



Donor 4488

Genetic Testing Summary

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

Last Updated: 01/04/23

Donor Reported Ancestry: Irish

Jewish Ancestry: No

| 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 |
| Cystic Fibrosis (CF) carrier screening | Negative by for 99 mutations in the CFTR gene | 1/300 |
| Spinal Muscular Atrophy (SMA) carrier screening | Negative for deletions of exon 7 and gene sequencing in the SMN1 gene | 1/4800 |
| HBB gene testing (Sickle Cell Disease and Beta Thalassemia) | Negative for 28 mutations in the HBB gene | 1/290 |
| Tay Sachs Enzyme Analysis | Non-carrier by hexosaminidase A analysis | |

*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.

**Results Recipient**Fairfax Cryobank - [REDACTED]
[REDACTED]
[REDACTED]
Report Date: 02/21/2013**Male**Name: 4488 4488
DOB: [REDACTED]
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 02/18/2013
Date Received: 02/20/2013
Barcode: [REDACTED]
Indication: No family history
(screening)**Female**

Not tested

Counsyl Test Results

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for **128 variants** associated with **3 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.*

**4488 4488**

4488 4488'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.*
- 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/.

Lab Directors:

William Seltzer, PhD, FACMG

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.



Male

Name: 4488 4488

DOB: [REDACTED]

Female

Not tested

Mutations Tested

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%.

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%.

Spinal Muscular Atrophy - Gene: SMN1. Variants (1): SMN1 copy number. Detection rate: Northern European 95%.

**Male**

Name: 4488 4488

DOB: [REDACTED]

Female

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 | 4488 4488 Residual Risk | Post-test Reproductive Risk | Pre-test Reproductive Risk |
|---------------------------------------------------------------------------------------------|-------------------------------|--------------------------------|-------------------------------|
| Cystic Fibrosis | 1 in 300 | 1 in 33,000 | 1 in 3,000 |
| Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) | 1 in 290 | 1 in 58,000 | 1 in 10,000 |
| Spinal Muscular Atrophy | SMN1: 3+ copies 1 in 4,800 | 1 in 670,000 | 1 in 4,800 |

Tay-Sachs Enzyme Analysis

Patient Name: 4488, [REDACTED]
Referring Physician: [REDACTED]
Specimen #: [REDACTED]
Patient ID: [REDACTED]

Client #: [REDACTED]

Fairfax Cryobank [REDACTED]

DOB: Not Given
SSN: ***-**-****

Date Collected: 02/18/2013
Date Received: 02/19/2013
Lab ID: 4488-130218
Hospital ID:
Specimen Type: White Blood Cells

RESULTS: Hexosaminidase Activity : 825 nmol/mg protein
Hexosaminidase Percent A: 64.3

| | Hex A | 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.

Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

Under the direction of:



Stanford Marenberg, PhD, MSc
Stanford Marenberg, Ph.D.

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

ENTERED
NP 03-06-13

Date: 02/22/2013

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