Donor 2751

Genetic Testing Summary

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

Last Updated: 04/03/24

Donor Reported Ancestry: Colombian 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	1/205
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/1061

*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.

GENETICS & IVF
Institute

Celebrating 25 Years of Excellence

Cytogenetic Report

Client Fairfax Cryobank

Address [redacted]

Reporting Phone # [redacted] **Fax #** [redacted] **Email** [redacted]

Patient name/Donor Alias Donor 2751 **Patient DOB** N/A

Donor # 2751-[redacted] **Specimen type** Peripheral Blood

Collection Date 04/08/2010 **Accession #** [redacted]

Date Received 04/08/2010

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	20	**Type of banding**	GTG
Cells analyzed	5	**Band resolution**	500
Cells karyotyped	3		
Modal chromosome #	46		

KARYOTYPE 46,XY

FISH

Probe(s) N/A

Nuclei scored N/A

ENTERED 5-21-10 DM

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

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

4/15/10
Date

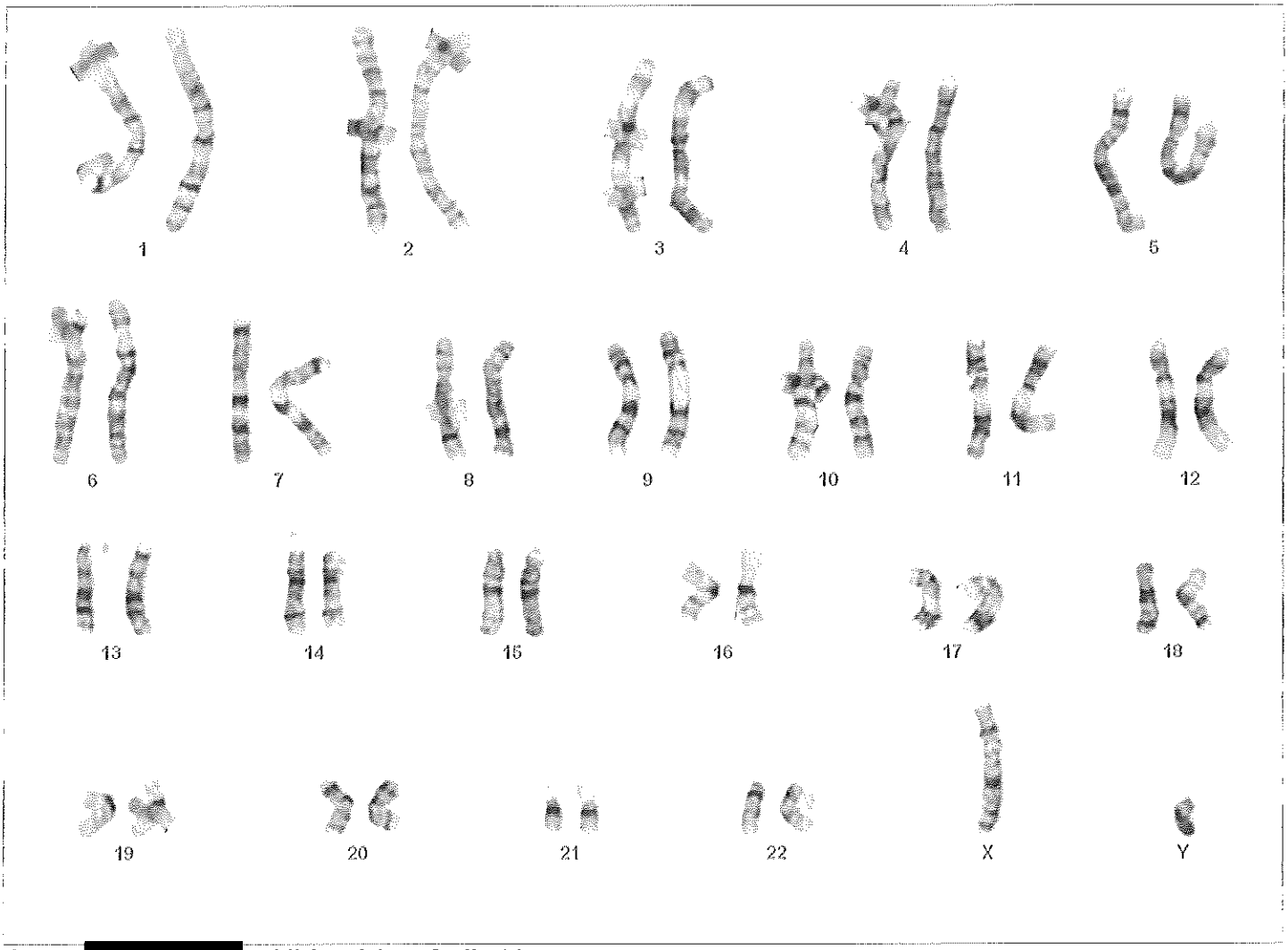
3015 Williams Drive • Fairfax, Virginia 22031 • (703) 698-7355 • (800) 552-4363 • E-mail: givf@givf.com • www.givf.com

