



## Donor 2815

### Genetic Testing Summary

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

Last Updated: 08/27/18

Donor Reported Ancestry: German, Irish, Italian, Bengali

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	1/190

\*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 2815,  
 Referring Physician:  
 Specimen #:  
 Patient ID:

Client #:  
 Case #:

Fairfax Cryobank  
 Genetics and IVF Institute

DOB: Not Given Date Collected: 01/19/2006  
 Sex: M Date Received: 01/21/2006  
 SSN: Lab ID:  
 Hospital ID:  
 Specimen Type: **BLDPER**

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 1/31/06  
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Ethnicity: Caucasian, Pakistani  
 Indication: Carrier test / Gamete donor

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

**INTERPRETATION**

This individual is negative for the mutations analyzed. This result reduces but does not eliminate the risk to be a CF carrier.

**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/pch/gdb/html/PDE/CFTTable1.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  $\Delta F508$  and the following polymorphisms: F508C, I506V and I507V. In some cases, specific allele identification requires enzymatic amplification followed by hybridization to oligonucleotide probes.

Signed: *Stephanie Hallam*

Date: 01/31/2006



Stephanie Hallam, Ph.D.

Page 1 of 1

Patient Name: 2815, Adult  
Referring Physician: [Redacted]  
Specimen #: [Redacted] Client #: 606452  
Patient ID: [Redacted]

Fairfax Cryobank [Redacted]  
Genetics and IVF Institute  
[Redacted]

DOB: Not Given Date Collected: 02/07/2006  
SSN: Date Received: 02/09/2006  
Lab ID: 2815-060207  
Hospital ID:  
Specimen Type: **Peripheral Blood**

Indication: No family history / 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**

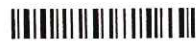
**INTERPRETATION:**

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.

