Donor 2701

Genetic Testing Summary

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

Last Updated: 06/25/24

Donor Reported Ancestry: Irish, Austrian, 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
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 97 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.

genzyme

Cystic Fibrosis Mutation Analysis

Patient Name: Donor 2701, .
Referring Physician: [redacted] D.
Specimen #: [redacted]
Patient ID: [redacted]
Client # [redacted]
Case #: [redacted]

Fairfax Cryobank
[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 11/17/2005
Date Received: 11/21/2005
Lab ID: [redacted]
Hospital ID:
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/CFTable1.html
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 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 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: 12/01/2005

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

genzyme

Chromosome Analysis

Patient Name: Adult, 2701
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

Fairfax Cryobank
[redacted]

DOB: Not Given
SSN:

Date Collected: 01/10/2006
Date Received: 01/12/2006
Lab ID: [redacted]
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	6	**Number of Cultures:**	2	**Banding Resolution:**	550
Metaphases Karyotyped:	2			**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: Liang Dong

Liang Dong, FACMG

Date: 01/19/2006

Testing Performed At Genzyme Genetics 655 East Huntington Drive Monrovia, CA 91016 E.Robert Wassman M.D., Laboratory Director 800-255-1616

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 410.247.9100

PATIENT INFORMATION
2701

DOB:
GENDER: M
Age: [redacted]

REPORT STATUS **Final**

ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
[redacted]
FAIRFAX CRYOBANK
[redacted]

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION: [redacted]
LAB REF NO:

COLLECTED: 11/17/2005 09:00
RECEIVED: 11/18/2005 22:16
REPORTED: 11/22/2005 11:45

COMMENTS: 2701-051117

Test Name	In Range	Out of Range	Reference Range	Lab
[redacted]	[redacted]		[redacted]	[redacted]
[redacted]	[redacted]		[redacted]	[redacted]
[redacted]	[redacted]		[redacted]	[redacted]
CBC W/DIFF & PLT				QBA
WHITE BLOOD CELL COUNT	4.1		3.8-10.8 THOUS/MCL	
RED BLOOD CELL COUNT	4.78		4.20-5.80 MILL/MCL	
HEMOGLOBIN	15.0		13.2-17.1 G/DL	
HEMATOCRIT	44.0		38.5-50.0 %	
MCV	92		80-100 FL	
MCH	31.3		27-33 PG	
MCHC	34.0		32-36 G/DL	
PLATELET COUNT	220		140-400 THOUS/MCL	
RDW	13.3		11.0-15.0 %	
MPV	8.5		7.5-11.5 FL	
ABSOLUTE NEUTROPHILS	2050		1500-7800 CELLS/MCL	
ABSOLUTE LYMPHOCYTES	1652		850-3900 CELLS/MCL	
ABSOLUTE MONOCYTES	283		200-950 CELLS/MCL	
ABSOLUTE EOSINOPHILS	94		15-550 CELLS/MCL	
ABSOLUTE BASOPHILS	21		0-200 CELLS/MCL	
NEUTROPHILS	50.0		%	
LYMPHOCYTES	40.3		%	
REACTIVE LYMPHOCYTES	0.0		%	
MONOCYTES	6.9		%	
EOSINOPHILS	2.3		%	
BASOPHILS	0.5		%	
COMMENT	Platelets appear adequate.			
BLOOD GROUP & RH				QBA
BLOOD GROUP	A			
RH TYPE	POSITIVE			
HEMOGLOBINOPATHY EVALUATION				QBA
RED BLOOD CELL COUNT	4.78		4.20-5.80 MILL/MCL	
HEMOGLOBIN	15.0		13.2-17.1 G/DL	
HEMATOCRIT	44.0		38.5-50.0 %	
MCV	92		80-100 FL	

DATA ENTERED

QUEST DIAGNOSTICS INCORPORATED

PATIENT INFORMATION
2701

DOB:
GENDER: M
Age: [redacted]

REPORT STATUS **Final**

ORDERING PHYSICIAN [redacted]

REPORTED: 11/22/2005 11:45

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION	(Continued)			
MCH	31.3		27-33 PG	
	Platelets appear adequate.			
RDW	13.3		11.0-15.0 %	
HEMOGLOBIN A1	97.8		>96.0 %	
HEMOGLOBIN F	NONE DETECTED		0.0-1.9	
HEMOGLOBIN A2	2.2		1.8-3.5 %	
HGB SCREEN INTERPRETATION				
	THE HEMOGLOBINOPATHY SCREEN IS NORMAL.			
ABNORMAL HEMOGLOBIN #1 %:	0.0		%	

This report was electronically signed by: Robert R. L. Smith, M.D.
Quest Diagnostics, Inc. - Baltimore
1901 Sulphur Spring Road
Baltimore, MD 21227
(410)247-9100

Performing Laboratory Information:

QBA Quest Diagnostics Incorporated 1901 Sulphur Spring Road Baltimore MD 21227 Laboratory Director: Robert R. L. Smith, M.D.