Donor 2829

Genetic Testing Summary

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

Last Updated: 08/28/24

Donor Reported Ancestry: German, Swedish, English 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

*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 Fib ɔsis Mutation Analysis

Patient Name: Donor, 2829 [redacted]
Referring Physician: Steve
Specimen
Patient ID: [redacted]

Client #: [redacted]
Case #: [redacted]

[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 07/12/2007
Date Received: 07/13/2007
Lab ID: 2829-070712
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

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 ΔF508 and the following polymorphisms: F508C, I506V and I507V. In some cases, specific allele identification requires enzymatic amplification followed by hybridization to oligonucleotide probes.

ENTERED 7/27/07 maf

Under the direction of: Zhaoqing Zhou

Zhaoqing Zhou, Ph.D.

Date: 07/23/2007

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

genzyme

Chromosome Analysis

Patient Name: Donor, 2829
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

Fairfax Cryobank [redacted]

DOB: Not Given
SSN:

Date Collected: 07/26/2007
Date Received: 07/27/2007
Lab ID: 2829-[redacted]
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	550
Metaphases Karyotyped:	2			**Dept. Section:**	B1

RESULTS: 46,XY
Male karyotype

INTERPRETATION:

This analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. The standard cytogenetic methodology utilized in this analysis does not routinely detect small rearrangements and low level mosaicism, and cannot detect microdeletions.

ENTERED 8-6-07

Signed: Suehan Berend, Ph.D., FACMG

Sue Berend

Date: 08/03/2007

Testing Performed At Genzyme Genetics 10421 University Center Drive Suite 100 Tampa, FL 33612 1-800-966-4440

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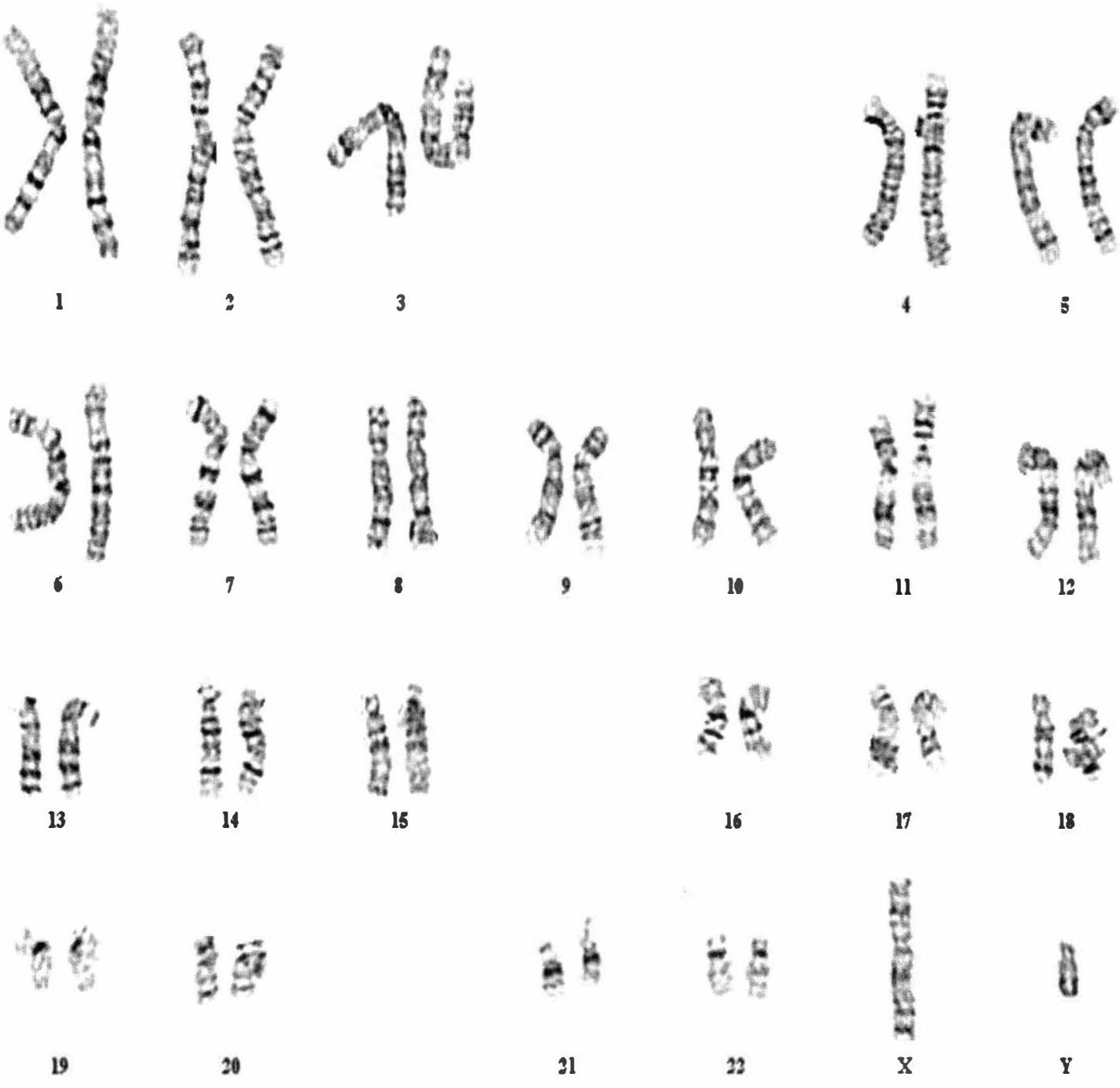

[redacted]
Patient Name: Donor, 2829
Image ID: DKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 07/27/2007
Date Reviewed: 08/03/2007
Reviewed By: SB2

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GENERAL
genetics

Quest Diagnostics

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 800.825.7330

PATIENT INFORMATION
ID,2829

DOB: AGE:
GENDER: M FASTING: N

REPORT STATUS **FINAL**

ORDERING PHYSICIAN
[redacted]

SPECIMEN INFORMAT[redacted]
SPECIMEN: [redacted]
REQUISITION:

ID:
PHONE:

[redacted]MATION
[redacted]

COLLECTED: 07/12/2007 08:00 ET
RECEIVED: 07/12/2007 21:32 ET
REPORTED: 07/17/2007 05:59 ET

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.40		4.20-5.80 Million/uL	QHO
HEMOGLOBIN	13.7		13.2-17.1 g/dL	
HEMATOCRIT	39.0		38.5-50.0 %	
MCV	88.7		80.0-100.0 fL	
MCH	31.2		27.0-33.0 pg	
RDW	12.1		11.0-15.0 %	
HEMOGLOBIN A1	97.4		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.6		1.8-3.5 %	
INTERPRETATION	NORMAL PHENOTYPE.			

[redacted]

ENTERED 7/17/07