Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4189

Genetic Testing Summary

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

Last Updated: 10/27/23

Donor Reported Ancestry: Bohemian, German, Polish, French, Irish, Norwegian 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
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Tay Sachs Enzyme Analysis	Non-carrier by hexosaminidase A analysis	

*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

Carrier Testing

Patient Name: Donor #4189, .
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

[redacted]stitute

DOB: Not Given
Sex: M
SSN:

Date Collected: 05/27/2010
Date Received: 05/28/2010
Lab ID: [redacted]
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Caucasian

Indication: Gamete donor

Disease	Result	Interpretation
Cystic Fibrosis	Negative	Carrier risk reduced from 1/25 (4%) to 1/343 (0.3%).
Tay-Sachs - Enzyme	Hex. Activity: 1103 nmol/mg protein Hex. Percent A: 62.1	Non carrier Plasma/Serum WBC Non carrier range: Hex A >= 55% >= 55% Carrier range : Hex A 20 - 48% 20 - 49%

COMMENTS:

DNA:
If ordered, results for dihydrolipoamide dehydrogenase deficiency, familial hyperinsulinism, nemaline myopathy, Usher syndrome type IF and Usher syndrome type III will be received separately.

The negative results from this analysis cannot eliminate the possibility that this individual carries a mutation not detected by this test. Unless otherwise noted, interpretations are based on a negative family history and the absence of symptoms.

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.

Enzyme: [White Blood Cells]
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.

METHOD / LIMITATIONS:

DNA is isolated from the sample and amplified for disease specific regions using the polymerase chain reaction (PCR). Mutations are identified by hybridization to allele specific oligonucleotides or by solution-phase multiplex allele-specific primer extension with subsequent mutation-specific hybridization and detection. 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.

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.

(REPORT CONTINUED ...)

Date: 06/10/2010

genzyme

Carrier Testing

Patient Name: Donor #4189, . **Referring Physician:** [redacted] **Specimen #:** [redacted] **Patient ID:** [redacted]

...Continued From Page 1

MUTATIONS ANALYZED / DETECTION RATE

Cystic Fibrosis

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

This 97 mutation assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V.

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

Under the direction of:

Ruth A Heim, PhD, FACMG

Ruth A. Heim, Ph.D., FACMG

Additional approvals by:

Enzyme:Stanford Marenberg, Ph.D.

SMAC

Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

genzyme

SMN1 Copy Number Analysis

Patient Name: . Donor #4189
DOB: Age:
SSN #: Gender: Male

606452 / 348795
[redacted]

Genzyme Specimen # [redacted]
Case #: [redacted] Patient ID #: [redacted]
Date Collected: 05/27/2010 Date Received: 05/28/2010

Referring Physician: [redacted]
Genetic Counselor:

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

Specimen Type: Peripheral Blood

Specimen(s) Received: 3 - 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.

ENTERED CMS 6142

Electronically Signed by: Lynne S. Rosenblum, Ph.D., FACMG, on 06/02/2010

Reported by:

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

GENETICS & IVF
Institute

Cytogenetic Report

Client [redacted]

Address [redacted]

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

Patient name/Donor Alias Donor # 4189 **Patient DOB** N/A

Donor # 4189-[redacted] **Specimen type** Peripheral blood

Collection Date 05/27/2010 **Accession #** [redacted]

Date Received 05/28/2010

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	20	**Type of banding**	GTG
Cells analyzed	5	**Band resolution**	500
Cells karyotyped	2		
Modal chromosome #	46		

