Donor 2145

Genetic Testing Summary

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

Last Updated: 04/03/23

Donor Reported Ancestry: French, Scottish, English 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 for 87 mutations in the CFTR gene	1/325
Alpha-1 Antitrypsin Deficiency carrier screening	Negative for S and Z mutations in the SERPINA1 gene	Reduced risk

*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 Fibrosis Mutation Analysis

Patient Name: Donor, 2145
Referring Physician: [redacted]
Specimen #: [redacted]
Patient ID: [redacted]
Client #: [redacted]
Case #: [redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 12/04/2000
Date Received: 04/30/2003
Lab ID: [redacted]
Hospital ID:
Specimen Type: **DNA**

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

Ethnicity: Caucasian

Indication: Carrier test / Gamete donor

RESULTS: Negative for the 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:

This analysis was performed on extracted DNA provided by Genetics and IVF Institute and identified as above.

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: 05/08/2003

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

GENETICS & IVF INSTITUTE

3022 Javier Road, Fairfax, Virginia 22031 (800) 654-GENE

Joseph D. Schulman, M.D., *Medical Director*

Name:	[redacted] **Donor 2145**	**Family No.:**	[redacted]
ID No.:	[redacted]	**Sample No.:**	
Specimen:	Peripheral blood	**Date Drawn:**	12/04/2000
Referred By:	[redacted]	**Received:**	12/04/2000

Test: α_1-Antitrypsin S and Z mutations.

PI*S Result: Negative.
PI*Z Result: Negative.

Conclusion: This individual is not a carrier of the S or Z α_1-antitrypsin mutations.

Comment: Deficiency in the protease inhibitor α_1-antitrypsin can cause chronic obstructive pulmonary disease (emphysema). Deficiencies in this enzyme occur through a variety of different mutations in the α_1-antitrypsin gene. Two, called *PI*Z* and *PI*S,* are particularly common. Individuals who inherit two *PI*Z* alleles have a high risk of developing emphysema. They also may experience transient hepatitis or permanent liver damage in childhood or later in life. Individuals who inherit one *PI*Z* and one *PI*S* allele also have a somewhat increased risk for emphysema and liver disease. Persons who have one α_1-antitrypsin allele that is intact and one that has the *PI*Z* mutation may have some increased risk of emphysema, especially with smoking. Since about 1 person in 20 in the U.S. is a carrier of a *PI*S* or *PI*Z* allele, healthy adults may want screening to determine if they and their partner are at risk of having a child with two deficient alleles. If results are positive, genetic counseling is indicated.

Note: This test examines the α_1-antitrypsin gene at the specific positions associated with the common S and Z mutations. Mutations other than S and Z would not be detected. This method differs from PI Typing, in which the protein itself is examined and classified as S, Z, M (normal), or another variant.

Dec 6, 2000
Date

Anne Maddalena, PhD, ABMG
Laboratory Director
W. Christine Spence, PhD, ABMG
Associate Director

GENETICS & IVF INSTITUTE

3022 Javier Road, Fairfax, Virginia 22031 (800) 654-GENE

Joseph D. Schulman, M.D., *Medical Director*

Patient: [redacted] **Donor 2145**

PB Lab. No.: [redacted]

Hospital/Chart No: Fairfax

D.O.B./Age: [redacted]

Physician Name: [redacted]

Preliminary: --

Source No.: 1.007

Final: 01-22-01

Date Received: 12-22-00 **Collected:** 12-22-00

Specimen: Blood

Test: Chromosome Analysis

Band Resolution:

400 550 850

Number of Cells Counted: 20

Modal Chromosome Count: 46

Number of Cells Karyotyped: 2

Karyotype: **46,XY**

Impression: **Normal Male Karyotype**

Comment:

1/22/01

Clinical Cytogeneticists
Wayne S. Stanley, Ph.D
Lillian D. Killos, Ph.D.
Julie Leana-Cox, Ph.D.

Most chromosome variants of no clinical significance, if present, are not reported.

