



## Donor 1438

### Genetic Testing Summary

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

Last Updated: 06/23/20

Donor Reported Ancestry: German

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Cystic Fibrosis (CF) carrier screening	Negative for 87 mutations testing in the CFTR gene	1/325

\*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: CLI 1438, [REDACTED]  
Referring Physician:  
Specimen #: [REDACTED]  
Patient ID: [REDACTED]

Client #: [REDACTED]  
Case #: [REDACTED]

DOB: Not Given  
Sex: M  
SSN:

Date Collected: 11/26/2002  
Date Received: 05/15/2003  
Lab ID:  
Hospital ID:  
Specimen Type: [REDACTED]

Fairfax Cryobank  
[REDACTED]

Ethnicity: Not Provided

Indication: Carrier test / Gamete donor

**RESULTS: Negative for the mutations analyzed**

**INTERPRETATION**

The sample provided is negative for the mutations analyzed.

**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	CF87 Detection rate	References
Caucasian	1/25 to 1/325	92.6%	Genet in Med 3:168, 2001 in conjunction with Genet in Med 4:90, 2002
African American	1/65 to 1/338	81%	Genet in Med 3:168, 2001
Hispanic	1/46 to 1/162	72%	Genet in Med 3:168, 2001
Ashkenazi Jewish	1/26 to 1/834	97%	Am J Hum Genet 51:951, 1994
Jewish, non-Ashkenazi		Varies by country of origin	Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997
Asian		Not Provided	Insufficient data
Other or Mixed Ethnicity		Not Provided	Detection rate not determined and varies with ethnicity

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of this condition. Although DNA-based testing is highly accurate, rare diagnostic errors may occur. Examples include misinterpretation because of genetic variants, blood transfusion, bone marrow transplantation, or erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

**METHOD**

DNA is isolated from the sample and tested for the 87 CF mutations listed. Regions of the CFTR gene are amplified enzymatically and hybridized to specific CF mutation oligonucleotide probes. Results are characterized as positive or negative, and specimens with positive results are tested for specific mutation identity. The assay discriminates between  $\Delta F508$  and the following polymorphisms: F508C, I506V, I506M and I507V.

This test was developed and its performance characteristics determined by Genzyme Genetics. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.

Under the direction of:



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Date: 05/23/2003

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