Donor 2446-CLI

Genetic Testing Summary

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

Last Updated: 10/10/22

Donor Reported Ancestry: Danish, Norwegian, Swedish 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 carrier screening	Negative by genotyping for 97 mutations in the CFTR gene (testing performed in 2010)	1/343
Spinal Muscular Atrophy (SMN1) carrier screening	Negative for deletions in the SMN1 gene	1/632

*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®

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 800.323.5917

SPECIMEN INFORMATION
SPECIMEN: WX426763V
REQUISITION: 3347092
LAB REF: 2446-100416

COLLECTED: 04/16/2010 11:10 CT
RECEIVED: 04/17/2010 04:03 CT
REPORTED: 04/21/2010 08:06 CT

PATIENT INFORMATION
DONOR,2446

DOB: AGE:
GENDER: M FASTING: N

ID:
PHONE:

REPORT STATUS **FINAL**

ORDERING PHYSICIAN
STERN,HARVEY J

CLIENT INFORMATION
[redacted] 4195000

Entered 4-28-10 lw

	Test Name	In Range	Out of Range	Reference Range	Lab
	HEMOGLOBINOPATHY EVALUATION				
	RED BLOOD CELL COUNT	5.42		4.20-5.80 Million/uL	CB
	HEMOGLOBIN	16.1		13.2-17.1 g/dL	
	HEMATOCRIT	46.4		38.5-50.0 %	
	MCV	85.6		80.0-100.0 fL	
	MCH	29.7		27.0-33.0 pg	
	RDW	13.0		11.0-15.0 %	
*	HEMOGLOBIN A	97.5		>96.0 %	CB
*	HEMOGLOBIN F	<1.0		<2.0 %	
*	HEMOGLOBIN A2 (QUANT)	2.5		1.8-3.5 %	
*	INTERPRETATION	NORMAL PHENOTYPE. NORMAL HEMOGLOBIN DISTRIBUTION, NO HGS, HGC OR OTHER ABNORMAL HEMOGLOBIN OBSERVED.			
	CBC (INCLUDES DIFF/PLT) (NO TEST INDICATED)				
	CBC (INCLUDES DIFF/PLT)				CB
	WHITE BLOOD CELL COUNT	7.1		3.8-10.8 Thousand/uL	
	RED BLOOD CELL COUNT	5.42		4.20-5.80 Million/uL	
	HEMOGLOBIN	16.1		13.2-17.1 g/dL	
	HEMATOCRIT	46.4		38.5-50.0 %	
	MCV	85.6		80.0-100.0 fL	
	MCH	29.7		27.0-33.0 pg	
	MCHC	34.7		32.0-36.0 g/dL	
	RDW	13.0		11.0-15.0 %	
	PLATELET COUNT	267		140-400 Thousand/uL	
	ABSOLUTE NEUTROPHILS	5410		1500-7800 cells/uL	
	ABSOLUTE LYMPHOCYTES	1328		850-3900 cells/uL	
	ABSOLUTE MONOCYTES	241		200-950 cells/uL	
	ABSOLUTE EOSINOPHILS	99		15-500 cells/uL	
	ABSOLUTE BASOPHILS	21		0-200 cells/uL	
	NEUTROPHILS	76.2		%	
	LYMPHOCYTES	18.7		%	
	MONOCYTES	3.4		%	
	EOSINOPHILS	1.4		%	
	BASOPHILS	0.3		%	

genzyme

SMN1 Copy [redacted]mber Analysis

Entered 4-28-10 lw

Patient Name: 2446 Donor
DOB: Age:
SSN #: Gender: Male

606452 / 348795
[redacted]

Genzyme Specimen #:61542755-6
Case #: 61428025 Patient ID #: 61279863
Date Collected: 04/16/2010 Date Received: 04/19/2010

Referring Physician: Steve Pool
Genetic Counselor:

Client Lab ID #: 2446100416
Hospital ID #:
Specimen ID #:

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

Clinical Data: Carrier Test/Gamete donor
Ethnicity: Caucasian

RESULTS: SMN1 copy number: 2 (Reduced Carrier Risk)

INTERPRETATION:

This individual has an SMN1 copy number of two. This result reduces but does not eliminate the risk to be a carrier of SMA. Ethnic specific risk reductions based on a negative family history and an SMN1 copy number of two are provided in the Comments section of this report.

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) in the survival motor neuron (SMN1) gene. Molecular testing assesses the number of copies of the SMN1 gene. Individuals with one copy of the SMN1 gene are predicted to be carriers of SMA. Individuals with two or more copies have a reduced risk to be carriers. (Affected individuals have 0 copies of the SMN1 gene.)

This copy number analysis cannot detect individuals who are carriers of SMA as a result of either 2 (or very rarely 3) copies of the SMN1 gene on one chromosome and the absence of the SMN1 gene on the other chromosome or small intragenic mutations within the SMN1 gene. This analysis also will not detect germline mosaicism or mutations in genes other than SMN1. Additionally, de novo mutations have been reported in approximately 2% of SMA patients.

Carrier Frequency and Risk Reductions for Individuals with No Family History of SMA				
Ethnicity	**Detection Rate[1]**	**A priori Carrier Risk[1]**	**Reduced Carrier Risk for 2 copy result**	**Reduced Carrier Risk for 3 copy result**
Caucasian	94.9%	1:35	1:632	1:3,500
Ashkenazi Jewish	90.2%	1:41	1:350	1:4,000
Asian	92.6%	1:53	1:628	1:5,000
Hispanic	90.6%	1:117	1:1061	1:11,000
African American	71.1%	1:66	1:121	1:3,000
Mixed Ethnicities	For counseling purposes, consider using the ethnic background with the most conservative risk estimates.			

METHOD/LIMITATIONS:
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. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

REFERENCES:
1. Carrier frequency and detection rate are calculated based on analysis of allele frequencies among > 1000 individuals from each ethnic group noted (Genzyme Genetics, data submitted for publication). 2. 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: Zhaoqing Zhou, Ph.D., FACMG, on 04/21/2010

Reported by: /

genzyme

Cystic Fibrosis Mutation Analysis

Patient Name: Donor, 2446
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

Cryogenic Laboratories, Inc. / Genetics and IVF Institute
[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 04/16/2010
Date Received: 04/19/2010
Lab ID: 2446100416
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 / LIMITATIONS:

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 fluorescence detection. Some mutations are then specifically identified by bi-directional dideoxysequencing. The assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

Under the direction of: Zhaoqing Zhou

Zhaoqing Zhou, Ph.D., FACMG

Date: 04/26/2010

SMAC

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