Genetics and IVF Preimplantation Genetics Laboratory

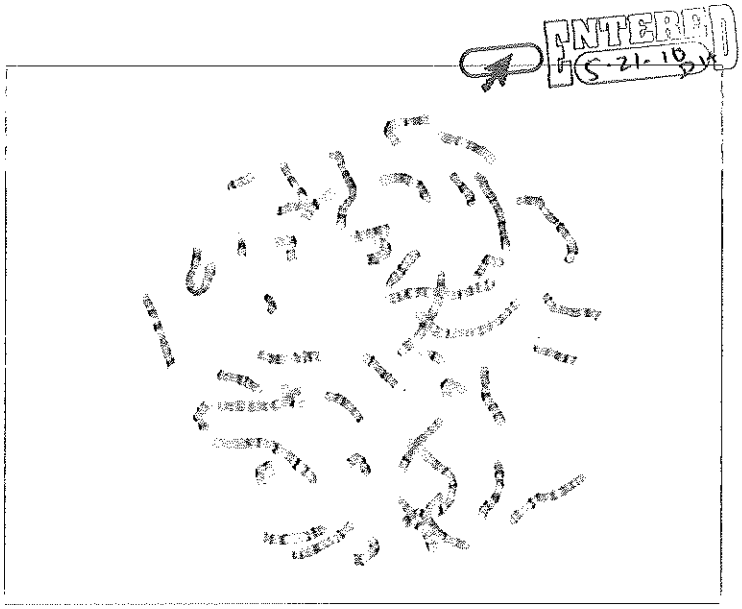
Patient name: DONOR 2751

Case name: [redacted]

46,XY



Case: [redacted] Slide: A3 Cell: 19



Quest Diagnostics
1901 Sulphur Spring Road • Baltimore, Maryland 21227-0588
Main Laboratory 410-247-0100 • D.C. Area 301-624-6900 • Outside Maryland 1-800-LAB-NOW

ROBERT R L SMITH, M.D.
Medical Director

No Tracking Number
SPECIMEN COLLECTED: 04/08/2010 11:00
COMPLETED REPORT: 04/09/2010 13:52

[redacted]

2751
PATIENT ID #: NP

PATIENT PHONE#: Not provided
PATIENT DOB: Not provided

PATIENT NAME	DATE	AGE	SEX	LAB NUMBER	
2751	04/08/2010	?	M	[redacted]	LAB REPORT

HEMATOLOGY:

WHITE BLOOD CELL COUNT	6.7	Thousand/uL	(3.8-10.8)
RED BLOOD CELL COUNT	5.04	Million/uL	(4.20-5.80)
HEMOGLOBIN	14.9	g/dL	(13.2-17.1)
HEMATOCRIT	44.2	%	(38.5-50.0)
MCV	88	fL	(80-100)
MCH	29.6	pg	(27-33)
MCHC	33.8	g/dL	(32-36)
RDW	13.6	%	(11.0-15.0)
PLATELET COUNT	169	Thousand/uL	(140-400)
MPV	9.6	fL	(7.5-11.5)
ABSOLUTE NEUTROPHILS	3712	cells/uL	(1500-7800)
ABSOLUTE LYMPHOCYTES	1970	cells/uL	(850-3900)
ABSOLUTE MONOCYTES	610	cells/uL	(200-950)
ABSOLUTE EOSINOPHILS	375	cells/uL	(15-500)
ABSOLUTE BASOPHILS	34	cells/uL	(0-200)
NEUTROPHILS	55.4	%	
LYMPHOCYTES	29.4	%	
REACTIVE LYMPHOCYTES	0.0	%	
MONOCYTES	9.1	%	
EOSINOPHILS	5.6	%	
BASOPHILS	0.5	%	

