# 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.

genzyme

# Cystic Fibrosis Mutation Analysis

**Patient Name:** Donor, 2669
**Referring Physician:** Steve Pool, M.D.
**Specimen #:** [redacted]
**Patient ID:** [redacted]
**Client #:** [redacted]
**Case #:** [redacted]

[redacted]

DOB: Not Given
Sex: M
SSN:

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/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 ΔF508 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: 05/09/2007



Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357