Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4409

Genetic Testing Summary

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

Last Updated: 08/20/18

Donor Reported Ancestry: Chinese 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 108 mutations in the CFTR gene	1/200
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	<1/500
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 37 mutations tested in the HBB gene	1/160 for Beta-Thalassemia <1/500 for Sickle Cell

*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 Recipient	Ordering Healthcare Professional	Male Details	Female Details
Fairfax Cryobank - [redacted] [redacted] Report Date: 04/07/2011	Fairfax Cryobank - [redacted]	Name: Donor 4409 DOB: [redacted] Ethnicity: East Asian Sample Type: Saliva (OG-300) Date of Collection: 03/24/2011 Barcode: [redacted] Indication: Egg or Sperm Donor	Not tested

Universal Genetic Test (Egg or Sperm Donor)

The Universal Genetic 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 4409

Donor 4409's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

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

Child Risk Summary

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

Note on hemoglobinopathies:
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.

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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 child risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Counsyl

Male	Female
Name: Donor 4409 DOB: [redacted]	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

Your child's risk:	Risk before testing:	
1 in 20,000	1 in 3,900	Reduced risk

Donor 4409: 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 160. 80% detection rate.

Gene: HBB. **Variants (35):** K17X, Q39X, 619 bp deletion, 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), IVS-I-1(G>T), -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, Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, Pro5fs, Gly16fs, Glu6fs, IVS-II-705, IVS-II-844, -30T>A, CAP+1 A>C, Hb E, Hb O-Arab.

Cystic Fibrosis

Your child's risk:	Risk before testing:	
1 in 69,000	1 in 30,000	Reduced risk

Donor 4409: No mutations detected. No call for 3199del6. 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 200. 56% detection rate.

Gene: CFTR. **Variants (108):** 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, S1235R, 394delTT, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), G91R, R117C, I148T, L206W, G330X, T338I, R352Q, S364P, G480C, I506V, F508C, C524X, S549I, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, Q890X, R1066C, R1070Q, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, R1283M, dele2-3 21kb, 3199del6, F311del, 574delA, 663delT, 935delA, 936delTA, 1161delC, 1609delCA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2105-2117del13insAGAAA, 3171delC, 3667del4, 3821delT, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1811+1.6kbA>G, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, W1204X(c.3612G>A).

Sickle Cell Disease

Your child's risk:	Risk before testing:	
Less than 1 in 1,000,000	less than 1 in 1,000,000	Reduced risk

Donor 4409: 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. >99% detection rate.

Gene: HBB. **Variants (37):** Hb S, K17X, Q39X, 619 bp deletion, 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), IVS-I-1(G>T), -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, Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, Pro5fs, Gly16fs, Glu6fs, IVS-II-705, IVS-II-844, -30T>A, CAP+1 A>C, Hb E, Hb D-Punjab, Hb O-Arab.

Spinal Muscular Atrophy

Your child's risk:	Risk before testing:	
1 in 150,000	1 in 11,000	Reduced risk

Donor 4409: 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. 93% detection rate.

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

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This test was developed and its performance characteristics determined by Counsyl, Inc. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician's workup.

Laboratory Director: Jessica Jacobson, MD
CLIA Number: 05D1102604

2200 Bridge Parkway, Suite 103, Redwood City, CA 94065
(888) COUNSYL | http://www.counsyl.com

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank [redacted]

Address [redacted]

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

Patient name/Donor Alias Donor # 4409 **Patient DOB** N/A

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

Collection Date 03/24/2011 **Accession #** [redacted]