COMMENT:

ENTERED 5-21-10 DH

SIGNATURE

DATE REPORTED

The above laboratory studies were performed by Quest Diagnostics, 1901 Sulphur Spring Road, Baltimore, MD 21227

20020I (4/09)

Quest Diagnostics
1901 Sulphur Spring Road • Baltimore, Maryland 21227-0388
Main Laboratory 410-247-9100 • D.C. Area 301-624-8000 • Outside Maryland 1-800-LAB-NOW1

ROBERT R.L. SMITH, M.D.
Medical Director

No Tracking Number
SPECIMEN COLLECTED: 04/08/2010 11:00
COMPLETED REPORT: 04/09/2010 13:52

[redacted]

2751
PATIENT ID #: NP

PATIENT PHONE#: Not provided
PATIENT DOB: Not provided

PATIENT NAME	DATE	AGE	SEX	LAB NUMBER	LAB REPORT
2751	04/08/2010	?	M	[redacted]	

CONTINUATION OF REPORT - PAGE 2

CHEMISTRY:

AST	21	U/L	(10-35)	
ALT	33	U/L	(9-60)	
*CHOLESTEROL	207	MG/DL	(125-200)	OK
HEMOGLOBIN A1	97.7 %	(>96.0)		
HEMOGLOBIN F	NONE DETECTED		(0.0-1.9)	
HEMOGLOBIN A2	2.3 %		(1.8-3.5)	
HGB SCREEN INTERPRETATION				

THE HEMOGLOBINOPATHY SCREEN IS NORMAL.

ABNORMAL HEMOGLOBIN #1 %: 0.0 %

THE ABOVE NORMAL VALUE RANGE(S) MAY NOT APPLY, SINCE AGE AND/OR SEX WERE NOT PROVIDED ON THE REQUISITION.

* * * PHYSICIAN NOTES * * *

We are unable to forward results as you requested due to incomplete mailing information. If you would still like a copy of these results forwarded to another party, please contact the Client Services Department at 410-247-0001 to provide the necessary information.

FAXED 5-21-10 DH

SIGNATURE

DATE REPORTED

The above laboratory studies were performed by Quest Diagnostics, 1901 Sulphur Spring Road, Baltimore MD 21227

200201 (4/92)

genzyme

Cystic Fibrosis Mutation Analysis

Patient Name: 2751, [redacted]
Referring Physician: [redacted]
Specimen #: [redacted]
Patient ID: [redacted]
Client #: [redacted]
Case #: [redacted]

Fairfax Cryobank / Genetics and IVF Institute
[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 04/08/2010
Date Received: 04/09/2010
Lab ID: 2751100408
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Hispanic

Indication: Carrier test / Gamete donor

RESULTS: Negative for the 97 mutations analyzed

INTERPRETATION

This individual's risk to be a carrier is reduced from 1/46 (2.2%) to 1/205 (0.5%), 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.

ENTERED 4-21-10

Under the direction of: Zhaoqing Zhou

Date: 04/19/2010

SMAC

Zhaoqing Zhou, Ph.D., FACMG

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

genzyme

SMN1 Copy Number Analysis

Patient Name: . 2751
DOB: Age:
SSN #: Gender: Male

[redacted]
Fairfax Cryobank / Genetics and IVF Institute
[redacted]

Genzyme Specimen # [redacted]
Case #: [redacted] Patient ID #: [redacted]
Date Collected: 04/08/2010 Date Received: [redacted]

Referring Physician: [redacted]
Genetic Counselor:

Client Lab ID #: 2751100408
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: Hispanic

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 5-21-10 DH

Electronically Signed by: Narasimhan Nagan, Ph.D., FACMG, on 04/13/2010
Reported by: /

Testing performed at Genzyme Genetics 3400 Computer Drive, Westborough, MA 01581 1-800-255-7357