



Donor 2691

Genetic Testing Summary

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

Last Updated: 08/28/24

Donor Reported Ancestry: German, English, French, Finnish

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 genotyping of 99 mutations in the CFTR gene	1/343

*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.

Patient Name: Donor, 2691

Referring Physician: [REDACTED]

Specimen #: [REDACTED]

Patient ID: [REDACTED]

Client #: [REDACTED]

Case #: [REDACTED]

DOB: Not Given

Sex: M

SSN: [REDACTED]

Date Collected: 10/29/2007

Date Received: 10/31/2007

Lab ID: 2691-071029

Hospital ID: [REDACTED]

Specimen Type: BLDPER

Ethnicity: Caucasian

Indication: Carrier test / Gamete donor

RESULTS: Negative for the 97 mutations analyzed**INTERPRETATION**

This individual's risk to be a carrier is reduced from 1/25 (4%) to 1/343 (0.3%), based on these results and a negative family history.

COMMENTS:

Mutation Detection Rates among Ethnic Groups		Detection rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild presentation (e.g. congenital absence of the vas deferens, pancreatitis) detection rates may vary from those provided here.	
Ethnicity	Carrier risk reduction when no family history	Detection rate	References
African American	1/65 to 1/338	81%	Genet in Med 3:168, 2001
Ashkenazi Jewish	1/26 to 1/834	97%	Am J Hum Genet 51:951, 1994
Asian		Not Provided	Insufficient data
Caucasian	1/25 to 1/343	93%	Genet in Med 3:168, 2001; Genet in Med 4:90, 2002
Hispanic	1/46 to 1/205	78%	Genet in Med 3:168, 2001; www.dhs.ca.gov/pcfh/gdb/html/PDE/CFStudy.htm
Jewish, non-Ashkenazi		Varies by country of origin	Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997
Other or Mixed Ethnicity		Not Provided	Detection rate not determined and varies with ethnicity

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.

METHOD

DNA is isolated from the sample and tested for the 97 CF mutations listed. Regions of the *CFTR* gene are amplified enzymatically and subjected to a solution-phase multiplex allele-specific primer extension with subsequent hybridization to a bead array and fluorescent detection. The assay discriminates between $\Delta F508$ and the following polymorphisms: F508C, I506V and I507V. In some cases, specific allele identification requires enzymatic amplification followed by hybridization to oligonucleotide probes.

Under the direction of:



Lynne Rosenblum-Vos
Lynne Rosenblum-Vos, Ph. D.

Date: 11/07/2007

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Patient Name: Donor, 2691

Referring Physician: [REDACTED]

Specimen #: [REDACTED]

Client #: [REDACTED]

Patient ID: [REDACTED]

DOB: Not Given

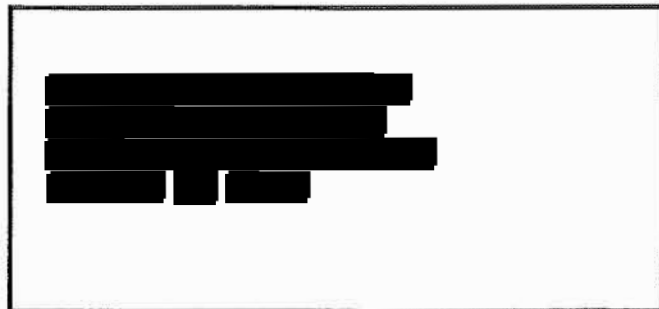
Date Collected: 11/15/2007

SSN:

Date Received: 11/16/2007

Lab ID: 2691-071115

Hospital ID:

Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted: 20

Metaphases Analyzed: 5

Number of Cultures: 2

Metaphases Karyotyped: 2

Banding Technique: GTW

Banding Resolution: 550

Dept. Section: B1

RESULTS: 46,XY**Male karyotype****INTERPRETATION:**

This analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. The standard cytogenetic methodology utilized in this analysis does not routinely detect small rearrangements and low level mosaicism, and cannot detect microdeletions.

Signed:

Date: 11/29/2007



David M. De Bauche, Ph.D.

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