CLI Donor 1130

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, Danish, Norwegian Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities 46, XY, 15ps+ where the 15ps+ is considered a normal variant with no clinical significance. See attached.
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.

genzyme Genetics

Cystic Fibr is Mutation Analysis

Patient Name: Donor, CLI 1130
Referring Physician:
Specimen #: [redacted]
Patient ID: [redacted]

Client #: [redacted]
Case #: [redacted]

Fairfax Cryobank
Genetics and IVF Institute
3015 Williams Drive
Suite 110
Fairfax VA 22031

DOB: Not Given
Sex: M
SSN:

Date Collected: 06/14/2004
Date Received: 06/16/2004
Lab ID: [redacted]
Hospital ID:
Specimen Type: BLOOD

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. The assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V, I506M and I507V.

Under the direction of: Elizabeth M. Rohlfs

Elizabeth M Rohlfs, Ph. D.

Date: 06/23/2004

Mount Sinai

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MOUNT SINAI LABORATORY (CYTOGENETICS) (212) 241-7518

June 29, 1995

Dr. Judith P. Willner
Director, Clinical Genetics
Department of Human Genetics

Patient: CL-1130
Specimen: Peripheral blood
Collection Dt.: 06/23/95
Received Dt.: 06/23/95
Lab No. [redacted]
Accession Dt. 06/23/95

Dear Dr. Willner:

Chromosome studies were performed on a peripheral blood specimen received from CL-1130 on June 23, 1995.

Model count:	46	Karyotype:	46,XY,15ps+
Cells counted:	20	Cells analyzed:	5
Cells photographed:	8	Cells karyotyped:	2
Banding used:	GTG (G-bands by trypsin using Giemsa)		

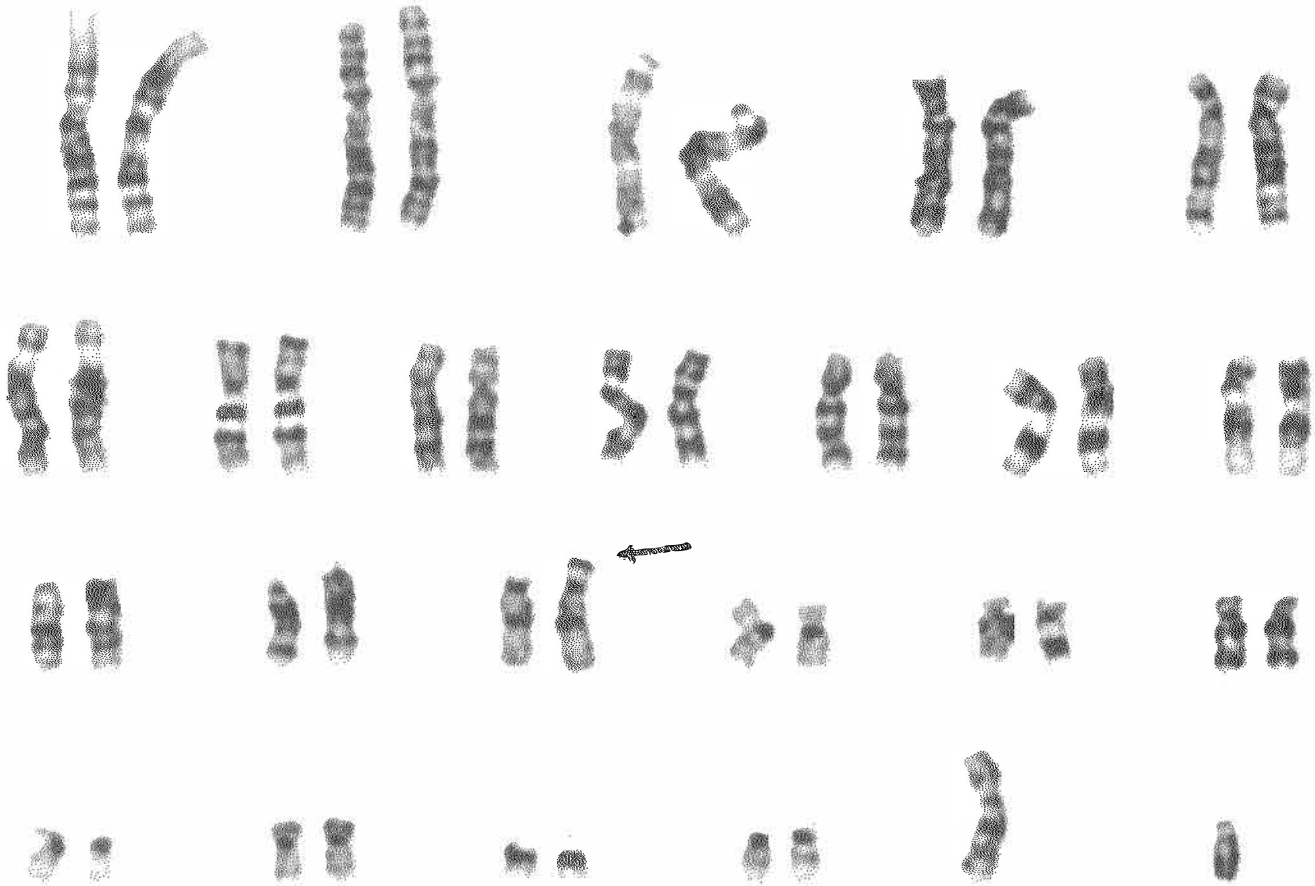
Chromosome analysis of 20 G-banded metaphase cells revealed an normal chromosome complement 46,XY,15ps+ in all the cells examined, including a large satellite on one of the chromosome 15's. This is considered a normal variant.

