



## Donor 4542

### Genetic Testing Summary

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

Last Updated: 06/24/22

Donor Reported Ancestry: Italian, Sicilian, English, 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 for 99 mutations in the CFTR gene	1/310
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 and gene sequencing in the SMN1 gene	1/700
Beta Thalassemia (HBB) Sickle Cell Disease (HBB)	Negative for 28 mutations in the HBB gene	1/500- Beta Thalassemia <1/500 – Sickle Cell Disease.

\*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



Report Date: 12/26/2011

Male

Name: DONOR 4542  
DOB: [REDACTED]  
Ethnicity: Northern European  
Sample Type: OG-500 Saliva  
Date of Collection: 12/16/2011  
Barcode: [REDACTED]  
Indication: Egg or Sperm Donor

Female

Not tested

## Counsyl Test Results (Egg or Sperm Donor)

Panel: Fairfax Cryobank Fundamental Panel

The Counsyl test uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for a number of Mendelian 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 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 4542*

*DONOR 4542'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:

- 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.*

To schedule a free appointment to speak with a genetic counselor about your results, please visit [www.counsyl.com/appointment](http://www.counsyl.com/appointment).

\*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, 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.

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. Lab Directors: Jessica Jacobson, MD, William K. Seltzer, PhD, FACMG.



Full Results

Below are the full test results for all diseases on the panel. Noted are the specific genetic mutations for which the patient tested positive or negative. If there was insufficient data to determine the genotype for any variant, this will be noted as "no call." Also 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.

✓ **Beta Thalassemia**      **Reproductive risk:** Less than 1 in 1,000,000      **Risk before testing:** 1 in 250,000

DONOR 4542: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 1,500. 83% detection rate.

Gene: HBB. Variants (27): 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.

✓ **Cystic Fibrosis**      **Reproductive risk:** 1 in 34,000      **Risk before testing:** 1 in 3,000

DONOR 4542: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 310. 91% detection rate.

Gene: CFTR. Variants (99): G85E, R117H, R334W, R347P, A155E, 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, 1949del8-1, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667delH, 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.

✓ **Sickle Cell Disease**      **Reproductive risk:** Less than 1 in 1,000,000      **Risk before testing:** less than 1 in 1,000,000

DONOR 4542: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 70% detection rate.

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.

✓ **Spinal Muscular Atrophy**      **Reproductive risk:** 1 in 97,000      **Risk before testing:** 1 in 4,800

DONOR 4542: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 700. 95% detection rate.

Gene: SMN1. Variants (1): Exon 7 deletion.

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