



Donor 2669

Genetic Testing Summary

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

Last Updated: 04/22/21

Donor Reported Ancestry: Chinese

Jewish Ancestry: No

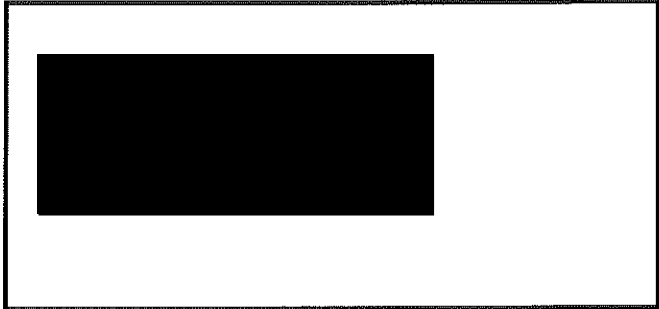
Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative for 97 mutations in the CFTR gene	See attached result

*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, 2669
 Referring Physician: Steve Pool, M.D.
 Specimen # [REDACTED]
 Patient ID: [REDACTED]

Client # [REDACTED]
 Case #: [REDACTED]



DOB: Not Given
 Sex: M
 SSN: [REDACTED]

Date Collected: 04/26/2007
 Date Received: 04/28/2007
 Lab ID: [REDACTED]
 Hospital ID:
 Specimen Type: BLDPER

Ethnicity: Asian
 Indication: Not Provided

RESULTS: Negative for the 97 mutations analyzed

INTERPRETATION
 This negative result may need further interpretation depending on the clinical indication.

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	Detection rate	References
African American	81%	Genet in Med 3:168, 2001
Ashkenazi Jewish	97%	Am J Hum Genet 51:951, 1994
Asian	Not Provided	Insufficient data
Caucasian	93%	Genet in Med 3:168, 2001; Genet in Med 4:90, 2002
Hispanic	78%	Genet in Med 3:168, 2001; www.dhs.ca.gov/pcftv/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:

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Date: 05/09/2007



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