Sincerely,

Arvind Babu, Ph.D.
Cytogeneticist

R.J. Desnick, Ph.D., M.D.
Professor and Chairman
Department of Human Genetics

Cytogenetics Laboratory
Department of Human Genetics
Mount Sinai School of Medicine
New York, NY



Case: [redacted] Slide: 2A Cell: 24 Patient: CL-1130

Case name: [redacted] Date: 06/26/95 +6,XY,15ps+

Quest Diagnostics®

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 800.323.5917

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION: [redacted]

COLLECTED: 06/14/2004 12:45 CT
RECEIVED: 06/15/2004 05:56 CT
REPORTED: 06/30/2004 11:40 CT

PATIENT INFORMATION
ID,1130

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

REPORT STATUS FINAL

ORDERING PHYSICIAN

CLIENT INFORMATION
[redacted]

COMMENTS: ADULT

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.01		4.20-5.80 MILL/MCL	CB
HEMOGLOBIN	15.9		13.2-17.1 G/DL	
HEMATOCRIT	46.4		38.5-50.0 %	
MCV	92.5		80.0-100.0 FL	
MCH	31.6		27.0-33.0 PG	
RDW	12.9		11.0-15.0 %	
HEMOGLOBIN A1	97.7		>96.0 %	CB
FETAL HEMOGLOBIN	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.3		1.8-3.5 %	
INTERPRETATION				
NORMAL PHENOTYPE.				
NORMAL HEMOGLOBIN DISTRIBUTION, NO HGS, HGC OR				
OTHER ABNORMAL HEMOGLOBIN OBSERVED.				
CHOLESTEROL, TOTAL		209 H	<200 MG/DL	CB
AST	23		2-50 U/L	CB
CBC (INCLUDES DIFF/PLT)				CB
WHITE BLOOD CELL COUNT	5.6		3.8-10.8 THOUS/MCL	
RED BLOOD CELL COUNT	5.01		4.20-5.80 MILL/MCL	
HEMOGLOBIN	15.9		13.2-17.1 G/DL	
HEMATOCRIT	46.4		38.5-50.0 %	
MCV	92.5		80.0-100.0 FL	
MCH	31.6		27.0-33.0 PG	
MCHC	34.2		32.0-36.0 G/DL	
RDW	12.9		11.0-15.0 %	
PLATELET COUNT	195		140-400 THOUS/MCL	
ABSOLUTE NEUTROPHILS	3242		1500-7800 CELLS/MCL	
ABSOLUTE LYMPHOCYTES	1904		850-3900 CELLS/MCL	
ABSOLUTE MONOCYTES	370		200-950 CELLS/MCL	
ABSOLUTE EOSINOPHILS	56		15-500 CELLS/MCL	
ABSOLUTE BASOPHILS	28		0-200 CELLS/MCL	
NEUTROPHILS	57.9		%	

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PATIENT INFORMATION
ID,1130

REPORT STATUS FINAL

QUEST DIAGNOSTICS INCORPORATED

ORDERING PHYSICIAN

DOB: AGE:

REPORTED: 06/30/2004 11:40 CT GENDER: M FASTING: U

Test Name	In Range	Out of Range	Reference Range	Lab
Hard Copy with additional information to follow				
LYMPHOCYTES	34.0		%	
MONOCYTES	6.6		%	
EOSINOPHILS	1.0		%	
BASOPHILS	0.5		%	

PERFORMING LABORATORY INFORMATION
CB QUEST DIAGNOSTICS WOOD DALE, 1355 MITTEL BOULEVARD, WOOD DALE, IL 60191
Laboratory Director: ANTHONY V. THOMAS, M.D., CLIA: 14D0417052