KARYOTYPE 46,XY

FISH

Probe(s) N/A

Nuclei scored N/A

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

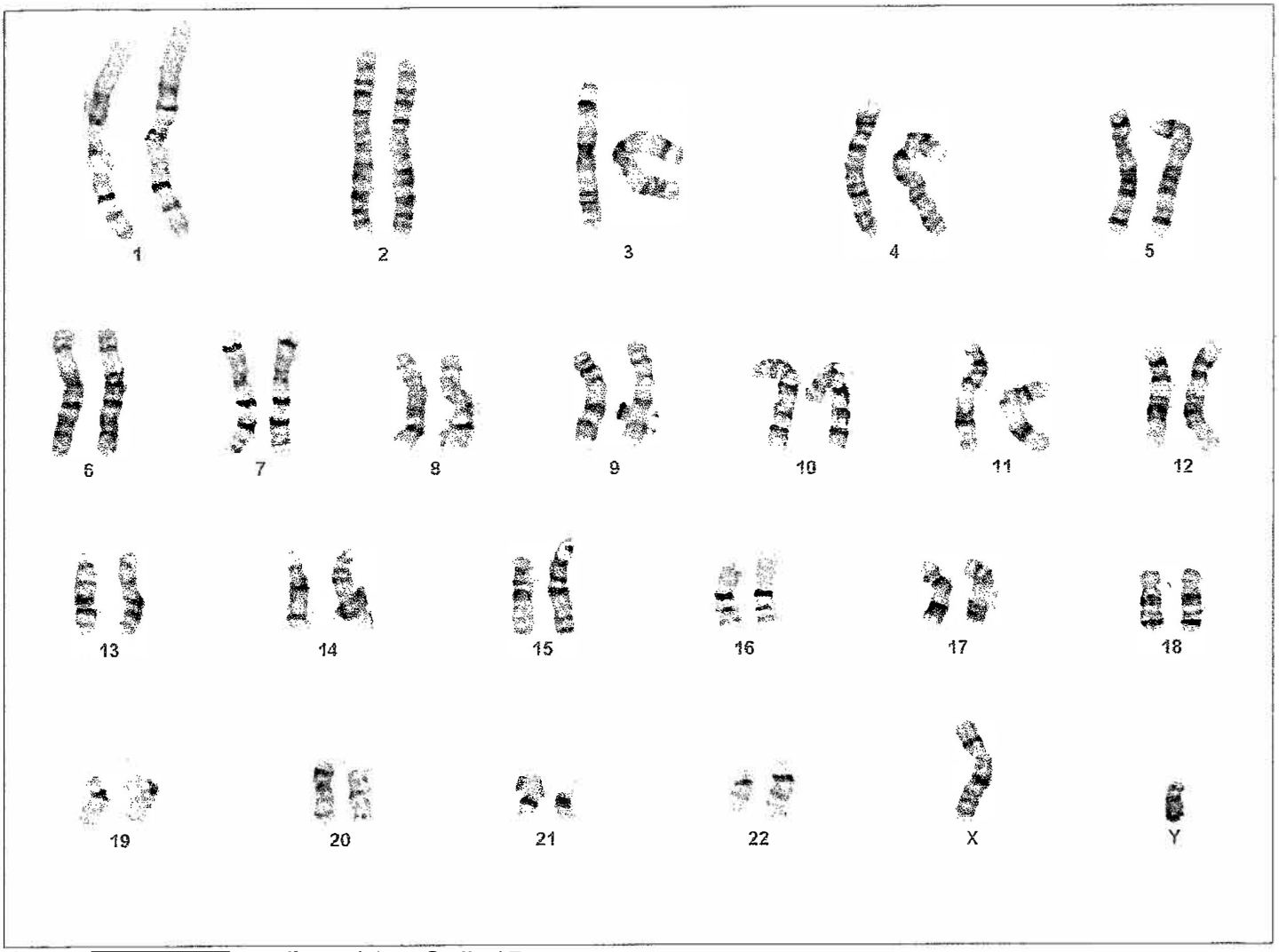
6/3/10
Date

Genetics and IVF Preimplantation Genetics Laboratory

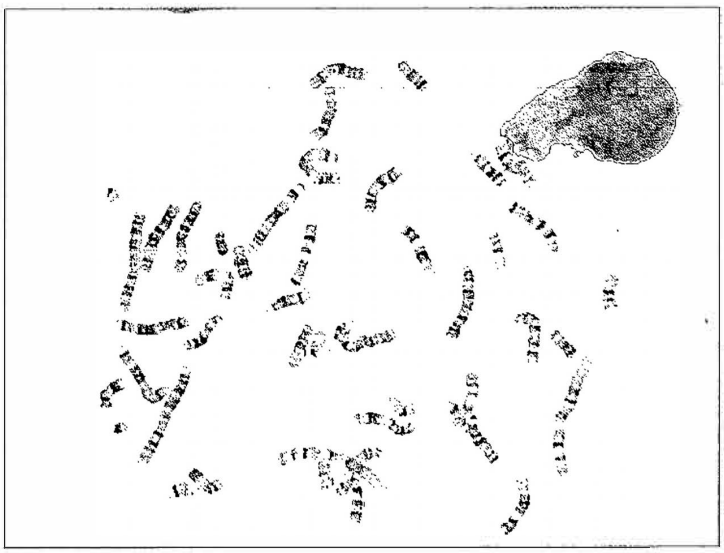
Patient name: DONOR # 4189

Case name: [redacted]

46,XY



Case: [redacted] Slide: A2 Cell: 17



Quest Diagnostics®

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 800.323.5917

PATIENT INFORMATION
DONOR,4189

DOB: AGE:
GENDER: M FASTING: N

REPORT STATUS **FINAL**

ORDERING PHYSICIAN
[redacted]

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION: [redacted]

ID:
PHONE:

CLIENT INFORMATION
[redacted] 4195000

COLLECTED: 05/27/2010 14:00 CT
RECEIVED: 05/28/2010 02:05 CT
REPORTED: 06/01/2010 07:29 CT

COMMENTS: REG 4189-100527

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.57		4.20-5.80 Million/uL	CB
HEMOGLOBIN	14.1		13.2-17.1 g/dL	
HEMATOCRIT	40.5		38.5-50.0 %	
MCV	88.6		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
RDW	12.7		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.			
CHOLESTEROL, TOTAL	153		125-200 mg/dL	CB
AST		47 H	10-35 U/L	CB
ALT	35		9-60 U/L	CB
CBC (INCLUDES DIFF/PLT)				CB
