Donor 4279

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 04/07/2026

Donor Reported Ancestry: German, Irish, English, Scottish Jewish Ancestry: Yes

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Carrier screening for 17 diseases by genotyping	Negative for 180 variants associated with 17 diseases	See attached report for a list of the included genes, the variant(s) tested for each gene, and the residual risks
Tay-Sachs Enzyme Analysis	Non Carrier	

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

Counsyl

Results Recipient	Male	Female
Fairfax Cryobank [redacted] Attn: [redacted]	Name: DONOR 4279 DOB: [redacted] Ethnicity: Ashkenazi Jewish Sample Type: [redacted] Saliva Date of Collection: 09/25/2012 Date Received: 09/27/2012 Barcode: [redacted] Indication: Egg or Sperm Donor	Not tested

Report Date: 10/02/2012

Counsyl Test Results (Egg or Sperm Donor)

The Counsyl test (**Fairfax Cryobank Jewish Panel**) uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for **180 variants** associated with **17 diseases**. This report indicates which mutations, if any, were detected for each mutation panel. Because only select mutations are tested, the percentage of carriers detected varies by ethnicity. A full list of mutations tested is given on page 2. A negative test result does not eliminate the possibility that the individual is a carrier. Interpretation is given as an estimate of the risk of conceiving a child affected with a disease, which is based on reported ethnicity, the test results, and an assumption of no family history.*

DONOR 4279

DONOR 4279'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:

- 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 testing and genetic counseling.
- Individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and may also benefit from carrier testing by CBC and hemoglobin electrophoresis or HPLC. *ACOG Practice Bulletin No. 78. Obstet Gynecol 2007;109:229-37.*
- Additional Tay-Sachs disease testing can be performed using a biochemical assay, which has an excellent detection rate regardless of ancestry. *Gross et al. Genet Med 2008:10(1):54-56.*
- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complementary appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

[redacted]

Lab Directors:

William Seltzer, PhD, FACMG

Hyunseok Kang

H. Peter Kang, MD

*Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately. Other possible sources of diagnostic error include sample mix-up, trace contamination, and technical errors. The reproductive risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation. For the purposes of risk calculations, it is assumed that mutations within the same gene are on different chromosomes.

This test was developed and its performance characteristics determined by Counsyl, Inc. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician's workup. CLIA Number: **#05D1102604**.

Counsyl

Male	Female
Name: DONOR 4279 DOB: [redacted]	Not tested

Risk Calculations

Below are the full test results for all diseases on the panel. Listed in this section is the patient's post-test risk of being a carrier of each disease as well as the odds that his future children could inherit each disease. A negative result does not rule out the possibility of being a carrier of untested mutations. Estimates of post-test carrier risk assume a negative family history.

Disease	DONOR 4279 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
ABCC8-Related Hyperinsulinism	1 in 670	1 in 180,000	1 in 18,000
Bloom Syndrome	1 in 11,000	< 1 in 1,000,000	1 in 48,000
Canavan Disease	1 in 2,700	1 in 590,000	1 in 12,000
Cystic Fibrosis	1 in 900	1 in 98,000	1 in 3,000
Familial Dysautonomia	1 in 6,100	1 in 750,000	1 in 3,800
Fanconi Anemia Type C	1 in 8,900	< 1 in 1,000,000	1 in 32,000
Gaucher Disease	1 in 310	1 in 19,000	1 in 1,000
Glycogen Storage Disease Type Ia	1 in 7,000	< 1 in 1,000,000	1 in 20,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 930	1 in 590,000	1 in 100,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 370	1 in 44,000	1 in 3,600
Lipoamide Dehydrogenase Deficiency	1 in 93,000	< 1 in 1,000,000	1 in 35,000
Maple Syrup Urine Disease Type 1B	1 in 9,600	< 1 in 1,000,000	1 in 37,000
Mucolipidosis IV	1 in 2,700	< 1 in 1,000,000	1 in 40,000
Niemann-Pick Disease, SMPD1-Associated	1 in 3,300	< 1 in 1,000,000	1 in 40,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 370	1 in 60,000	1 in 6,600
Usher Syndrome Type 1F	1 in 400	1 in 160,000	1 in 40,000
Usher Syndrome Type 3	1 in 6,000	< 1 in 1,000,000	1 in 57,000

