Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4202

Genetic Testing Summary

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

Last Updated: 08/23/18

Donor Reported Ancestry: Korean, French, Polish 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: 12/09/2010	Fairfax Cryobank - [redacted]	Name: Donor 4202 DOB: [redacted] Ethnicity: East Asian Sample Type: Saliva (OG-300) Date of Collection: 11/29/2010 [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 4202

Donor 4202'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.

Beta Thalassemia

Cystic Fibrosis

Spinal Muscular Atrophy

12/17/10

*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.

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

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Counsyl

Male
Name: Donor 4202
[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

Your child's risk: 1 in 20,000

Risk before testing: 1 in 3,900

Reduced risk

Donor 4202: 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: 1 in 69,000

Risk before testing: 1 in 30,000

Reduced risk

Donor 4202: No mutations detected. No call for R1066C. 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: Less than 1 in 1,000,000

Risk before testing: less than 1 in 1,000,000

Reduced risk

Donor 4202: 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 150,000

Risk before testing: 1 in 11,000

Reduced risk

Donor 4202: 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.

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

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GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

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

Patient name/Donor Alias Donor #4202 **Patient DOB** N/A

Donor # 4202-101129 **Specimen type** Peripheral Blood

Collection Date 11/29/2010 **Accession #** 10-113CG

Date Received 11/30/2010

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	20	**Type of banding**	GTG
Cells analyzed	5	**Band resolution**	550
Cells karyotyped	2		
Modal chromosome #	46		

