Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4341

Genetic Testing Summary

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

Last Updated: 01/14/22

Donor Reported Ancestry: German, English, Irish, Scottish 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 99 mutations in the CFTR gene	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/610
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/290
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	Male	Female
Fairfax Cryobank - Fairfax Attn: Dr. Harvey Stern [redacted] NPI: [redacted] Report Date: 03/28/2013	Name: DONOR 4341 DOB: [redacted] Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 03/25/2013 Date Received: 03/27/2013 Barcode: [redacted] Indication: Egg or Sperm Donor	Not tested

Counsyl Test Results (Egg or Sperm Donor)

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for **128 variants** associated with **3 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 full list of mutations tested is given on page 2. 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 4341

DONOR 4341'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:

- The Counsyl test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional testing and genetic counseling.
- 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.*
- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complimentary appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

Lab Directors:

William Seltzer, PhD, FACMG

H. Peter Kang, MD

*Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately. 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. For the purposes of risk calculations, it is assumed that mutations within the same gene are on different chromosomes.

Counsyl

Male | Female

Name: DONOR 4341
DOB: [redacted]

Not tested

Mutations Tested

Cystic Fibrosis - 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. **Detection rate:** Northern European 91%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - 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. **Detection rate:** Northern European 83%.

Spinal Muscular Atrophy - Gene: SMN1. **Variants (1):** SMN1 copy number. **Detection rate:** Northern European 95%.

Counsyl

Male	Female
Name: DONOR 4341 DOB [redacted]	Not tested

Risk Calculations

Below are the full test results for all diseases on the panel. 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. A negative result does not rule out the possibility of being a carrier of untested mutations. Estimates of post-test carrier risk assume a negative family history.

Disease	DONOR 4341 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000	1 in 3,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000	1 in 10,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000	1 in 4,800

Integrated GENETICS

Tay-Sachs Enzyme Analysis

Patient Name: Donor #4341
Referring Physician: [redacted]
Specimen # [redacted]
Patient ID: [redacted]
Client # [redacted]

DOB: Not Given
SSN: ***-**-****

Date Collected: 03/25/2013
Date Received: 03/27/2013
Lab ID:
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 988 nmol/mg protein**
Hexosaminidase Percent A: 74.2

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥54%	≥54%
Expected Carrier Range:	Hex A	20 - 49%	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.

COPY

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Under the direction of: [redacted]

Stanford Marenberg, PHD, MDCC
Stanford Marenberg, Ph.D.

Date: 03/29/2013

Testing Performed At Esoterix Genetic Laboratories, LLC 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

VRL Laboratories
6665 S. Kenton Street, Ste 205, Centennial, CO 80111
Phone 855-VRL-LABS, Fax 303-799-1584

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* FINAL *
* *
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Requesting Facility: FAIRFAX CRYOBANK [redacted]

VRL Accession Nbr: [redacted]
Date Received: 03/26/2013 09:30
Date Of Final Report: 03/27/2013 11:26
Date Report Generated: 03/27/2013 11:26
Gender: MALE Date Of Birth: [redacted]

Donor ID-1: 4341
Donor ID-2: [redacted]
Donor ID-3:
Donor ID-4:

Tube Type	Collection Date/Time	Refrigeration Date/Time	Centrifugation Date/Time	Transfusion Status	Sample Type
RED	03/25/2013 15:00				LIVING
EDTA	03/25/2013 15:00				LIVING

TEST REQUESTED	RESULTS	REFERENCE RANGE
**** CBC		
WBC	7.0	3.8-10.8 THOUS/MCL
RBC	4.70	4.20-5.80 MILL/MCL
HEMOGLOBIN	13.6	13.4-18.0 GM/DL
HEMATOCRIT	42.1	40.0-54.0 %
MCV	89.7	80.0-100.0 FL
MCH	28.9	27.0-33.0 PG
MCHC	32.3	32.0-36.0 GM/DL
RDW	14.6	11.0-15.0 %
PLATELET COUNT	142	140-400 THOUS/MCL
MPV	11.2	7.5-11.5 FL
ABSOLUTE NEUTROPHILS	3430	1500-7800 CELLS/MCL
ABSOLUTE LYMPHOCYTES	2828	850-3900 CELLS/MCL
ABSOLUTE MONOCYTES	595	200-950 CELLS/MCL
ABSOLUTE EOSINOPHILS	112	0-500 CELLS/MCL
ABSOLUTE BASOPHILS	35	0-200 CELLS/MCL
NEUTROPHILS	49.0	%
LYMPHOCYTES	40.4	%
MONOCYTES	8.5	%
EOSINOPHILS	1.6	%
BASOPHILS	0.5	%

