Donor 4352

Genetic Testing Summary

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

Last Updated: 03/13/23

Donor Reported Ancestry: German, English, Dutch, Native American 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 carrier screening	Negative for 99 variants 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	Negative for 28 variants in the HBB gene	1/290

*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 - [redacted] Attn: Dr. Harvey Stern [redacted] NPI: [redacted] Report Date: 07/11/2013	Name: DONOR # 4352 DOB: [redacted] Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 07/08/2013 Date Received: 07/10/2013 Barcode: [redacted] Indication: Egg or Sperm Donor	Not tested

Counsyl Test Results Summary (Egg or Sperm Donor)

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses copy number analysis and targeted genotyping as described in the methods section on page 2 to determine carrier status associated with **3 diseases**. Please refer to page 3 for a complete list of diseases and genes included in this panel.

DONOR # 4352

DONOR # 4352'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

- 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/.

ENTERED 7.26.13

Counsyl

Male

Name: DONOR # 4352
DOB: [redacted]

Female

Not tested

Methods and Limitations

DONOR # 4352: targeted genotyping and copy number analysis.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 127 variants associated with 2 diseases. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

Copy number analysis: Targeted copy number analysis is used to determine the copy number of exon 7 of the SMN1 gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of SMN1 are carriers with two SMN1 genes on one chromosome and a SMN1 deletion on the other chromosome. In addition, a small percentage of SMA cases are caused by nondeletion mutations in the SMN1 gene. Thus, a test result of two SMN1 copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more SMN1 gene copies. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

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, bone marrow transplantation, blood transfusions and technical errors. 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 evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet Gynecol 2007;109:229-37*).

This test was developed and its performance characteristics determined by Counsyl, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's workup. Literature citations validating reported variants are available upon request. CLIA Number: **#05D1102604.**

Lab Director:

Hyunseok Kang

H. Peter Kang, MD

Counsyl

Male	Female
Name: DONOR # 4352 DOB [redacted]	Not tested

Diseases 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 (copy number analysis only) - Gene: SMN1. **Variant (1):** SMN1 copy number. **Detection rate:** Northern European 95%.

Counsyl

Male	Female
Name: DONOR # 4352 DOB [redacted]	Not tested

Risk Calculations

Below are the risk calculations for all diseases tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Disease	DONOR # 4352 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000

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

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Requesting Facility: FAIRFAX CRYOBANK
A GENETICS & IVF INSTITUTE CRYOBANK
3015 WILLIAMS DR., STE. 110
FAIRFAX, VA 22031

VRL Accession Nbr: [redacted]
Date Received: 07/09/2013 09:40
Date Of Final Report: 07/11/2013 07:40
Date Report Generated: 07/11/2013 07:40
Gender: MALE Date Of Birth: UNKNOWN

Donor ID-1: 4352
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	07/08/2013 15:00				LIVING
EDTA	07/08/2013 15:00				LIVING

TEST REQUESTED	RESULTS	REFERENCE RANGE
**** CBC		
WBC	5.3	3.8-10.8 THOUS/MCL
RBC	4.76	4.20-5.80 MILL/MCL
HEMOGLOBIN	15.1	13.4-18.0 GM/DL
HEMATOCRIT	46.0	40.0-54.0 %
MCV	96.5	80.0-100.0 FL
MCH	31.8	27.0-33.0 PG
MCHC	33.0	32.0-36.0 GM/DL
RDW	14.1	11.0-15.0 %
PLATELET COUNT	166	140-400 THOUS/MCL
MPV	9.9	7.5-11.5 FL
ABSOLUTE NEUTROPHILS	3265	1500-7800 CELLS/MCL
ABSOLUTE LYMPHOCYTES	1585	850-3900 CELLS/MCL
ABSOLUTE MONOCYTES	175	200-950 CELLS/MCL
ABSOLUTE EOSINOPHILS	223	0-500 CELLS/MCL
ABSOLUTE BASOPHILS	53	0-200 CELLS/MCL
NEUTROPHILS	61.6	%
LYMPHOCYTES	29.9	%
MONOCYTES	3.3	%
EOSINOPHILS	4.2	%
BASOPHILS	1.0	%

CLIA [redacted]
FDA [redacted]

Laboratory Director: Zahra Mehdizadeh Kashi, PhD, HCLD
Dr. Michael J Bauer, MD (NY)

GENETICS & IVF
Institute

ENTERED 7-26-13

Cytogenetic Report

Client Fairfax Cryobank [redacted]