FISH

Probe(s) N/A

Nuclei scored N/A

KARYOTYPE 46,XY

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

12/16/10
Date

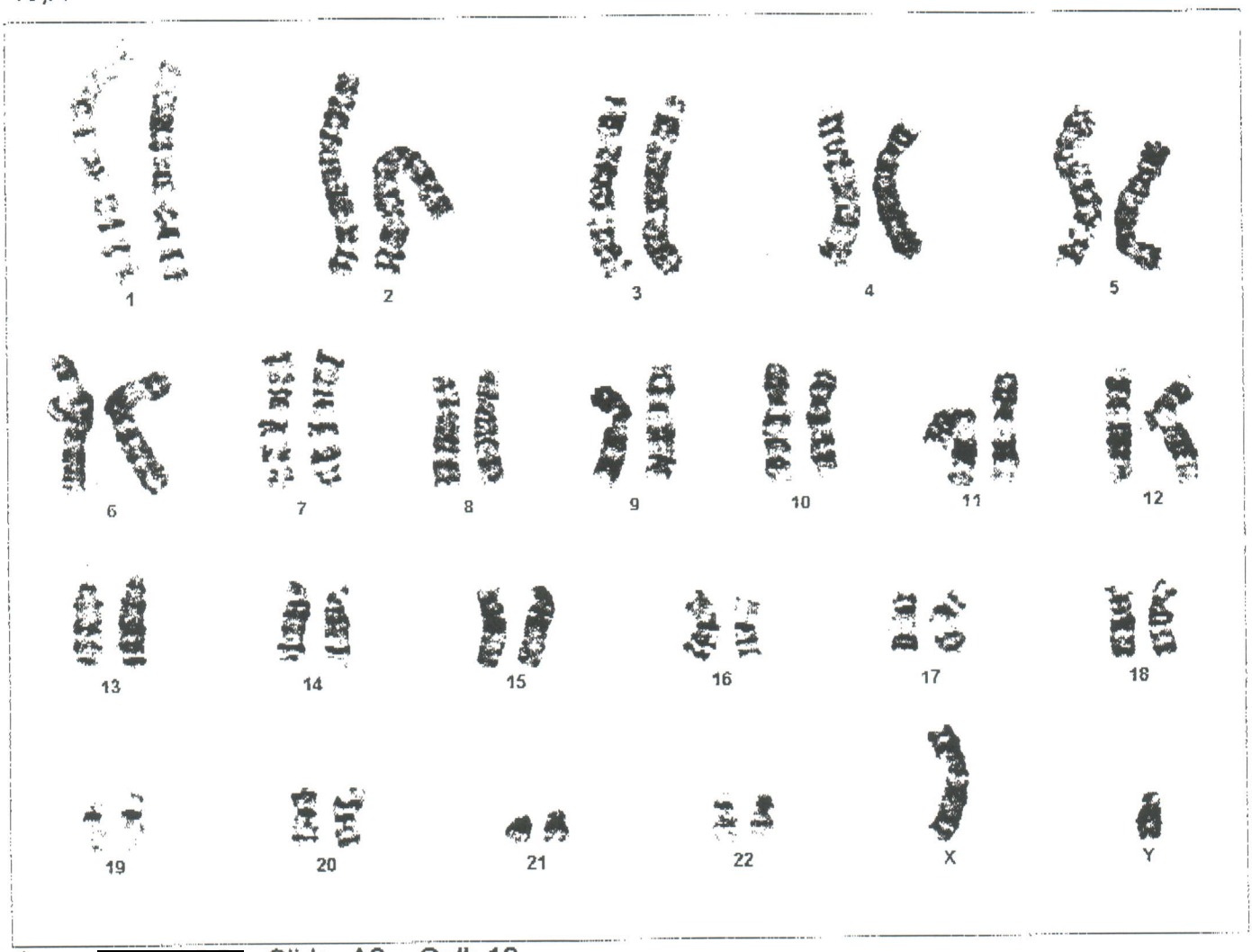
12/17/10

Genetics and [redacted]F Preimplantation Genetics Laboratory

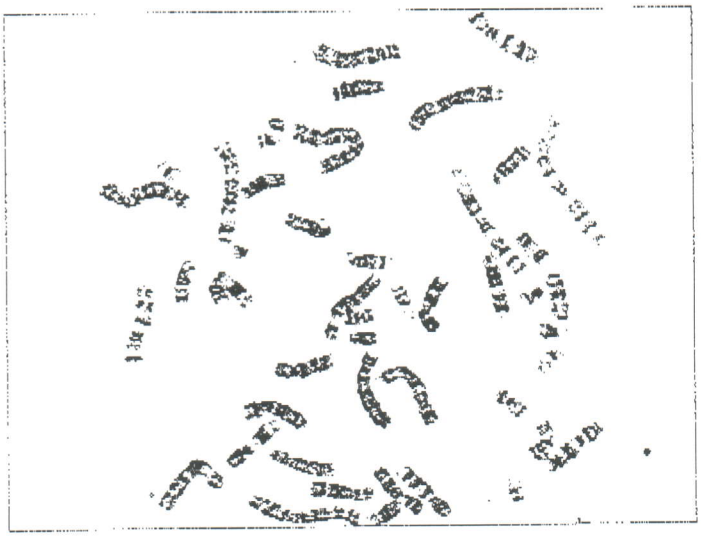
Patient name: Donor # 4202

Case name: [redacted]

46,XY



Case: [redacted] Slide: A2 Cell: 18



Quest Diagnostics

Report Status: Final

DONOR, 4202

Patient Information	Specimen Information	Client Information
DONOR, 4202 **DOB: Not Given** **AGE: Not Given** Gender: M Fasting: U Phone: NG Patient ID: [redacted]	Specimen: [redacted] Requisition: [redacted] Collected: 11/29/2010 Received: 11/30/2010 / 03:13 CST Reported: 12/02/2010 / 13:39 CST	Client #: 41550 AUS0000 [redacted] FAIRFAX CRYOBANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.09		4.20-5.80 Million/uL	IG
HEMOGLOBIN	16.3		13.2-17.1 g/dL	
HEMATOCRIT	48.3		38.5-50.0 %	
MCV	94.9		80.0-100.0 fL	
MCH	32.0		27.0-33.0 pg	
RDW	13.7		11.0-15.0 %	
HEMOGLOBIN A	97.9		>96.0 %	IG
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.1		1.8-3.5 %	
INTERPRETATION				
Normal phenotype.				
CHOLESTEROL, TOTAL	169		125-200 mg/dL	IG
AST	20		10-35 U/L	IG
ALT	22		9-60 U/L	IG
CBC (INCLUDES DIFF/PLT)				IG
WHITE BLOOD CELL COUNT	4.4		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	5.09		4.20-5.80 Million/uL	
HEMOGLOBIN	16.3		13.2-17.1 g/dL	
HEMATOCRIT	48.3		38.5-50.0 %	
MCV	94.9		80.0-100.0 fL	
MCH	32.0		27.0-33.0 pg	
MCHC	33.7		32.0-36.0 g/dL	
RDW	13.7		11.0-15.0 %	
PLATELET COUNT	181		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	2702		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1153		850-3900 cells/uL	
ABSOLUTE MONOCYTES	352		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	172		15-500 cells/uL	
ABSOLUTE BASOPHILS	22		0-200 cells/uL	
NEUTROPHILS	61.4		%	
LYMPHOCYTES	26.2		%	
MONOCYTES	8.0		%	
EOSINOPHILS	3.9		%	
BASOPHILS	0.5		%	
ABO GROUP AND RH TYPE				IG
ABO GROUP	B			
RH TYPE	RH(D) POSITIVE			

PERFORMING SITE:

IG QUEST DIAGNOSTICS-IRVING, 4770 REGENT BLVD., IRVING, TX 75063 Laboratory Director: SUZANNE H. KREISBERG, MD, CLIA: 45D0697943

RECEIVED 12/17/10

CLIENT SERVICES: 800.824.6152

COLLECTED: 11/29/2010

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