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2/16/06  
MCA

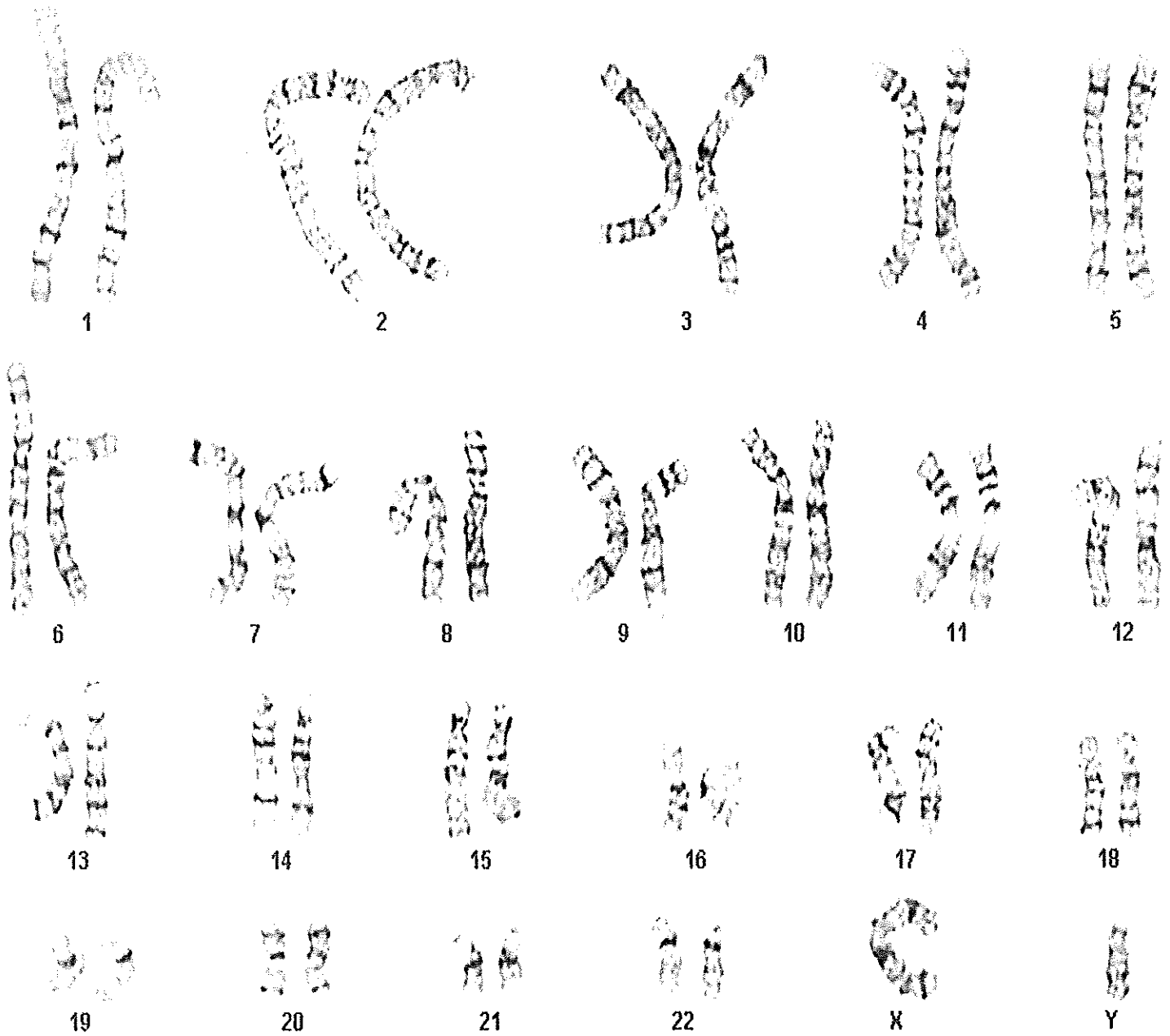
Signed: *Atieh Hajianpour*

Date: 02/15/2006



Atieh Hajianpour, FACMG

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Specimen #: [REDACTED]  
Specimen Type: Peripheral Blood  
Patient Name: 2815, Adult  
Reviewed By: AH1  
Karyotype: 46,XY

Dept ID: B1  
Date Received: 02/09/2006  
Date Reviewed: 02/15/2006





PATIENT INFORMATION  
ID, 2815

REPORT STATUS **FINAL** REPRINT

QUEST DIAGNOSTICS INCORPORATED  
CLIENT SERVICE 800.825.7330

DOB: AGE:  
GENDER: M FASTING: N  
SSN:  
ID:  
PHONE:

ORDERING PHYSICIAN

CLIENT INFORMATION  
N19104437 HO19  
FAIRFAX CRYO BANK

SPECIMEN INFORMATION

SPECIMEN:  
REQUISITION:

COLLECTED: 01/19/2006 13:10 ET  
RECEIVED: 01/19/2006 20:19 ET  
REPORTED: 01/24/2006 05:45 ET

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1/25/06  
mid

Test Name	In Range	Out of Range	Reference Range	Lab
<b>HEMOGLOBINOPATHY EVALUATION</b>				
RED BLOOD CELL COUNT	4.97		4.20-5.80 MILL/MCL	QHO
HEMOGLOBIN	15.0		13.2-17.1 G/DL	
HEMATOCRIT	44.0		38.5-50.0 %	
MCV	88.5		80.0-100.0 FL	
MCH	30.3		27.0-33.0 PG	
RDW	13.2		11.0-15.0 %	
HEMOGLOBIN A1	97.8		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.2		1.8-3.5 %	
INTERPRETATION	NORMAL PHENOTYPE.			
CHOLESTEROL, TOTAL	155		<200 MG/DL	QHO
AST	25		3-50 U/L	QHO
ALT	29		3-60 U/L	QHO
<b>CBC (INCLUDES DIFF/PLT)</b>				
WHITE BLOOD CELL COUNT	7.8		3.8-10.8 THOUS/MCL	QHO
RED BLOOD CELL COUNT	4.97		4.20-5.80 MILL/MCL	
HEMOGLOBIN	15.0		13.2-17.1 G/DL	
HEMATOCRIT	44.0		38.5-50.0 %	
MCV	88.5		80.0-100.0 FL	
MCH	30.3		27.0-33.0 PG	
MCHC	34.2		32.0-36.0 G/DL	
RDW	13.2		11.0-15.0 %	
PLATELET COUNT	248		140-400 THOUS/MCL	
ABSOLUTE NEUTROPHILS	5226		1500-7800 CELLS/MCL	
ABSOLUTE LYMPHOCYTES	2106		850-3900 CELLS/MCL	
ABSOLUTE MONOCYTES	312		200-950 CELLS/MCL	
ABSOLUTE EOSINOPHILS	156		15-500 CELLS/MCL	
ABSOLUTE BASOPHILS	0		0-200 CELLS/MCL	
NEUTROPHILS	67		%	
LYMPHOCYTES	27		%	
MONOCYTES	4		%	
EOSINOPHILS	2		%	
BASOPHILS	0		%	



PATIENT INFORMATION  
ID, 2815

REPORT STATUS **FINAL** REPRINT

QUEST DIAGNOSTICS INCORPORATED

ORDERING PHYSICIAN  
[REDACTED]

REPORTED: 01/24/2006 05:45 ET

DOB: AGE:  
GENDER: M FASTING: N

Test Name	In Range	Out of Range	Reference Range	Lab
ABO GROUP & RH TYPE				QHO
ABO GROUP	A			
RH TYPE	RH (D) POSITIVE			

**PERFORMING LABORATORY INFORMATION**

QHO QUEST DIAGNOSTICS-HORSHAM, 900 BUSINESS CENTER DRIVE, HORSHAM, PA 19044, Laboratory Director: HERMAN HURWITZ, MD, FCAP