Address [redacted]

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

Patient name/Donor Alias Donor # 4352 **Patient DOB** N/A

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

Collection Date 07/08/2013 **Accession #** [redacted]

Date Received 07/09/2013

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

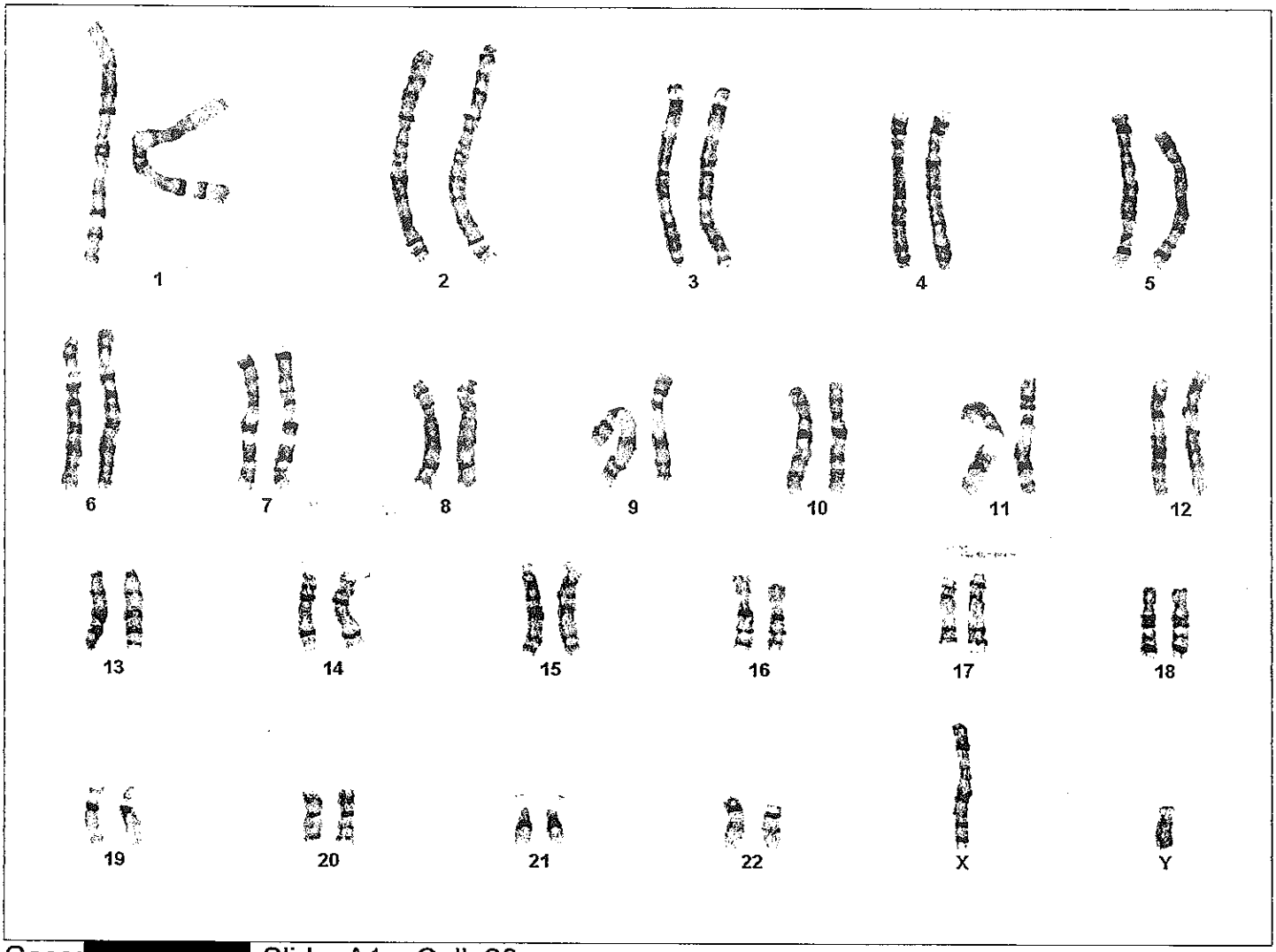
7/23/13
Date

Genetics and IVF Preimplantation Genetics Laboratory

Patient name: DONOR # 4352

Case name: [redacted]

46,XY



Case: [redacted] Slide: A1 Cell: 20