CLI Donor 2281

Genetic Testing Summary

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

Last Updated: 03/12/24

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 87 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.

genzyme Genetics

Cystic Fibr[illegible]s Mutation Analysis

Patient Name: Donor 2281, .
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

Fairfax Cryobank
[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 08/15/2003
Date Received: 08/16/2003
Lab ID: [redacted]
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Caucasian

Indication: Carrier test / Gamete donor

RESULTS: Negative for the 87 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	CF87 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 87 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. The assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V, I506M and I507V.

This test was developed and its performance characteristics determined by Genzyme Genetics. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.

Under the direction of:

Lynne Rosenblum-Vos

Lynne Rosenblum-Vos, Ph. D.

Date: 08/22/2003

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

genzyme Genetics

Cystic fibrosis mutations analyzed

ΔF508	R1162X	1898+1G>A
ΔI507	R117C	1898+5G>T
ΔF311	R117H	1949del84
A455E	R1283M	2043delG
A559T	R334W	2143delT
C524X	R347H	2183delAA>G
D1152H	R347P	2184delA
D1270N	R352Q	2307insA
E60X	R553X	2789+5G>A
G178R	R560T	2869insG
G330X	S1196X	3120+1G>A
G480C	S1251N	3120G>A
G542X	S1255X	3659delC
G551D	S364P	3662delA
G85E	S549I	3791delC
G91R	S549N	3821delT
I148T	S549R	3849+10kbC>T
K710X	T338I	3849+4A>G
L206W	V520F	3876delA
M1101K	W1089X	3905insT
N1303K	W1282X	394delTT
P574H	Y1092X	405+1G>A
Q1238X	Y563D	405+3A>C
Q359K/T360K	1078delT	444delA
Q493X	1161delC	574delA
Q552X	1609delCA	621+1G>T
Q890X	1677delTA	711+1G>T
R1066C	1717-1G>A	711+5G>A
R1158X	1812-1G>A	712-1G>T

genzyme Genetics

Chromosome Analysis

Patient Name: Donor, #2281
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

Fairfax Cryobank
[redacted]

DOB: Not Given
SSN:

Date Collected: 08/29/2003
Date Received: 08/30/2003
Lab ID: [redacted]
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: No clinical indication provided

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	7	**Number of Cultures:**	2	**Banding Resolution:**	500
Metaphases Karyotyped:	3			**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.

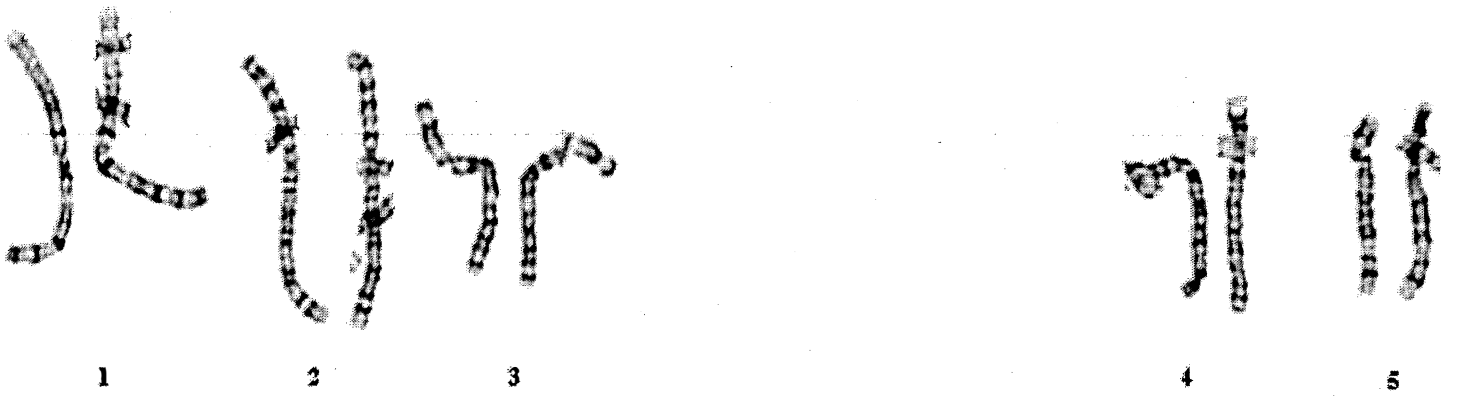
Signed:

