



## CLI Donor 4554

### Genetic Testing Summary

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

Last Updated: 8/14/2025

Donor Reported Ancestry: Hmong

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobinopathy 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 genotyping	1 in 190
Spinal Muscular Atrophy (SMA) carrier screening	Negative	1 in 720
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative by genotyping	1 in 130 and <1 in 500, respectively

\*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.

## Cytogenetic Report

Client Cryogenic Laboratories

Address

Reporting Phone #

Email N/A

Patient name/Donor Alias Donor # 4554

Patient DOB N/A

Donor #

Specimen type Peripheral Blood

Collection Date 05/24/2012

Accession #

Date Received 05/25/2012

### RESULTS

#### CYTOGENETIC ANALYSIS

#### FISH

Cells counted 20

Type of banding GTG

Probe(s) N/A

Cells analyzed 5

Band resolution 550

Nuclei scored N/A

Cells karyotyped 2

Modal chromosome # 46


KARYOTYPE 46,XY

#### INTERPRETATION

Normal male karyotype

No clonal numerical or structural abnormalities were identified. This normal cytogenetic result does not exclude the possibility of the presence of subtle rearrangements beyond the technical limits of detection with this test.

Comments

  
Wayne S. Stanley, Ph.D., FACMG  
Clinical Cytogeneticist

6/7/12  
Date

**Results Recipient**

Cryogenic Laboratories

Attn: [REDACTED]  
[REDACTED]  
[REDACTED]  
Report Date: 05/22/2012**Male**

Name: DONOR 4554

DOB: [REDACTED]

Ethnicity: Southeast Asian

Sample Type: [REDACTED]

Date of Collection: 05/16/2012

Date Received: 05/18/2012

Barcode: [REDACTED]

Indication: Egg or Sperm Donor

**Female**

Not tested

**Counsyl Test Results (Egg or Sperm Donor)**

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 **4 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 4554**

DONOR 4554'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.*
- Genetic counseling is recommended. 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).

**Lab Directors:**

Jessica Jacobson, MD

William Seltzer, PhD, FACMG

\* **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.

## 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 4554 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
✓ Beta Thalassemia	1 in 130	1 in 9,500	1 in 1,300
✓ Cystic Fibrosis	1 in 190	1 in 66,000	1 in 30,000
✓ Sickle Cell Disease	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
✓ Spinal Muscular Atrophy	1 in 720	1 in 150,000	1 in 11,000

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Patient Information	Specimen Information	Client Information
<b>ID4554, DONOR</b>  <b>DOB:</b> [REDACTED] <b>Gender:</b> M <b>Fasting:</b> U <b>Phone:</b> NG <b>Patient ID:</b> [REDACTED]	<b>Specimen:</b> [REDACTED] <b>Requisition:</b> [REDACTED] <b>Lab Ref #:</b> [REDACTED]  <b>Collected:</b> 05/16/2012 / 10:15 CDT <b>Received:</b> 05/17/2012 / 03:44 CDT <b>Reported:</b> 05/18/2012 / 13:37 CDT	[REDACTED]  [REDACTED]  [REDACTED]

Test Name	In Range	Out Of Range	Reference Range	Lab
✓ HEMOGLOBINOPATHY EVALUATION ✓				
RED BLOOD CELL COUNT	4.81		4.20-5.80 Million/uL	CB
HEMOGLOBIN	14.8		13.2-17.1 g/dL	
HEMATOCRIT	44.2		38.5-50.0 %	
MCV	92.0		80.0-100.0 fL	
MCH	30.8		27.0-33.0 pg	
RDW	13.8		11.0-15.0 %	
HEMOGLOBIN A	97.5		>96.0 %	CB
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.5		1.8-3.5 %	
INTERPRETATION				

Normal phenotype.

Normal hemoglobin distribution, no HgS, HgC or other abnormal hemoglobin observed.

✓ CHOLESTEROL, TOTAL ✓	173		125-200 mg/dL	CB
✓ AST ✓	35		10-40 U/L	CB
✓ ALT ✓	37		9-60 U/L	CB
✓ CBC (INCLUDES DIFF/PLT) ✓				CB
WHITE BLOOD CELL COUNT	8.9		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.81		4.20-5.80 Million/uL	
HEMOGLOBIN	14.8		13.2-17.1 g/dL	
HEMATOCRIT	44.2		38.5-50.0 %	
MCV	92.0		80.0-100.0 fL	
MCH	30.8		27.0-33.0 pg	
MCHC	33.5		32.0-36.0 g/dL	
RDW	13.8		11.0-15.0 %	
PLATELET COUNT	164		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	4762		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	2910		850-3900 cells/uL	
ABSOLUTE MONOCYTES	534		200-950 cells/uL	
ABSOLUTE EOSINOPHILS			15-500 cells/uL	
ABSOLUTE BASOPHILS	36		0-200 cells/uL	
NEUTROPHILS	53.5		%	
LYMPHOCYTES	32.7		%	
MONOCYTES	6.0		%	
EOSINOPHILS	7.4		%	
BASOPHILS	0.4		%	
✓ ABO GROUP AND RH TYPE ✓				CB
ABO GROUP	O			
RH TYPE	RH (D) POSITIVE			

PERFORMING SITE:

CB QUEST DIAGNOSTICS WOOD DALE, 1355 MITTEL BOULEVARD, WOOD DALE, IL 60191-1024 Laboratory Director: ANTHONY V. THOMAS, MD, CLIA: 14D0417052