



## Donor 2494

### Genetic Testing Summary

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

Last Updated: 10/29/24

Donor Reported Ancestry: German

Jewish Ancestry: No

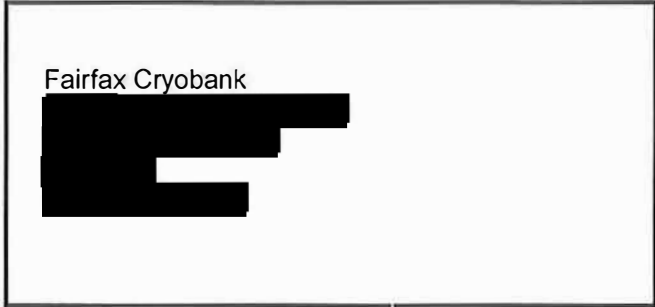
Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by genotyping of 86 mutations in the CFTR gene	1/325

\*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, <sup>FFX</sup> GLI 2494 <sup>YAU 3/20/06</sup>  
 Referring Physician: [REDACTED]  
 Specimen #: [REDACTED]  
 Patient ID: [REDACTED]

Client #: [REDACTED]  
 Case #: [REDACTED]



DOB: Not Given      Date Collected: 02/17/2005  
 Sex: M              Date Received: 02/19/2005  
 SSN:                Lab ID: [REDACTED]  
                       Hospital ID:  
                       Specimen Type: **BLDPER**

**Ethnicity:** Caucasian

**Indication:** Carrier test / Gamete donor

**RESULTS:** Negative for the 86 mutations analyzed

**INTERPRETATION**

This individual's risk to be a carrier is reduced from 1/25 (4%) to 1/325 (0.3%), based on these results, a negative family history and the absence of symptoms.

**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
Caucasian	1/25 to 1/325	92.6%	Genet in Med 3:168, 2001 in conjunction with Genet in Med 4:90, 2002
African American	1/65 to 1/338	81%	Genet in Med 3:168, 2001
Hispanic	1/46 to 1/162	72%	Genet in Med 3:168, 2001
Ashkenazi Jewish	1/26 to 1/834	97%	Am J Hum Genet 51:951, 1994
Jewish, non-Ashkenazi		Varies by country of origin	Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997
Asian		Not Provided	Insufficient data
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 86 CF mutations listed. Regions of the CFTR gene are amplified enzymatically and hybridized to specific CF mutation oligonucleotide probes. Results are characterized as positive or negative, and specimens with positive results are tested for specific mutation identity using either the same methodology or 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, I506M and I507V.

Under the direction of:

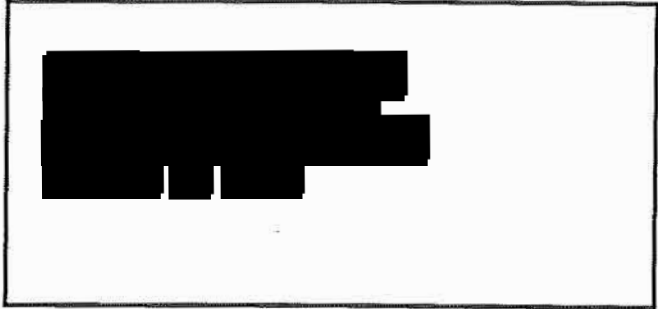
Date: 02/25/2005



Ruth A. Heim, Ph. D.

Page 1 of 1

*FFX of 3-21-06*  
Patient Name: Donor, *OLI 2494 Donor is Fairfax Brand*  
Referring Ph: *Steve Pool, M.D. 3-21-06 SF*  
Specimen #: [Redacted] Client #: [Redacted]  
Patient ID: [Redacted] 1



DOB: Not Given Date Collected: 03/14/2005  
SSN: [Redacted] Date Re: [Redacted] 5/2005  
Lab ID: [Redacted]  
Hospital ID: [Redacted]  
Specimen Type: Peripheral Blood

Indication: Gamete donor

Metaphases Counted: 20	Banding Technique: GTW	
Metaphases Analyzed: 5	Number of Cultures: 2	Banding Resolution: 500
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.

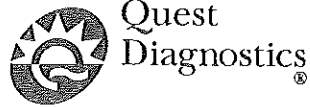
Signed:

*Jay W. Moore*  
Jay W. Moore, Ph.D. FFACMG

Date: 03/24/2005



Page 1 of 1



PATIENT NAME D, DONOR#2494	PATIENT ID ADULT	ROOM NO.	AGE	SEX	PHYSICIAN
LAB REF. #	COLLECTION DATE & TIME 02/17/2005 1300	LOG IN DATE 02/17/2005	REPORT DATE 02/21/2005	TIME 7:50AM	

REMARKS  
REGS# 2494

CENTRAL  
TIME  
FASTING: U

REPORT STATUS	TEST	RESULT	UNITS	REFERENCE RANGE	SITE CODE
		IN RANGE	OUT OF RANGE		

ate of Birth: NG					
HEMOGLOBINOPATHY EVALUATION					
RED BLOOD CELL COUNT	4.78			MILL/MCL	4.20-5.10 CB
HEMOGLOBIN		15.6 H		G/DL	13.2-15.5
HEMATOCRIT	44.3			%	38.5-45.0
MCV	92.9			FL	80.0-100.0
MCH	32.6			PG	27.0-33.0
RDW	12.7			%	11.0-15.0
HEMOGLOBIN A1	97.5			%	>96.0 CB
FETAL HEMOGLOBIN	<1.0			%	<2.0
HEMOGLOBIN A2 (QUANT)	2.5			%	1.8-3.5
INTERPRETATION					

*OK 3/1/05*

NORMAL PHENOTYPE.  
NORMAL HEMOGLOBIN DISTRIBUTION, NO HGS, HGC OR  
OTHER ABNORMAL HEMOGLOBIN OBSERVED.

CHOLESTEROL, TOTAL	187			MG/DL	<200 CB
BT	19			U/L	2-50 CB

BC (INCLUDES DIFF/PLT)					
WHITE BLOOD CELL COUNT	5.9			THOUS/MCL	3.8-10.8 CB
RED BLOOD CELL COUNT	4.78			MILL/MCL	4.20-5.10
HEMOGLOBIN		15.6 H		G/DL	13.2-15.5
HEMATOCRIT	44.3			%	38.5-45.0
MCV	92.9			FL	80.0-100.0
MCH	32.6			PG	27.0-33.0
MCHC	35.1			G/DL	32.0-36.0
RDW	12.7			%	11.0-15.0
PLATELET COUNT	200			THOUS/MCL	140-400
ABSOLUTE NEUTROPHILS	3552			CELLS/MCL	1500-7800
ABSOLUTE LYMPHOCYTES	1971			CELLS/MCL	850-3900
ABSOLUTE MONOCYTES	325			CELLS/MCL	200-950
ABSOLUTE EOSINOPHILS	35			CELLS/MCL	15-500
ABSOLUTE BASOPHILS	18			CELLS/MCL	0-200
NEUTROPHILS	60.2			%	
LYMPHOCYTES	33.4			%	
MONOCYTES	5.5			%	
EOSINOPHILS	0.6			%	
BASOPHILS	0.3			%	

>> REPORT CONTINUED ON NEXT PAGE - ID, DONOR#2494 WX313535F <<