



Donor 1886

Genetic Testing Summary

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

Last Updated: 03/18/22

Donor Reported Ancestry: Chinese

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	Insufficient data to estimate residual risk
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.



QUEST DIAGNOSTICS INCORPORATED
 CLIENT SERVICE 800.824.6152

PATIENT INFORMATION
 DONOR1886, ONLY

REPORT STATUS FINAL

ORDERING PHYSICIAN

SPECIMEN INFORMATION

SPECIMEN: [REDACTED]
 REQUISITION: [REDACTED]

DOB: AGE:
 GENDER: M
 SSN:
 ID: 1886-021212
 PHONE:

CLIENT INFORMATION

G41550 9999999
 FAIRFAX CRYOBANK
 [REDACTED]

COLLECTED: 12/12/2002 08:40 CT
 RECEIVED:
 REPORTED: 12/18/2002 08:03 CT

Test Name	In Range	Out of Range	Reference Range	Lab
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HEMOGLOBINOPATHY EVALUATION

RED BLOOD CELL COUNT	5.46		4.20-5.80 MILL/MCL	IG
HEMOGLOBIN	17.1		13.2-17.1 G/DL	
HEMATOCRIT	50.0		38.5-50.0 %	
MCV	91.5		80.0-100.0 FL	
MCH	31.3		27.0-33.0 PG	
RDW	12.3		11.0-15.0 %	
HEMOGLOBIN A1	>96.0		>96.0 %	IG
FETAL HEMOGLOBIN	<2.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.5		1.5-3.5 %	

INTERPRETATION

Hemoglobin variant analysis by HPLC reveals a normal pattern.

L.C. HARVEY, M.D., ELECT-SIGNATURE

HIV 1 HIV 2 AB, EIA, POSITIVES REFLEXED TO HIV-1 WB

HIV 1 HIV 2 ANTIBODY

SCREEN

HIV-1/HIV-2 AB SCREEN, EIA NONREACTIVE

Reference Range:

NONREACTIVE

No antibodies to HIV-1 and HIV-2 were detected. If the clinical situation warrants, a repeat of this test in six months on a freshly drawn sample may rule out the possibility of a false negative due to inadequate time for seroconversion to have occurred.

HIV 1 WESTERN BLOT

Reference Range:

NEGATIVE

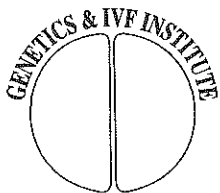
TNP-Supplemental testing not performed.

CHOLESTEROL, TOTAL	165		<200 MG/DL	IG
AST	25		2-50 U/L	IG
ALT	24		2-60 U/L	IG

HTLV I/II ANTIBODY, EIA

* HTLV-I/II ANTIBODY NONREACTIVE

Reference Range:



GENETICS & IVF INSTITUTE
3022 Javier Road Fairfax, Virginia 22031 (800) 654-GENE

CYTOGENETIC RESULTS

Patient: DONOR # 1886

PB Lab. No.: B15253

Hospital/Chart No.: [REDACTED]

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

Physician Name: [REDACTED]

Source No.: 1.007

Collected: 01-16-2003 **Date Received:** 01-17-2003

Final: 01-27-2003

Specimen: Blood

Chromosome Analysis

Type(s) of Banding: GTW

Band Resolution: 450-500

Total Cells Examined: 20


Cells Analyzed Microscopically: 5

Digitized Karyotypes: 2

Karyotype: 46,XY

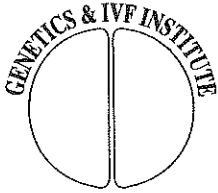
Interpretation: Normal male karyotype.

No consistent numerical or structural abnormalities were noted.



Clinical Cytogeneticists
Wayne S. Stanley, Ph.D. Denise A. Batista, Ph.D.
Lillian D. Killos, Ph.D. Chien-Song K. Shan, M.D.
Julie Leana-Cox, Ph.D.

Most chromosome variants of no clinical significance, if present, are not reported. This analysis does not rule out the possibility of subtle structural chromosome abnormalities, low frequency chromosome mosaicism, or defects of non-chromosomal etiology.



GENETICS & IVF INSTITUTE
3022 Javier Road Fairfax, Virginia 22031 (800) 654-GENE

Name: Donor 1886
ID No.: [REDACTED]
Specimen: Peripheral blood
Referred By: Megan Taylor

Family No.: [REDACTED]
Sample No.: [REDACTED]
Date Drawn: 12/12/2002
Received: 12/13/2002

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 18, 2002

Date


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

This test was developed and its performance characteristics determined by Genetics & IVF Institute. It has not been cleared or approved by the U.S. FDA. The FDA has determined that such clearance or approval is not necessary. Pursuant to the requirements of CLIA '88 this laboratory has established the test's accuracy and precision.

Patient Name: Donor, 1886
Referring Physician:
Specimen #:
Patient ID:

Client #:
Case #:

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

DOB: Not Given Date Collected: 05/12/2003
Sex: M Date Received: 05/13/2003
SSN: Lab ID:
 Hospital ID:
 Specimen Type: **BLDPER**

Ethnicity: Asian

Indication: Carrier test / Gamete donor

RESULTS: Negative for the mutations analyzed

INTERPRETATION
The sample provided is negative for the mutations analyzed. C

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:

Stephanie Hallam

Date: 05/20/2003



Stephanie Hallam, Ph.D.

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