Genetics & IVF Institute
Cytogenetics Laboratory

Case name: [redacted]

Patient name: [redacted]

Result: 46,XY

Case comment:

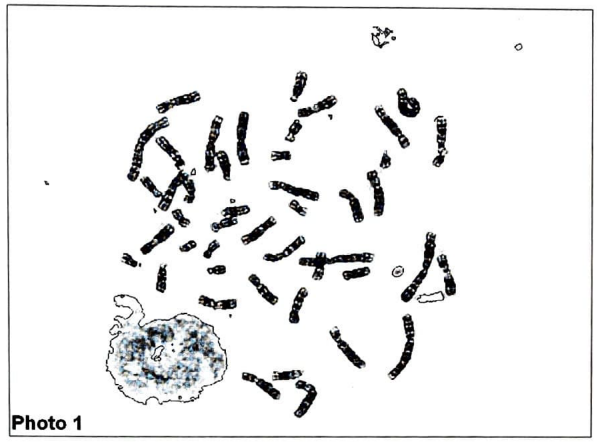
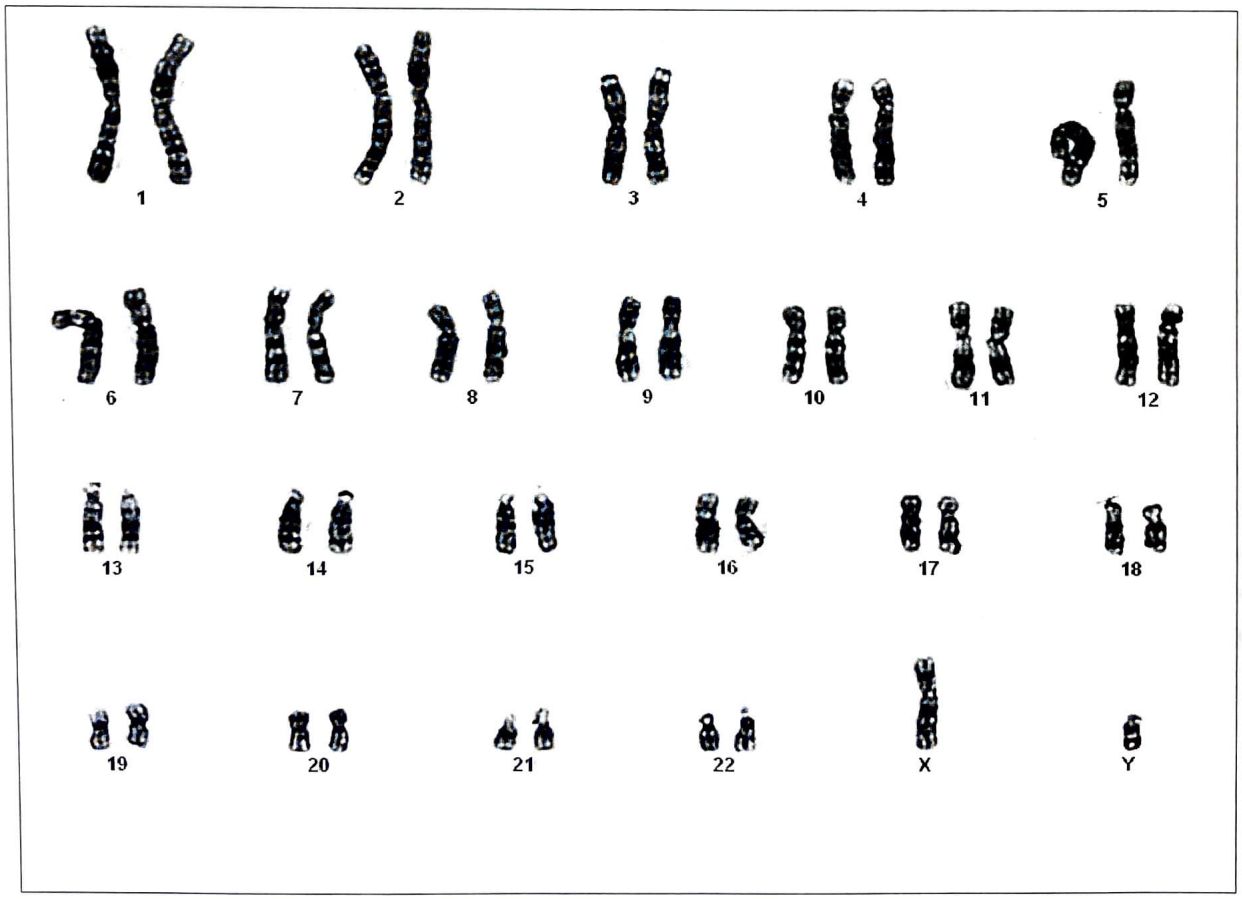


