Donor 2993

Genetic Testing Summary

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

Last Updated: 04/27/21

Donor Reported Ancestry: German, Irish, Native American 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 97 mutations in the CFTR gene	1/343
Tay Sachs Enzyme Analysis	Non-Carrier by Hexosaminidase activity	
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 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.

genzyme

Cystic Fibrosis Mutation Analysis

Patient Name: Donor #2993, .
Referring P[redacted]
Specimen #:[redacted]
Patient ID:[redacted]

Client #: 606452
Case #: 61540005

Fairfax Cryobank /[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 09/13/2010
Date Received: 09/14/2010
Lab ID:
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: Ruth A Heim, PhD, FACMG

10/6/10

Date: 09/21/2010

Ruth A. Heim, Ph.D., FACMG

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

genzyme
GENETICS

MUTATIONS ANALYZED

ΔF311	3120+1G>A	712-1G>T	Q359K/T360K	S549N
ΔF508	3120G>A	935delA	Q493X	S549R T>G
ΔI507	3171delC	936delTA	Q552X	T338I
1078delT	3199del6	A455E	Q890X	V520F
1288insTA	3659delC	A559T	R1066C	W1089X
1677delTA	3667del4	C524X	R1158X	W1204X
1717-1G>A	3791delC	CFTRdele2,3	R1162X	W1282X
1812-1G>A	3849+10kbC>T	D1152H	R117C	Y1092X C>A
1898+1G>A	3876delA	E60X	R117H	Y1092X C>G
1898+5G>T	3905insT	E92X	R334W	Y122X
1949del84	394delTT	G178R	R347H	
2043delG	4016insT	G330X	R347P	
2055del9>A	405+1G>A	G480C	R352Q	
2105del13ins5	405+3A>C	G542X	R553X	
2108delA	406-1G>A	G551D	R560T	
2143delT	444delA	G85E	R709X	
2183delAA>G	457TAT>G	K710X	R75X	
2184delA	574delA	L206W	R764X	
2184insA	621+1G>T	M1101K	S1196X	
2307insA	663delT	N1303K	S1251N	
2789+5G>A	711+1G>T	P574H	S1255X	
2869insG	711+5G>A	Q1238X	S364P	

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.

genzyme

Tay-Sachs Enzyme Analysis

Patient Name: Donor #2993, .
Referring Physician: Steve Pool, M.D.
Specimen #: [redacted]
Patient ID: [redacted]

Client #: 606452

Fairfax Cryobank / [redacted]

DOB: Not Given
SSN:

Date Collected: 09/13/2010
Date Received: 09/14/2010
Lab ID:
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 1434 nmol/mg protein**
Hexosaminidase Percent A: 56.6

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥55%	≥55%
Expected Carrier Range:	Hex A	20 - 48%	20 - 49%

INTERPRETATION: NON CARRIER

This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

10/6/10

Under the direction of: Stanford Marenberg, PHD, ABCC

Stanford Marenberg, Ph.D.

Date: 09/22/2010

Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

genzyme

SMN1 Copy Number Analysis

Patient Name: . Donor #2993
DOB: Age:
SSN #: Gender: Male

606452 / 310544
Fairfax Cryobank / [redacted]

Genzyme Specimen #:61653936-6
Case #: [redacted] Patient ID #: 61367865
Date Collected: 09/13/2010 Date Received: 09/14/2010

Referring Physician: Steve Pool
Genetic Counselor:

Client Lab ID #:
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

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RECEIVED 10/6/10

Electronically Signed by: Hui Zhu, Ph.D. FACMG, on 09/21/2010

Reported by: /