Date Received 03/25/2011

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

IMSG·19·11

4/5/11
Date

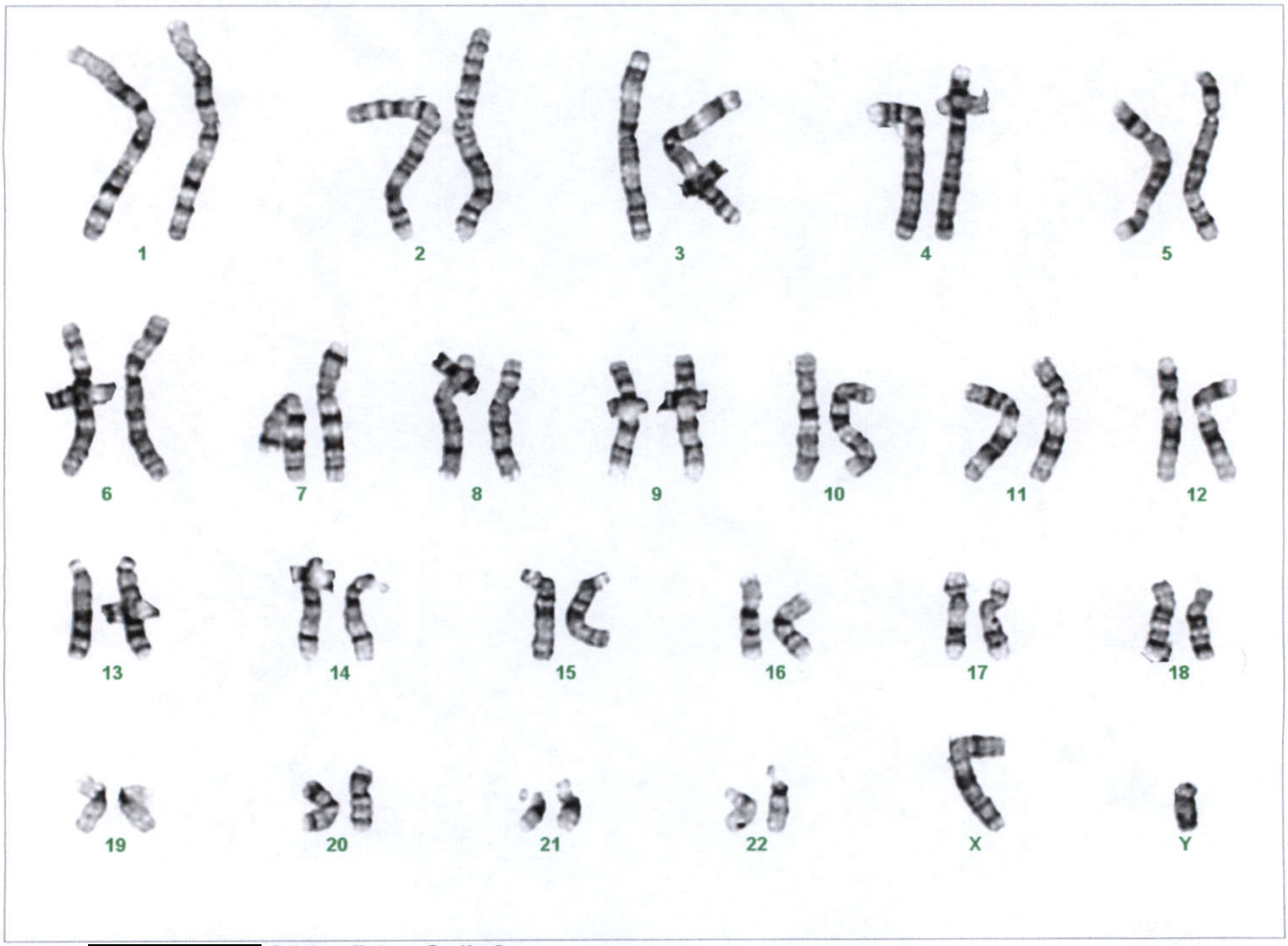
3015 Williams Drive | Fairfax, Virginia 22031 | 703-698-7355 | 800-552-4363 | givf@givf.com | www.givf.com