[redacted]

Counsyl

Male	Female
Name: DONOR 4279 DOB: [redacted]	Not tested

Mutations Tested

ABCC8-Related Hyperinsulinism - **Gene:** ABCC8. **Variants (3):** F1388del, V187D, 3992-9G>A. **Detection rate:** Ashkenazi Jewish 90%.

Bloom Syndrome - **Gene:** BLM. **Variants (1):** 2281del6ins7. **Detection rate:** Ashkenazi Jewish 99%.

Canavan Disease - **Gene:** ASPA. **Variants (4):** E285A, Y231X, A305E, IVS2-2A>G. **Detection rate:** Ashkenazi Jewish 98%.

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:** Ashkenazi Jewish 97%.

Familial Dysautonomia - **Gene:** IKBKAP. **Variants (2):** IVS20+6T>C, R696P. **Detection rate:** Ashkenazi Jewish >99%.

Fanconi Anemia Type C - **Gene:** FANCC. **Variants (3):** IVS4+4A>T, 322delG, R548X. **Detection rate:** Ashkenazi Jewish 99%.

Gaucher Disease - **Gene:** GBA. **Variants (10):** N370S, L444P, 84GG, IVS2+1G>A, V394L, R496H, D409H, D409V, R463C, R463H. **Detection rate:** Ashkenazi Jewish 95%.

Glycogen Storage Disease Type Ia - **Gene:** G6PC. **Variants (7):** R83C, Q347X, Q27fsdelC, 459insTA, R83H, G188R, Q242X. **Detection rate:** Ashkenazi Jewish 99%.

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:** Ashkenazi Jewish 83%.

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:** Ashkenazi Jewish 92%.

Lipoamide Dehydrogenase Deficiency - **Gene:** DLD. **Variants (2):** 105insA, G229C. **Detection rate:** Ashkenazi Jewish >99%.

Maple Syrup Urine Disease Type 1B - **Gene:** BCKDHB. **Variants (3):** R183P, G278S, E372X. **Detection rate:** Ashkenazi Jewish 99%.

Mucolipidosis IV - **Gene:** MCOLN1. **Variants (2):** 511_6944del, IVS3-2A>G. **Detection rate:** Ashkenazi Jewish 96%.

Niemann-Pick Disease, SMPD1-Associated - **Gene:** SMPD1. **Variants (4):** fsP330, L302P, R496L, p.R608del. **Detection rate:** Ashkenazi Jewish 97%.

Spinal Muscular Atrophy - **Gene:** SMN1. **Variants (1):** SMN1 copy number. **Detection rate:** Ashkenazi Jewish 91%.

Usher Syndrome Type 1F - **Gene:** PCDH15. **Variants (1):** R245X. **Detection rate:** Ashkenazi Jewish 75%.

Usher Syndrome Type 3 - **Gene:** CLRN1. **Variants (1):** N48K. **Detection rate:** Ashkenazi Jewish 98%.

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Integrated GENETICS
LabCorp Specialty Testing Group

Tay-Sachs Enzyme Analysis

Patient Name: Donor 4279, .
Referring Physician:
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

DOB: [redacted]
SSN: ***-**-****

Date Collected: 09/25/2012
Date Received: 09/26/2012
Lab ID:
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 1340 nmol/mg protein**
Hexosaminidase Percent A: 61.2

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥54%	≥54%
Expected Carrier Range:	Hex A	20 - 49%	20 - 49%

INTERPRETATION: NON CARRIER

This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

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Under the direction of: Stanford Marenberg, PHD, MBCC

Stanford Marenberg, Ph.D.

Date: 09/29/2012

Testing Performed At Esoterix Genetic Laboratories, LLC 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436