GENETICS & IVF
Institute

Cytogenetic Report

ENTERED 4.12.13

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted] Fax # [redacted] Email [redacted]

Patient name/Donor Alias Donor # 4341 Patient DOB N/A

Donor # [redacted] Specimen type Peripheral Blood

Collection Date 03/25/2013 Accession # [redacted]

Date Received 03/25/2013

RESULTS

CYTOGENETIC ANALYSIS

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

KARYOTYPE 46,XY

FISH

Probe(s) N/A

Nuclei scored N/A

INTERPRETATION

Normal male karyotype

No clonal 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

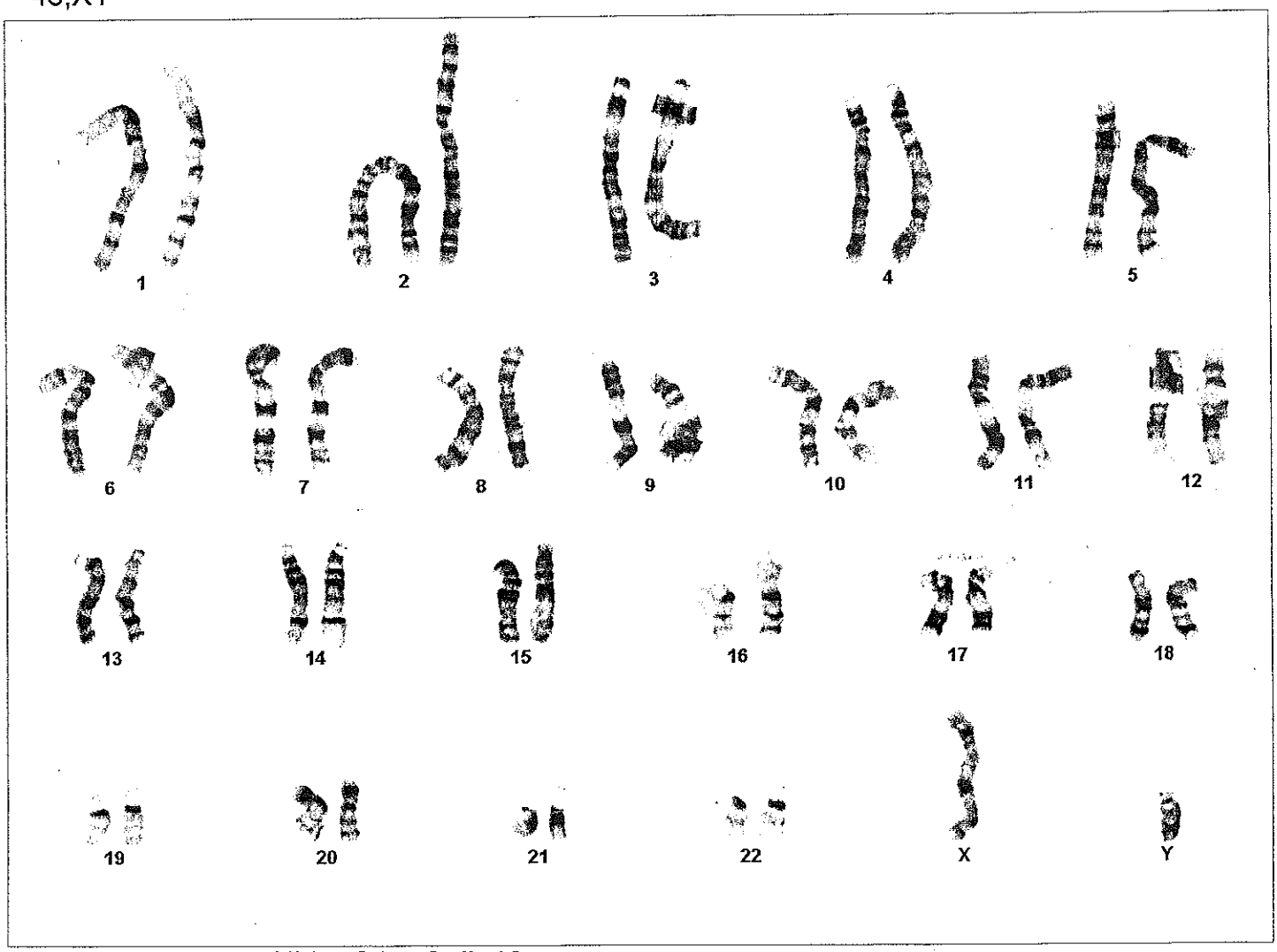
4/9/13
Date

Genetics and IVF Preimplantation Genetics Laboratory

Patient name: DONOR #4341

Case name [redacted]

46,XY



Case: 13-040CG Slide: C1 Cell: 12