Genetics and IVF Preimplantation Genetics Laboratory

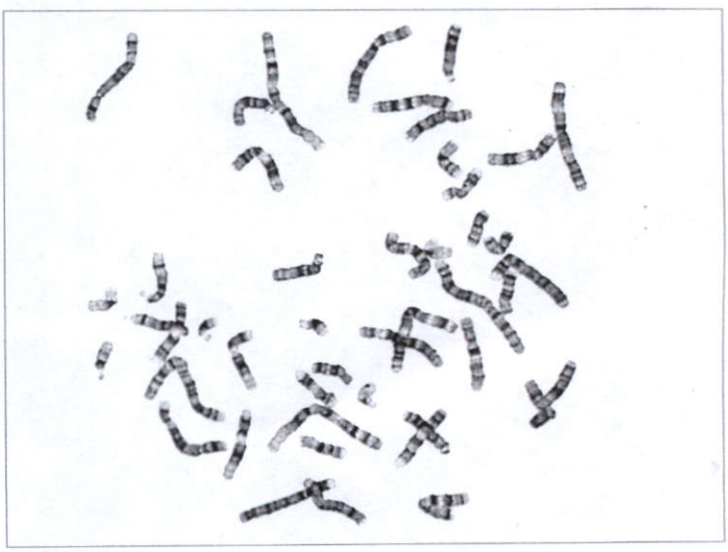
Patient name: DONOR 4409

Case name: [redacted]

46,XY



Case: [redacted] Slide: B1 Cell: 6



Quest Diagnostics®

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 866.697.8378

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION:

PATIENT INFORMATION
ID,4409

DOB: AGE:
GENDER: M FASTING: U

ID:
PHONE:

REPORT STATUS FINAL

ORDERING PHYSICIAN
STERN,HARVEY J

CLIENT INFORMATION
[redacted] H013
FAIRFAX CRYO BANK
[redacted]

COLLECTED: 03/30/2011 07:30 ET
RECEIVED: 03/30/2011 23:37 ET
REPORTED: 04/04/2011 15:10 ET

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.14		4.20-5.80 Million/uL	QHO
HEMOGLOBIN	15.9		13.2-17.1 g/dL	
HEMATOCRIT	47.1		38.5-50.0 %	
MCV	91.5		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
RDW	12.6		11.0-15.0 %	
HEMOGLOBIN A	97.6		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.4		1.8-3.5 %	
INTERPRETATION	Normal phenotype.			
CBC (INCLUDES DIFF/PLT)				QHO
WHITE BLOOD CELL COUNT	6.2		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	5.14		4.20-5.80 Million/uL	
HEMOGLOBIN	15.9		13.2-17.1 g/dL	
HEMATOCRIT	47.1		38.5-50.0 %	
MCV	91.5		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
MCHC	33.8		32.0-36.0 g/dL	
RDW	12.6		11.0-15.0 %	
PLATELET COUNT	221		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	3410		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	2480		850-3900 cells/uL	
ABSOLUTE MONOCYTES	248		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	62		15-500 cells/uL	
ABSOLUTE BASOPHILS	0		0-200 cells/uL	
NEUTROPHILS	55		%	
LYMPHOCYTES	40		%	
MONOCYTES	4		%	
EOSINOPHILS	1		%	
BASOPHILS	0		%	

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COLLECTED: 03/30/2011 07:30 ET
REPORTED: 04/04/2011 15:10 ET

PATIENT INFORMATION
ID,4409

DOB: AGE:
GENDER: M FASTING: U

REPORT STATUS FINAL

ORDERING PHYSICIAN
STERN,HARVEY J

PERFORMING LABORATORY INFORMATION
QHO QUEST DIAGNOSTICS-HORSHAM, 900 BUSINESS CENTER DRIVE, HORSHAM, PA 19044-3408
Laboratory Director: HERMAN HURWITZ, MD, FCAP, CLIA: 39D0204404

LIST OF RESULTS PRINTED IN THE OUT OF RANGE COLUMN: