Donor 2997

Genetic Testing Summary

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

Last Updated: 1/3/23

Donor Reported Ancestry: Spanish, Dutch

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 for 99 mutations in the CFTR gene	1/310
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7n the SMN1 gene	1/700
HBB testing	Negative for 28 mutations in the HBB gene	Beta Thalassemia: 1/1500 Sickle Cell Disease: <1/500

*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.

Counsyl

Results F[redacted]'bient
Fairfax Cryobank [redacted]
Report Date: 10/31/2011

Male
Name: DONOR 2997
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Sample Type: OG-500 Saliva
Date of Collection: 10/18/2011
Barcode: [redacted]
Indication: Egg or Sperm Donor

Female
Not tested

Counsyl Test Results (Egg or Sperm Donor)

The Counsyl test uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for a number of Mendelian diseases. This report indicates which mutations, if any, were detected for each mutation panel. Because only select mutations are tested, the percentage of carriers detected varies by ethnicity. A negative test result does not eliminate the possibility that the individual is a carrier. Interpretation is given as an estimate of the risk of conceiving a child affected with a disease, which is based on reported ethnicity, the test results, and an assumption of no family history.*

DONOR 2997

DONOR 2997's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Reproductive Risk Summary

No increased reproductive risks to highlight. Please refer to the following pages for detailed information about the results.

Clinical notes:

- Individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and may also benefit from carrier testing by CBC and hemoglobin electrophoresis or HPLC. *ACOG Practice Bulletin No. 78. Obstet Gynecol 2007;109:229-37.*

To schedule a free appointment to speak with a genetic counselor about your results, please visit www.counsyl.com/appointment.

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*Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, and technical errors. The reproductive risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Counsyl

Male
Name: DONOR 2997
DOB [redacted]

Female
Not tested

Full Results

Below are the full test results for all diseases on the panel. Noted are the specific genetic mutations for which the patient tested positive or negative. If there was insufficient data to determine the genotype for any variant, this will be noted as "no call." Also listed in this section is the patient's post-test risk of being a carrier of each disease as well as the odds that his future children could inherit each disease.

Beta Thalassemia

Reproductive risk: Less than 1 in 1,000,000
Risk before testing: 1 in 250,000
Reduced risk

DONOR 2997: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 1,500. 83% detection rate.

Gene: HBB. Variants (27): K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab.

Cystic Fibrosis

Reproductive risk: 1 in 34,000
Risk before testing: 1 in 3,000
Reduced risk

DONOR 2997: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 310. 91% detection rate.

Gene: CFTR. Variants (99): G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, E92X, Y122X, G178R, R347H, Q493X, V520F, S549N, P574H, M1101K, D1152H, 2143delT, 394delTT, 444delA, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), R117C, L206W, G330X, T338I, R352Q, S364P, G480C, C524X, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, R764X, Q890X, R1066C, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, 3199del6, 574delA, 663delT, 935delA, 936delTA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667del4, 3791delC, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, 3849+4A>G, Q359K/T360K.

Sickle Cell Disease

Reproductive risk: Less than 1 in 1,000,000
Risk before testing: less than 1 in 1,000,000
Reduced risk

DONOR 2997: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 70% detection rate.

Gene: HBB. Variants (28): Hb S, K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab.

Spinal Muscular Atrophy

Reproductive risk: 1 in 97,000
Risk before testing: 1 in 4,800
Reduced risk

DONOR 2997: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 700. 95% detection rate.

Gene: SMN1. Variants (1): Exon 7 deletion.

Genetics

Chromosome Analysis

atient Name: Donor, 2997
eferring Physician:
pecimen #: [redacted]
atient ID: [redacted]

Client #: [redacted]

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

OB: Not Given
SN:

Date Collected: 10/18/2011
Date Received: 10/20/2011
Lab ID:
Hospital ID:
Specimen Type: **Peripheral Blood**

dication: Gamete donor

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

RESULTS: 46,XY
Male karyotype

ITERPRETATION:

his analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. he standard cytogenetic methodology utilized in this analysis does not routinely detect subtle rearrangements low-level mosaicism and cannot detect microdeletions. Also, it cannot detect molecular cytogenetic normalities (such as microdeletions and microduplications) that may be detectable by array comparative enomic hybridization (aCGH).

gned: Veena Suri

Veena Suri, Ph.D.

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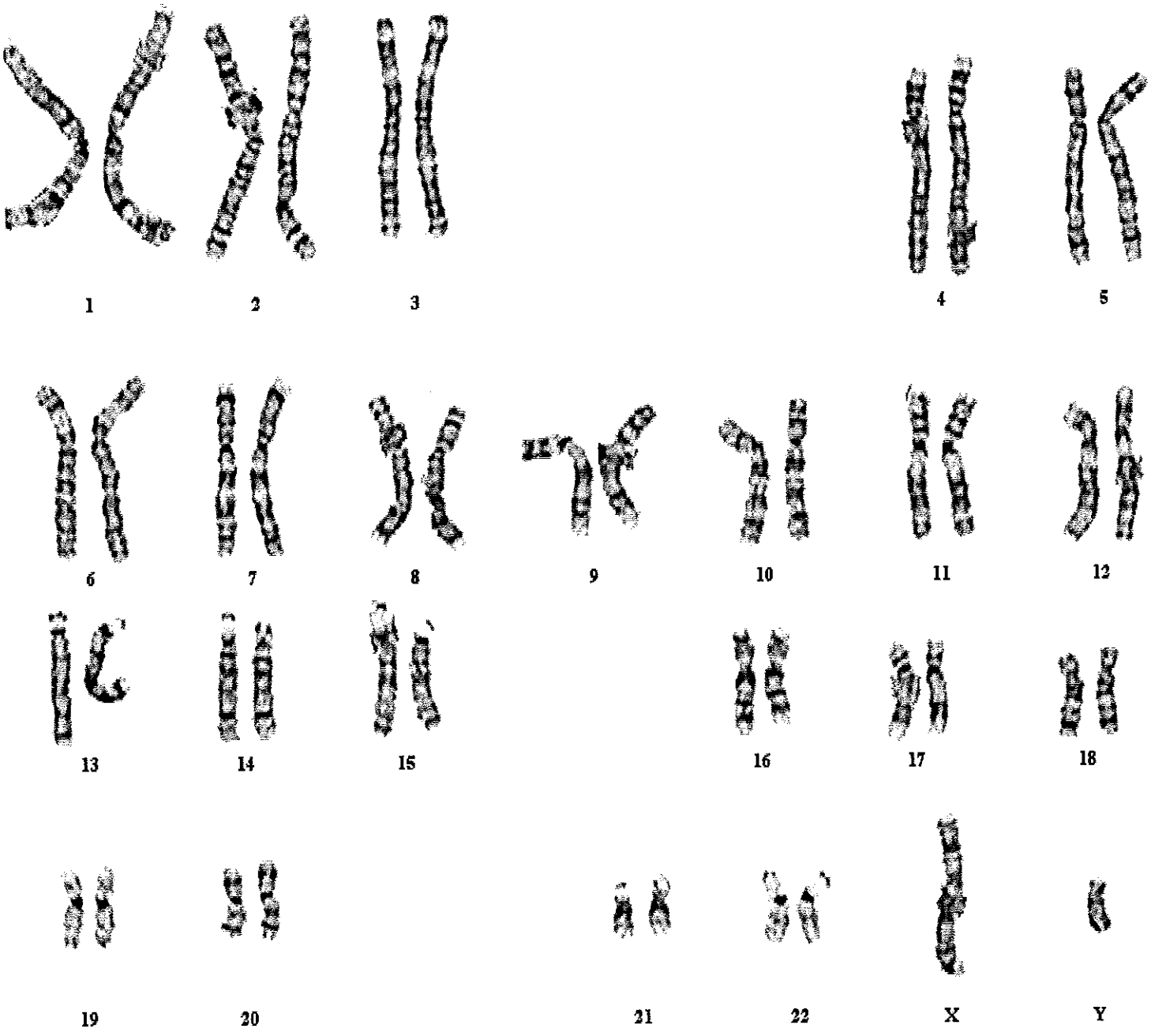
Date: 10/26/2011

Genetics



Specimen #[redacted]
Specimen Type: BLDPER
Patient Name: Donor, 2997
Image ID: BKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 10/20/2011
Date Reviewed: 10/26/2011
Reviewed By: VSU

Quest Diagnostics

Report Status: Final

DONOR, 2997

Patient Information	Specimen Information	Client Information
DONOR, 2997 **DOB:** [redacted] **AGE:** [redacted] Gender: M Fasting: U Phone: NG Patient ID: [redacted]	Specimen: [redacted] Requisition: 0000032 Collected: 10/18/2011 Received: 10/19/2011 / 06:59 CDT Reported: 10/24/2011 / 10:10 CDT	Client #: [redacted] FAIRFAX CRYOBANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.10		4.20-5.80 Million/uL	IG
HEMOGLOBIN	15.8		13.2-17.1 g/dL	
HEMATOCRIT	47.1		38.5-50.0 %	
MCV	92.4		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
RDW	13.4		11.0-15.0 %	
HEMOGLOBIN A	98.1		>96.0 %	IG
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	1.9		1.8-3.5 %	
INTERPRETATION				
Normal phenotype.				
CHOLESTEROL, TOTAL	200		125-200 mg/dL	IG
AST	14		10-40 U/L	IG
ALT	18		9-60 U/L	IG
CBC (INCLUDES DIFF/PLT)				IG
WHITE BLOOD CELL COUNT	6.4		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	5.10		4.20-5.80 Million/uL	
HEMOGLOBIN	15.8		13.2-17.1 g/dL	
HEMATOCRIT	47.1		38.5-50.0 %	
MCV	92.4		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
MCHC	33.5		32.0-36.0 g/dL	
RDW	13.4		11.0-15.0 %	
PLATELET COUNT	263		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	4429		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1523		850-3900 cells/uL	
ABSOLUTE MONOCYTES	339		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	83		15-500 cells/uL	
ABSOLUTE BASOPHILS	26		0-200 cells/uL	
NEUTROPHILS	69.2		%	
LYMPHOCYTES	23.8		%	
MONOCYTES	5.3		%	
EOSINOPHILS	1.3		%	
BASOPHILS	0.4		%	
ABO GROUP AND RH TYPE				IG
ABO GROUP	A			
RH TYPE	RH(D) POSITIVE			

PERFORMING SITE:

IG QUEST DIAGNOSTICS-IRVING, 4770 REGENT BLVD., IRVING, TX 75063 Laboratory Director: ELISABETH S BROCKIE,D.O., CLIA: 45D0697943

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CLIENT SERVICES: 800.824.6152 COLLECTED: 10/18/2011 PAGE 1 OF 1