Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4401

Genetic Testing Summary

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

Last Updated: 08/29/18

Donor Reported Ancestry: Irish, Italian 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	1/270
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	<1/500 for Beta-Thalassemia <1/500 for Sickle Cell
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	

*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: 01/28/2011	Fairfax Cryobank - [redacted]	Name: Donor 4401 DOB: [redacted]2 Ethnicity: Mixed or Other Caucasian Sample Type: Saliva (OG-300) Date of Collection: 01/17/2011 Barcode: [redacted] Indication: No family history (screening)	Not tested

Universal Genetic Test

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 4401

Donor 4401'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

Your Universal Genetic Test indicates that your future children have a reduced risk for the diseases tested, including those listed below which are common in your ethnicity.

Cystic Fibrosis

Spinal Muscular Atrophy

ENTERED IMS 29.11

*Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. The child risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation. Individuals of African, Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and should also be offered carrier testing by CBC and hemoglobin electrophoresis or HPLC.

Counsyl

Male	Female
Name: Donor 4401 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: Less than 1 in 1,000,000
Risk before testing: 1 in 250,000
Reduced risk

Donor 4401: 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. 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: 1 in 30,000
Risk before testing: 1 in 3,100
Reduced risk

Donor 4401: 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 270. 90% 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: Less than 1 in 1,000,000
Risk before testing: less than 1 in 1,000,000
Reduced risk

Donor 4401: 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: 1 in 97,000
Risk before testing: 1 in 4,800
Reduced risk

Donor 4401: 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. 95% detection rate.

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

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

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

Quest Diagnostics®

QUEST DIAGNOSTICS INCORPORATED

COLLECTED: 01/17/2011 12:30 ET
REPORTED: 01/26/2011 15:03 ET

PATIENT INFORMATION
ID,4401[redacted]
DOB: AGE:
GENDER: FASTING: U

REPORT STATUS FAX COPY

ORDERING PHYSICIAN
[redacted]

Test Name	In Range	Out of Range	Reference Range	Lab

KARYOTYPE:
46,XY

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INTERPRETATION and COMMENTS:
NORMAL MALE karyotype

Within the limits of standard cytogenetic methodologies, the chromosomes had normal G-banding patterns without apparent structural abnormality or rearrangement.

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods, or rare events such as low level mosaicism or very subtle rearrangements.

Electronic Signature on File

Nicole C. Christacos, Ph.D.

Technical Director
Diplomate, ABMG
Cytogenetics Laboratory
703-802-7156

RESULTS RECEIVED 01/26/11
Reference lab accession: [redacted]

[redacted] QHO

[redacted]

[redacted] QHO

[redacted]

Quest Diagnostics®

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 866.697.8378

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

COLLECTED: 02/03/2011
RECEIVED: 02/04/2011 03:11 ET
REPORTED: 02/07/2011 15:03 ET

PATIENT INFORMATION
ID,4401

DOB: AGE:
GENDER: M FASTING: Y

ID: 4401-[redacted]
PHONE:

REPORT STATUS FAX COPY

ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
[redacted] HO13

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.69		4.20-5.80 Million/uL	QHO
HEMOGLOBIN	15.4		13.2-17.1 g/dL	
HEMATOCRIT	45.2		38.5-50.0 %	
MCV	96.3		80.0-100.0 fL	
MCH	32.9		27.0-33.0 pg	
RDW	13.0		11.0-15.0 %	
HEMOGLOBIN A	96.6		>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	4.8		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.69		4.20-5.80 Million/uL	
HEMOGLOBIN	15.4		13.2-17.1 g/dL	
HEMATOCRIT	45.2		38.5-50.0 %	
MCV	96.3		80.0-100.0 fL	
MCH	32.9		27.0-33.0 pg	
MCHC	34.2		32.0-36.0 g/dL	
RDW	13.0		11.0-15.0 %	
PLATELET COUNT	208		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	2736		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1584		850-3900 cells/uL	
ABSOLUTE MONOCYTES	432		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	96		15-500 cells/uL	
ABSOLUTE BASOPHILS	0		0-200 cells/uL	
NEUTROPHILS	57		%	
LYMPHOCYTES	33		%	
MONOCYTES	9		%	
EOSINOPHILS	2		%	
BASOPHILS	0		%	

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genzyme

Tay-Sachs Enzyme Analysis

Patient Name: 4401, .
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]1

DOB: Not Given
SSN:

Date Collected: 01/17/2011
Date Received: 01/18/2011
Lab ID: 4401-110117
Hospital ID:
Specimen Type: **White Blood Cells**

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[redacted]
[redacted]

RESULTS: **Hexosaminidase Activity : 1464 nmol/mg protein**
Hexosaminidase Percent A: 60.3

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥55%	≥55%
Expected Carrier Range:	Hex A	20 - 48%	20 - 49%

INTERPRETATION: NON CARRIER

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.

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Under the direction of: Stanford Marenberg, PHD, ABCC

Stanford Marenberg, Ph.D.

Date: 01/21/2011

Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436