WHITE BLOOD CELL COUNT	5.3		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.57		4.20-5.80 Million/uL	
HEMOGLOBIN	14.1		13.2-17.1 g/dL	
HEMATOCRIT	40.5		38.5-50.0 %	
MCV	88.6		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
MCHC	34.9		32.0-36.0 g/dL	
RDW	12.7		11.0-15.0 %	
PLATELET COUNT	191		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	2995		1500-7800 cells/uL	

6/1/10 OK

ENTERED IMS 6.14.0

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PATIENT INFORMATION
DONOR,4189

REPORT STATUS **FINAL**

ORDERING PHYSICIAN [redacted]

DOB: AGE:
GENDER: M FASTING: N

COLLECTED: 05/27/2010 14:00 CT
REPORTED: 06/01/2010 07:29 CT

Test Name	In Range	Out of Range	Reference Range	Lab
ABSOLUTE LYMPHOCYTES	1860		850-3900 cells/uL	
ABSOLUTE MONOCYTES	286		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	127		15-500 cells/uL	
ABSOLUTE BASOPHILS	32		0-200 cells/uL	
NEUTROPHILS	56.5		%	
LYMPHOCYTES	35.1		%	
MONOCYTES	5.4		%	
EOSINOPHILS	2.4		%	
BASOPHILS	0.6		%	
ABO GROUP AND RH TYPE				CB
ABO GROUP	O ✓			
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, MD
CLIA: 14D0417052

A duplicate report has been faxed to the following:
Faxed to: (651) 489-0340 on: 05/31/2010 5:14:20 AM

ENTERED IMS 6.4.10