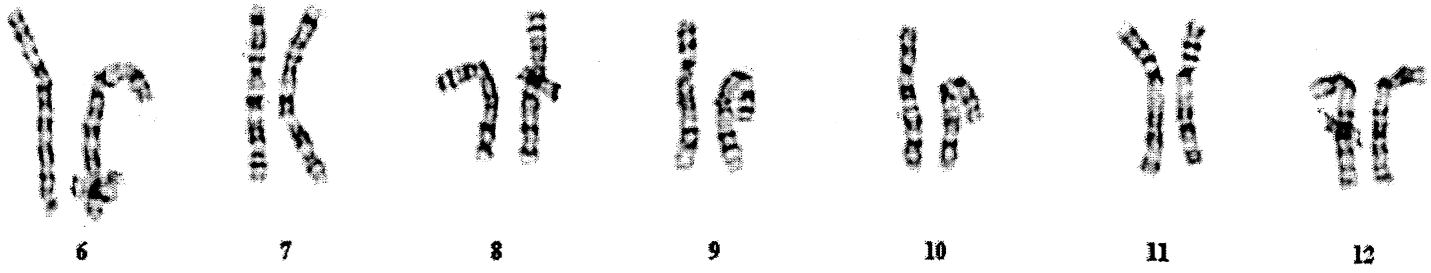
Jay W. Moore, Ph.D. FFACMG

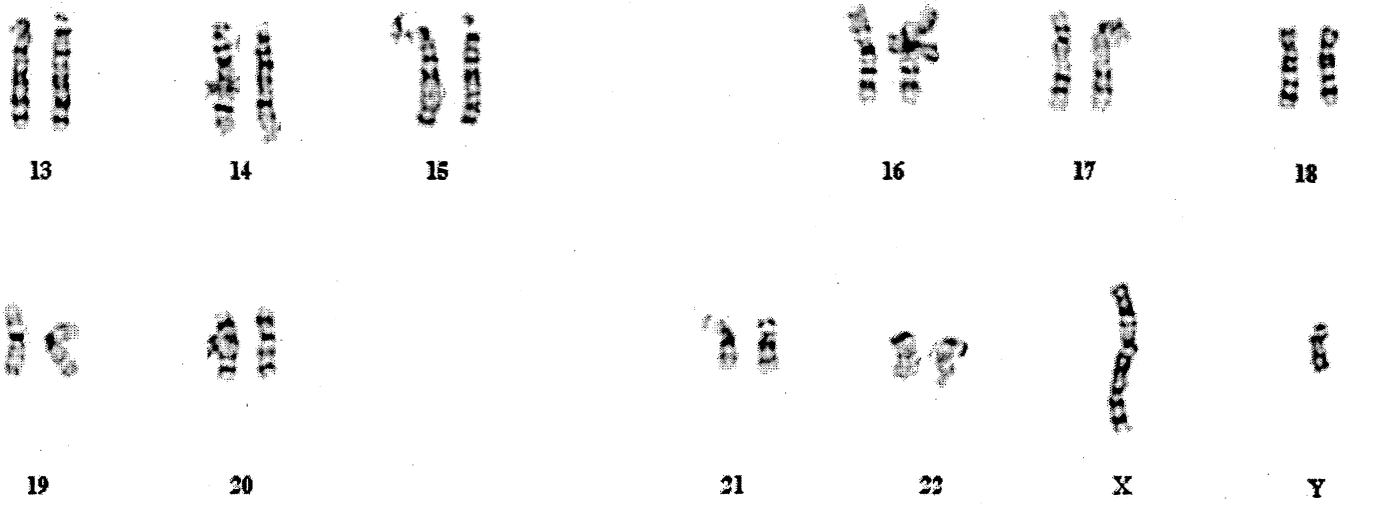
Date: 09/08/2003

Testing Performed At Genzyme Genetics 10421 University Center Drive Suite 100 Tampa, FL 33612 1-800-966-4440

genzyme
Genetics







Specimen #: [redacted]
Specimen Type: BLDPER
Patient Name: Donor, #2281
Image ID: [redacted]
Karyotype: 46,XY

