

## Donor 2835

## **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: Chinese

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**

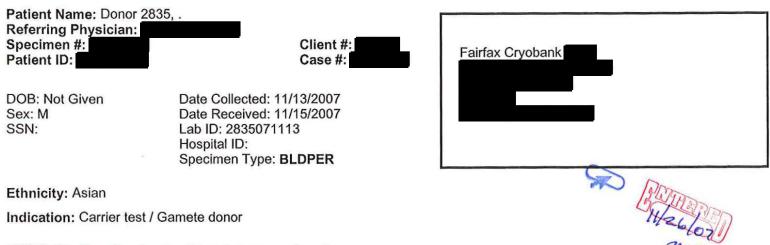
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/ and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative 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.



# Cystic Fil. Jsis Mutation Analysis



#### **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 Detection rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild   among Ethnic Groups Detection rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild				
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/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.

Signed:

Ruth & Heim, PhD, FACMG

Ruth A. Heim, Ph.D., FACMG

Date: 11/23/2007

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

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Quest Diagnostics	$C_{i}^{*}$	PATIENT INFORMATION ID,2835	SPORT STATUS FINAL	
QUEST DIAGNOSTICS INCORPORATED			ORDERING PHYSICIAN	
CLIENT SERVICE 800.825.7330		DOB: AGE:		
	. 91	GENDER: M FASTING: N		
SPECIMEN INFORMATION			CLIENT INFORMATION	
SPECIMEN:		ID: 2835.	N19104437	HO13
REQUISITION:		PHONE :	FAIRFAX CRYO BANK	11000111210404
COLLECTED: 12/06/2007	12:00 ET			
RECEIVED: 12/06/2007	21:09 ET			Dr.
REPORTED: 12/07/2007	06:50 ET		12/7/07	
Test Name		In Range Out of Rang	e Reference Range	Lab
			and and a constant and an	
CBC (INCLUDES DIFF/PLI	C)			QHO
WHITE BLOOD CELL COU	JNT	4.5	3.8-10.8 Thousand/uL	12.2
RED BLOOD CELL COUNT	C	4.57	4.20-5.80 Million/uL	
HEMOGLOBIN		14.7	13.2-17.1 g/dL	
HEMATOCRIT		41.3	38.5-50.0 %	
MCV		90.3	80.0-100.0 fL	
MCH		32.1	27.0-33.0 pg	
MCHC		35.5	32.0-36.0 g/dL	
RDW		12.4	11.0-15.0 %	
PLATELET COUNT		224	140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS		2655	1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	3	1530	850-3900 cells/uL	
ABSOLUTE MONOCYTES		225	200-950 cells/uL	
ABSOLUTE EOSINOPHILS	3	90	15-500 cells/uL	
ABSOLUTE BASOPHILS		0	0-200 cells/uL	
NEUTROPHILS		59	20	
LYMPHOCYTES		34	90	
MONOCYTES		5	00	
EOSINOPHILS		2	20	
BASOPHILS		0	00	

#### PERFORMING LABORATORY INFORMATION

QHO QUEST DIAGNOSTICS-HORSHAM, 900 BUSINESS CENTER DRIVE, HORSHAM, PA 19044, Laboratory Director: HERMAN HURWITZ, MD, FCAP CLIA: 39D0204404

LIST OF RESULTS PRINTED IN THE OUT OF RANGE COLUMN:

genzyme	$\subset$	So ALTHANK		Chromosome A	nalysis
Patient Name: Donor, 283 Referring Physician: Specimen # Patient ID: 8	35	Client #:	Ð	Fairfax Cryobank	
DOB: Not Given SSN:	Date Collecte Date Receive Lab ID: 2835 Hospital ID: Specimen Ty	<u>d: 12/07/</u> 2007			
Indication: Gamete don Metaphases Counted: Metaphases Analyzed: Metaphases Karyotype	20 5	Number of Cultures:	2	Banding Technique: Banding Resolution: Dept. Section:	GTW 500 B1
RESULTS: 46,XY Male kar	votype				

### 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: 

Suchan Berend, Ph.D., FACMO

Date: 12/17/200

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1	2	3			4	5
6	7	8	9	10	11	13
13	14	15		16	17	18
19	30		21	22	x	Y

Specimen #: Specimen Type: BLDPER Patient Name: Donor, 2835 Image ID: BKE1 Karyotype: 46,XY

Dept ID: B1 Date Received: 12/07/2007 Date Reviewed: 12/17/2007 Reviewed By: SB2

genzyme GENERAL genetics