



Donor 2979

Genetic Testing Summary

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

Last Updated: 06/25/24

Donor Reported Ancestry: English, German

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 99 mutations in the CFTR gene	1/343
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.

Patient Name: Donor #2979, .

Referring Physician: [REDACTED]

Specimen #: [REDACTED]

Patient ID: [REDACTED]

Client #: [REDACTED]

Case #: [REDACTED]

Fairfax Cryobank / [REDACTED]

DOB: Not Given

Sex: M

SSN: [REDACTED]

Date Collected: 10/29/2009

Date Received: 10/30/2009

Lab ID: [REDACTED]

Hospital ID: [REDACTED]

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/pch/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 $\Delta F508$ and the following polymorphisms: F508C, I506V and I507V. In some cases, specific allele identification requires enzymatic amplification followed by hybridization to oligonucleotide probes.

[REDACTED]
12.18.09
rn

Under the direction of:



Narasimhan Nagan, Ph.D., FACMG

Date: 11/06/2009

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

COPY

Patient Name: . Donor #2979

DOB:

Age:

SSN #:

Gender: Male

Genzyme Specimen #

Case #:

Patient ID #: 1

Date Collected: 10/29/2009

Date Received: 10/30/2009

Fairfax Cryobank /

Referring Physician:

Genetic Counselor:

Client Lab ID #:

Hospital ID #:

Specimen ID #:

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

Specimen Type: Peripheral Blood

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.

ENTERED
12.18.09

Electronically Signed by: Narasimhan Nagan, Ph.D., FACMG, on 11/04/2009

Reported by: 7

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Celebrating 25 Years of Excellence

Cytogenetic Report

Client **Fairfax Cryobank** [REDACTED]

Address [REDACTED]
[REDACTED]

Reporting Phone # [REDACTED] Fax # [REDACTED] Email **N/A**

Patient name/Donor Alias **Donor 2979**

Patient DOB **N/A**

Donor # **2979-091207**

Specimen type **Peripheral Blood**

Collection Date **12/07/2009**

Accession # **09-085CG**

Date Received **12/08/2009**

RESULTS

CYTOGENETIC ANALYSIS

Cells counted **20**

Type of banding **GTG**

Cells analyzed **5**

Band resolution **550**

Cells karyotyped **2**

Modal chromosome # **46**

FISH

Probe(s) **N/A**

Nuclei scored **N/A**

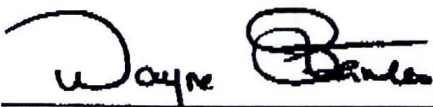
KARYOTYPE 46,XY

INTERPRETATION

Normal male karyotype.

No numerical or structural abnormalities were identified. This normal cytogenetic result does not exclude the possibility of the presence of subtle rearrangements beyond the technical limits of detection with this test.

Comments **The results shown here are from a redraw due to previous poor specimen quality.**

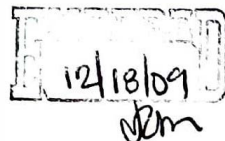


Wayne S. Stahley, Ph.D., FACMG
Clinical Cytogeneticist

12/16/09

Date

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12/18/09
JEM

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