Dept ID: B1
Date Received: 08/30/2003
Date Reviewed: 09/08/2003
Reviewed By: JWM

genzyme
GENERAL
genetics

Quest
Diagnostics
1901 Sulphur Spring Road • Baltimore, Maryland 21227-0580
Main Laboratory 410-247-9100 • D.C. Area 301-621-6900 • Outside Maryland 1-800-LAB-XCEL

ROBERT R.L. SMITH, M.D.
Medical Director

[redacted]

[redacted]
3015 WILLIAMS DR STE 110 (N1,A)
FAIRFAX VA 22031

DONOR #2281
PATIENT ID #: 2281[redacted]
COLL. DATE & TIME: 08-15-03

SPECIMEN COLLECTED: 08/15/2003
COMPLETED REPORT: 08/19/2003 15:04

PATIENT NAME	DATE	AGE	SEX	LAB NUMBER	LAB REPORT
DONOR #2281	08/15/2003	?	M	[redacted]	

HEMATOLOGY:

*WHITE BLOOD CELL COUNT	3.5	THOUS/MCL	(3.8-10.8)
RED BLOOD CELL COUNT	5.03	MILL/MCL	(4.20-5.80)
HEMOGLOBIN	15.7	G/DL	(13.2-17.1)
HEMATOCRIT	47.3	%	(38.5-50.0)
MCV	94	FL	(80-100)
MCH	31.1	PG	(27-33)
MCHC	33.1	G/DL	(32-36)
*RDW	15.2	%	(11.0-15.0)
PLATELET COUNT	145	THOUS/MCL	(140-400)
MPV	7.6	FL	(7.5-11.5)
ABSOLUTE NEUTROPHILS	1740	CELLS/MCL	(1500-7800)
ABSOLUTE LYMPHOCYTES	1292	CELLS/MCL	(850-3900)
ABSOLUTE MONOCYTES	256	CELLS/MCL	(200-950)
ABSOLUTE EOSINOPHILS	175	CELLS/MCL	(15-500)
ABSOLUTE BASOPHILS	39	CELLS/MCL	(0-200)
NEUTROPHILS	49.7	%	
LYMPHOCYTES	36.9	%	
REACTIVE LYMPHOCYTES	0.0	%	(0-9)
MONOCYTES	7.3	%	
EOSINOPHILS	5.0	%	
BASOPHILS	1.1	%	

COMMENT:

Platelets appear adequate.

Robert R.L. Smith, M.D.

SIGNATURE

DATE REPORTED

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Medical Director

[redacted]

[redacted]
3015 WILLIAMS DR STE 110 (N1,A)
FAIRFAX VA 22031

DONOR #2281
PATIENT ID #: 2281[redacted]
COLL. DATE & TIME: 08-15-03
,

SPECIMEN COLLECTED: 08/15/2003
COMPLETED REPORT: 08/19/2003 15:04

PATIENT NAME	DATE	AGE	SEX	LAB NUMBER	
DONOR #2281	08/15/2003	?	M	[redacted]	LAB REPORT

[redacted]

HEMOGLOBIN A1----------	97.5 % ()96.0)	
HEMOGLOBIN F----------	NONE DETECTED	(0.0-1.9)
HEMOGLOBIN A2----------	2.5 %	(1.8-3.5)
HGB SCREEN INTERPRETATION----------		

THE HEMOGLOBINOPATHY SCREEN IS NORMAL.

ABNORMAL HEMOGLOBIN #1 %:----------	0.0 %
HTLV I-II ANTIBODY----------	Nonreactive

[See note: a]

Please be advised that Quest Diagnostics Nichols Institute, Chantilly, VA, is no longer performing testing to determine the suitability of patient specimens for blood or organ donation.
The test code ordered is for non-donor use, therefore, testing was performed for clinical purposes only.

Reference value: Nonreactive

[redacted]

Robert R.L. Smith, MD

SIGNATURE

DATE REPORTED