CLI Donor 1982

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: English, Irish, Native American 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 99 mutations in the CFTR gene	1/343
Tay Sachs Enzyme Analysis	Non-carrier by Hexosaminidase A analysis	

*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.

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Cystic Fibrosis Mutation Analysis

Patient Name: Donor 1982, .
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

Fairfax Cryobank
[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 09/18/2006
Date Received: 09/19/2006
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/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 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: Zhaoqing Zhou

Zhaoqing Zhou, Ph.D.

Date: 09/25/2006

Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-2[illegible]-7357

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MUTATIONS ANALYZED

ΔF311	3120+1G>A	712-1G>T	Q359K/T360K	S549N
ΔF508	3120G>A	935delA	Q493X	S549R T>G
ΔI507	3171delC	936delTA	Q552X	T338I
1078delT	3199del6	A455E	Q890X	V520F
1288insTA	3659delC	A559T	R1066C	W1089X
1677delTA	3667del4	C524X	R1158X	W1204X
1717-1G>A	3791delC	CFTRdele2,3	R1162X	W1282X
1812-1G>A	3849+10kbC>T	D1152H	R117C	Y1092X C>A
1898+1G>A	3876delA	E60X	R117H	Y1092X C>G
1898+5G>T	3905insT	E92X	R334W	Y122X
1949del84	394delTT	G178R	R347H	
2043delG	4016insT	G330X	R347P	
2055del9>A	405+1G>A	G480C	R352Q	
2105del13ins5	405+3A>C	G542X	R553X	
2108delA	406-1G>A	G551D	R560T	
2143delT	444delA	G85E	R709X	
2183delAA>G	457TAT>G	K710X	R75X	
2184delA	574delA	L206W	R764X	
2184insA	621+1G>T	M1101K	S1196X	
2307insA	663delT	N1303K	S1251N	
2789+5G>A	711+1G>T	P574H	S1255X	
2869insG	711+5G>A	Q1238X	S364P	

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.

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Chromosome Analysis

Patient Name: Donor, 1982
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

[redacted]

DOB: Not Given
SSN:

Date Collected: 09/26/2006
Date Received: 09/27/2006
Lab ID: ID#1[redacted]
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted:	30			**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.

