Donor 4114

Genetic Testing Summary

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

Last Updated: 5/25/23

Donor Reported Ancestry: Romanian 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 for 97 mutations in the CFTR gene	1/343
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon in the SMN1 gene	1/648

*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, 4114
Referring Physician: [redacted]
Specimen #: [redacted]
Patient ID: [redacted]
Client #: [redacted]
Case #: [redacted]

[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 05/30/2008
Date Received: 05/31/2008
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 and family relationship information provided and the current understanding of the molecular genetics of this condition.

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:

SMAC

Lynne Rosenblum-Vos

Lynne Rosenblum-Vos, Ph. D.

Date: 06/06/2008

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

genzyme

SMN1 Copy (mber Analysis

Patient Name: 4114 Donor
DOB: Age:
SSN #: Gender: M

[redacted]

Genzyme Specimen # [redacted]
Case #: [redacted] Patient ID #: [redacted]
Date Collected: 05/30/2008 Date Received: 05/31/2008

Referring Physician [redacted]
Genetic Counselor:

Client Lab ID #: [redacted]
Hospital ID #:
Specimen ID #:

Specimen Type: Peripheral Blood
Specimen(s) Received: 2 - Yellow (ACD) 10 ml round bottom tube(s)

Clinical Data: Gamete donor
Ethnicity: Caucasian

RESULTS: SMN1 copy number: 2

INTERPRETATION:

This individual's risk to be a carrier of SMA is reduced from approximately 1/41 to 1/648, based on an SMN1 copy number of two and a negative family history.

COMMENT:
Spinal muscular atrophy (SMA) is an autosomal recessive disease of variable age of onset and severity caused by mutations, most often deletions or gene conversions, resulting in zero copies of the survival motor neuron (SMN1) gene. Approximately 1/41 individuals without a family history of SMA is a carrier. This analysis identifies approximately 94% of carriers. Individuals with one copy of the SMN1 gene are predicted to be carriers of SMA. Individuals with two or more copies of the SMN1 gene have a reduced risk to be carriers of SMA.

This copy number analysis cannot detect the ~6% of individuals who are carriers of SMA as a result of: 1) 2 copies of the SMN1 gene on one chromosome and a deletion or gene conversion of SMN1 gene on the other chromosome or 2) small intragenic mutations within the SMN1 gene. This analysis also will not detect germline mosaicism or mutations in genes other than SMN1. SMA carriers falling into any of these categories have an SMN1 copy number result of 2 by dosage analysis. Additionally, de novo mutations have been reported in approximately 2% of SMA patients. Other false negative or false positive results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, or erroneous representation of family relationships.

METHOD:
Specimen DNA is isolated and amplified by real-time polymerase chain reaction (PCR) for exon 7 of the SMN1 gene and two reference genes. A mathematical algorithm is used to calculate the number of copies of SMN1. Sequencing of the primer and probe binding sites for the SMN1 real-time PCR reaction is performed on all fetal samples, and on samples from individuals with 1 copy of SMN1 on carrier testing, to rule out the presence of sequence variants which could interfere with analysis and interpretation.

REFERENCES:
Smith M, Calabro V, Chong B, et al. 2007. Eur J Hum Genet 15:759-766.
Online review of SMA: http://www.genereviews.org/profiles/sma

The test was developed and its performance characteristics have been determined by Genzyme. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjunction with clinical assessment when available.

Electronically Signed by: Hui Zhu, Ph.D. FACMG on 06/17/2008

Reported by: MS/jw

genzyme

Chromosome Analysis

Patient Name: Donor # 4114
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

[redacted]

DOB: Not Given
SSN:

Date Collected: 06/27/2008
Date Received: 06/30/2008
Lab ID:
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**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.

Signed: Veena Suri

Date: 07/08/2008

Veena Suri, Ph.D.

Testing Performed At Genzyme Genetics 521 West 57th Street New York, NY 10019 1-800-447-8881

Quest Diagnostics

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 800.323.5917

PATIENT INFORMATION
ID,4114

DOB: AGE:
GENDER: M FASTING: U

REPORT STATUS **FINAL**

ORDERING PHYSICIAN

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION:

ID:
PHONE:

CLIENT INFORMATION
[redacted]

COLLECTED: 05/30/2008 12:00 CT
RECEIVED: 05/31/2008 03:44 CT
REPORTED: 06/04/2008 07:43 CT

COMMENTS: REG# 4114-080530

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.86		4.20-5.80 Million/uL	CB
HEMOGLOBIN	14.5		13.2-17.1 g/dL	
HEMATOCRIT	42.4		38.5-50.0 %	
MCV	87.3		80.0-100.0 fL	
MCH	29.8		27.0-33.0 pg	
RDW	13.5		11.0-15.0 %	
HEMOGLOBIN A1	97.3		>96.0 %	CB
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.7		1.8-3.5 %	
INTERPRETATION	NORMAL PHENOTYPE. NORMAL HEMOGLOBIN DISTRIBUTION, NO HGS, HGC OR OTHER ABNORMAL HEMOGLOBIN OBSERVED.			
CHOLESTEROL, TOTAL		**122 L**	125-200 mg/dL	CB
AST	26		10-35 U/L	CB
ALT	18		9-60 U/L	CB
CBC (INCLUDES DIFF/PLT)				CB
WHITE BLOOD CELL COUNT	3.9		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.86		4.20-5.80 Million/uL	
HEMOGLOBIN	14.5		13.2-17.1 g/dL	
HEMATOCRIT	42.4		38.5-50.0 %	
MCV	87.3		80.0-100.0 fL	
MCH	29.8		27.0-33.0 pg	
MCHC	34.2		32.0-36.0 g/dL	
RDW	13.5		11.0-15.0 %	
PLATELET COUNT	204		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	2172		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1283		850-3900 cells/uL	
ABSOLUTE MONOCYTES	406		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	16		15-500 cells/uL	
ABSOLUTE BASOPHILS	23		0-200 cells/uL	

Quest Diagnostics

QUEST DIAGNOSTICS INCORPORATED

PATIENT INFORMATION
ID,4114

REPORT STATUS **FINAL**

ORDERING PHYSICIAN

DOB: AGE:

COLLECTED: 05/30/2008 12:00 CT
REPORTED: 06/04/2008 07:43 CT

GENDER: M FASTING: U

Test Name	In Range	Out of Range	Reference Range	Lab
NEUTROPHILS	55.7		%	
LYMPHOCYTES	32.9		%	
MONOCYTES	10.4		%	
EOSINOPHILS	0.4		%	
BASOPHILS	0.6		%	
ABO GROUP AND RH TYPE				CB
ABO GROUP	B			
RH TYPE	RH (D) POSITIVE			

PERFORMING LABORATORY INFORMATION

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

A duplicate report has been faxed to the following:
Faxed to: [redacted] on: 06/03/2008 11:04:31 AM