000,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	1 in 290	1 in 110,000
Glutaric Acidemia Type 1	1 in 110	1 in 46,000
Glycogen Storage Disease Type Ia	1 in 250	1 in 180,000
Glycogen Storage Disease Type Ib	1 in 350	1 in 500,000
Glycogen Storage Disease Type III	1 in 290	1 in 180,000
Glycogen Storage Disease Type V	1 in 160	1 in 100,000
GRACILE Syndrome	< 1 in 500	< 1 in 1,000,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 84	1 in 7,900
Hereditary Fructose Intolerance	< 1 in 500	< 1 in 1,000,000
Hereditary Thymine-Uraciluria	1 in 100	1 in 40,000
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 3243 DOB: 01/01/1901	Not tested

Disease	DONOR 3243 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 160	1 in 100,000
Inclusion Body Myopathy 2	1 in 1,500	1 in 87,000
Isovaleric Acidemia	1 in 470	1 in 470,000
Joubert Syndrome 2	< 1 in 500	< 1 in 1,000,000
Krabbe Disease	1 in 150	1 in 89,000
Limb-Girdle Muscular Dystrophy Type 2D	1 in 450	1 in 800,000
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 150	1 in 90,000
Maple Syrup Urine Disease Type 1B	1 in 250	1 in 250,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 110	1 in 50,000
Megalencephalic Leukoencephalopathy With Subcortical Cysts	1 in 46	1 in 7,200
Metachromatic Leukodystrophy	1 in 200	1 in 160,000
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 250	1 in 250,000
Nijmegen Breakage Syndrome	1 in 160	1 in 100,000
Northern Epilepsy	< 1 in 500	< 1 in 1,000,000
Pendred Syndrome	1 in 71	1 in 20,000
PEX1-Related Zellweger Syndrome Spectrum	1 in 350	1 in 160,000
Phenylalanine Hydroxylase Deficiency	1 in 45	1 in 4,500
Polyglandular Autoimmune Syndrome Type 1	1 in 140	1 in 28,000
Pompe Disease	1 in 170	1 in 110,000
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 54	1 in 2,000
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 250	1 in 250,000
Smith-Lemli-Opitz Syndrome	< 1 in 500	< 1 in 1,000,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 560	1 in 110,000
Steroid-Resistant Nephrotic Syndrome	1 in 890	< 1 in 1,000,000
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 170	1 in 120,000
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 87	1 in 30,000
X-Linked Juvenile Retinoschisis	< 1 in 500	1 in 50,000