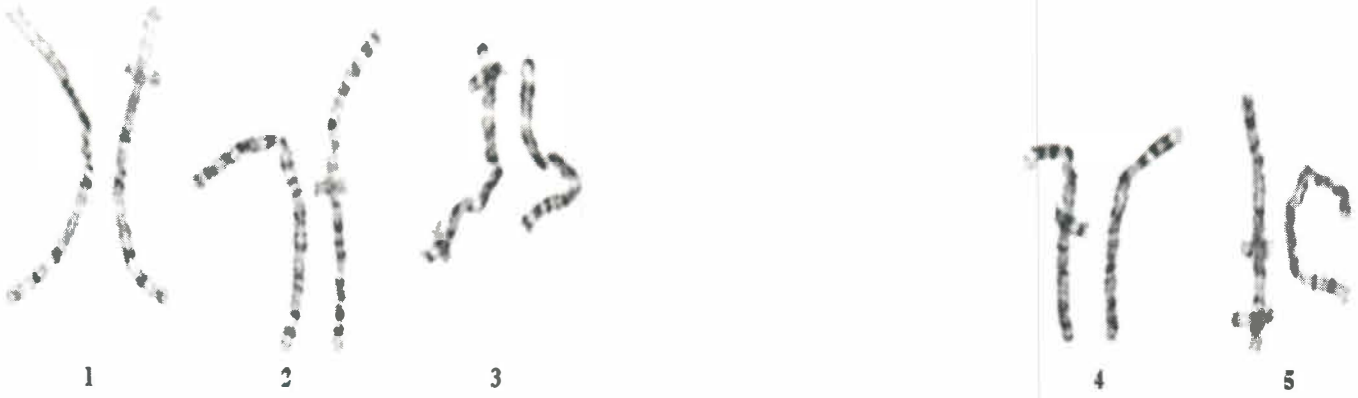
10/4/06

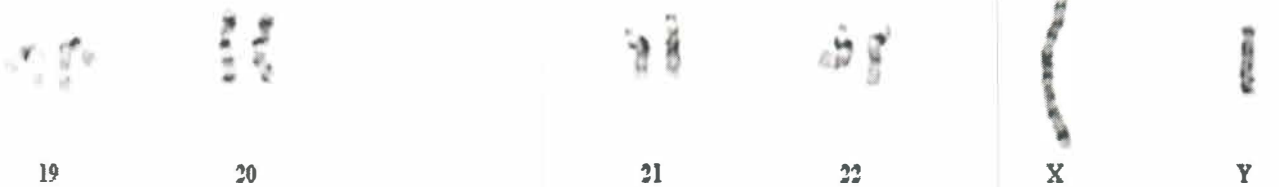
Signed:

Jay W. Moore, Ph.D. FFACMG

Date: 10/04/2006

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





Specimen #[redacted]
Specimen Type: BLDPER
Patient Name: Donor, 1982
Image ID: [redacted]
Karyotype: 46,XY

Dept ID: B1
Date Received: 09/27/2006
Date Reviewed: 10/04/2006
Reviewed By: JWM

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QUEST DIAGNOSTICS INCORPORATED

PATIENT INFORMATION
DONOR,1982

DOB: Age:
GENDER: M

REPORT STATUS **Final**

ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
[redacted]
FAIRFAX CRYOBANK
[redacted]

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

ID: [redacted]

COLLECTED: 09/18/2006 10:00
RECEIVED: 09/19/2006 04:24
REPORTED: 09/20/2006 05:38

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
HEMOGLOBINOPATHY INDICES				IG
RED BLOOD CELL COUNT	5.59		4.20-5.80 Million/uL	
HEMOGLOBIN	16.5		13.2-17.1 g/dL	
HEMATOCRIT	47.2		38.5-50.0 %	
MCV	84.4		80.0-100.0 fL	
MCH	29.5		27.0-33.0 pg	
RDW	13.2		11.0-15.0 %	
HEMOGLOBINOPATHY EVALUATION				IG
HEMOGLOBIN A1	97.9		>96.0 %	
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.1		1.8-3.5 %	
INTERPRETATION	NORMAL PHENOTYPE.			
CHOLESTEROL, TOTAL	153		<200 mg/dL	IG
AST	13		3-50 U/L	IG
ALT	10		3-60 U/L	IG
CBC (INCLUDES DIFF/PLT)				IG
WHITE BLOOD CELL COUNT	5.4		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	5.59		4.20-5.80 Million/uL	
HEMOGLOBIN	16.5		13.2-17.1 g/dL	
HEMATOCRIT	47.2		38.5-50.0 %	
MCV	84.4		80.0-100.0 fL	
MCH	29.5		27.0-33.0 pg	
MCHC	34.9		32.0-36.0 g/dL	
RDW	13.2		11.0-15.0 %	
PLATELET COUNT	183		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	3553		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1404		850-3900 cells/uL	
ABSOLUTE MONOCYTES	346		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	81		15-500 cells/uL	
ABSOLUTE BASOPHILS	16		0-200 cells/uL	
NEUTROPHILS	65.8		%	
LYMPHOCYTES	26.0		%	
MONOCYTES	6.4		%	
EOSINOPHILS	1.5		%	
BASOPHILS	0.3		%	

ENTERED 9/22/06

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Tay-Sachs Enzyme Analysis

Patient Name: Donor #1982, .
Referring Physician: [redacted].
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

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[redacted]

DOB: Not Given
SSN:

Date Collected: 05/01/2008
Date Received: 05/03/2008
Lab ID:
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 1777 nmol/mg protein**
Hexosaminidase Percent A: 65.3

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥55%	≥55%
Expected Carrier Range:	Hex A	20 - 48%	20 - 49%

INTERPRETATION: NON CARRIER

This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

ENTERED 5/14/08

Under the direction of: Stanford Marenberg, PHD, ABCC

Stanford Marenberg, Ph.D.

Date: 05/12/2008

Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436