Photo 1



Technologist: SK

SPECIALTY LABORATORIES
2211 Michigan Avenue 310-828-6543
Santa Monica, CA 90404-3900 800-421-4449

SPECIALTY # [redacted]
CLIENT #

PATIENT NAME: DONOR, #2145

PHYSICIAN: [redacted]

NOTES:
PATIENT ID: [redacted]
SPECIMEN ID:

FAIRFAX CRYOBANK
ATTN: STEVE POOL, PHD
3015 WILLIAMS DR STE 110

FAIRFAX VA 22031

DOB: N/A AGE: unknown
SEX: Male

DRAWN:	12/04/00 12:00
RECEIVED:	12/05/00 15:30
PRINTED:	12/16/00 01:03
FINAL REPORT:	12/16/00 01:03 FINAL

TEST NAME	RESULTS		REFERENCE RANGE
HEMOGLOBIN VARIANT SCREEN			
Hemoglobin A	96.7	%	(93.5-98.3)
Hemoglobin A2	2.9	%	(2.1-3.0)
Hemoglobin F	< 1.0	%	(< 1.0)
Hemoglobin C	Negative		Negative
Hemoglobin D	Negative		Negative
Hemoglobin G	Negative		Negative
Hemoglobin S	Negative		Negative
CHOLESTEROL, TOTAL			
Cholesterol, Total	158	mg/dL	(< 200)

REFERENCE RANGES for Cholesterol, Total:

Age	mg/dL	Risk Level
Less than 2	No range established	
2 - 18 years	< 170	Desirable
	170 - 199	Borderline High
	> 200	High
Adult	< 200	Desirable
	200 - 239	Borderline High
	> 240	High

REPORT COMPLETED PLEASE FILE

Tests Requested:
CUSTOM CRYOBANK PROFILE A, ALT (SGPT), AST (SGOT), HEMOGLOBIN VARIANT SCREEN, CHOLESTEROL, TOTAL

James B. Peter, M.D., Ph.D

QUEST DIAGNOSTICS INCORPORATED
1 SULPHUR SPRING ROAD
BALTIMORE, MD 21227
(410) 247-9100

[redacted]9)
[redacted] DR STE 110 (N1,M)
FAIRFAX VA 22031

DONOR#2145

SPECIMEN COLLECTED: 12/04/2000 12:00
COMPLETED REPORT: 12/05/2000 13:43

DONOR#2145 12/04/2000 ? M [redacted]6

HEMATOLOGY:

RBC--------	5.05	M/CMM(4.4-5.9)	WBC-------	5.7	K/CMM(	3.9-11.3)
HGB--------	15.5	GM/DL(14.0-18.0)	BANDS-----	0	%	(0-10)
HCT--------	43.7	% (38.0-50.0)	POLYS-----	60	%	(42-78)
MCV--------	86	FL (80-100)	LYMPHS----	28	%	(15-45)
MCH--------	30.8	PG (26.0-33.0)	MONOS-----	7	%	(0-12)
MCHC-------	35.6	G/DL (31.0-36.0)	EOSIN-----	4	%	(0-7)
			BASOS-----	1	%	(0-2)
			ATYP LYMPH	0	%	(0-4)

COMMENT:

Platelets appear adequate.

PLATELET COUNT----------	231	K/CMM	(140-440)
MPV----------	7.8	FL	(6.3-10.3)
MONOCYTES, ABSOLUTE----------	399	/CMM	(0-1356)
Reference range change effective 11/10/2000.			
LYMPHOCYTES, ABSOLUTE----------	1596	/CMM	(850-4100)
Reference range change effective 11/10/2000.			
BASOPHILS, ABSOLUTE----------	57	/CMM	(0-226)
Reference range change effective 11/10/2000.			
SEGMENTED NEUTROPHILS,ABSOLUTE---	3420	/CMM	(1638-8814)
Reference range change effective 11/10/2000.			
EOSINOPHILS, ABSOLUTE----------	228	/CMM	(0-791)
Reference range change effective 11/10/2000.			
RDW----------	12.3	%	(11.5-14.5)

IMMUNOHEMATOLOGY:
BLOOD GROUP---------- A