Donor 4552

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: Norwegian, Polish, English, Scottish, Irish 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	1/310
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/700
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/1500 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.

GENETICS & IVF
Institute

ENTERED 6/21/12

Cytogenetic Report

Client	[redacted]
Address	
Reporting Phone #	

Patient name/Donor Alias	Donor # 4552	**Patient DOB**	N/A
Donor #	4552-120604	**Specimen type**	Peripheral Blood
Collection Date	06/04/2012	**Accession #**	12-066CG
Date Received	06/05/2012		

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

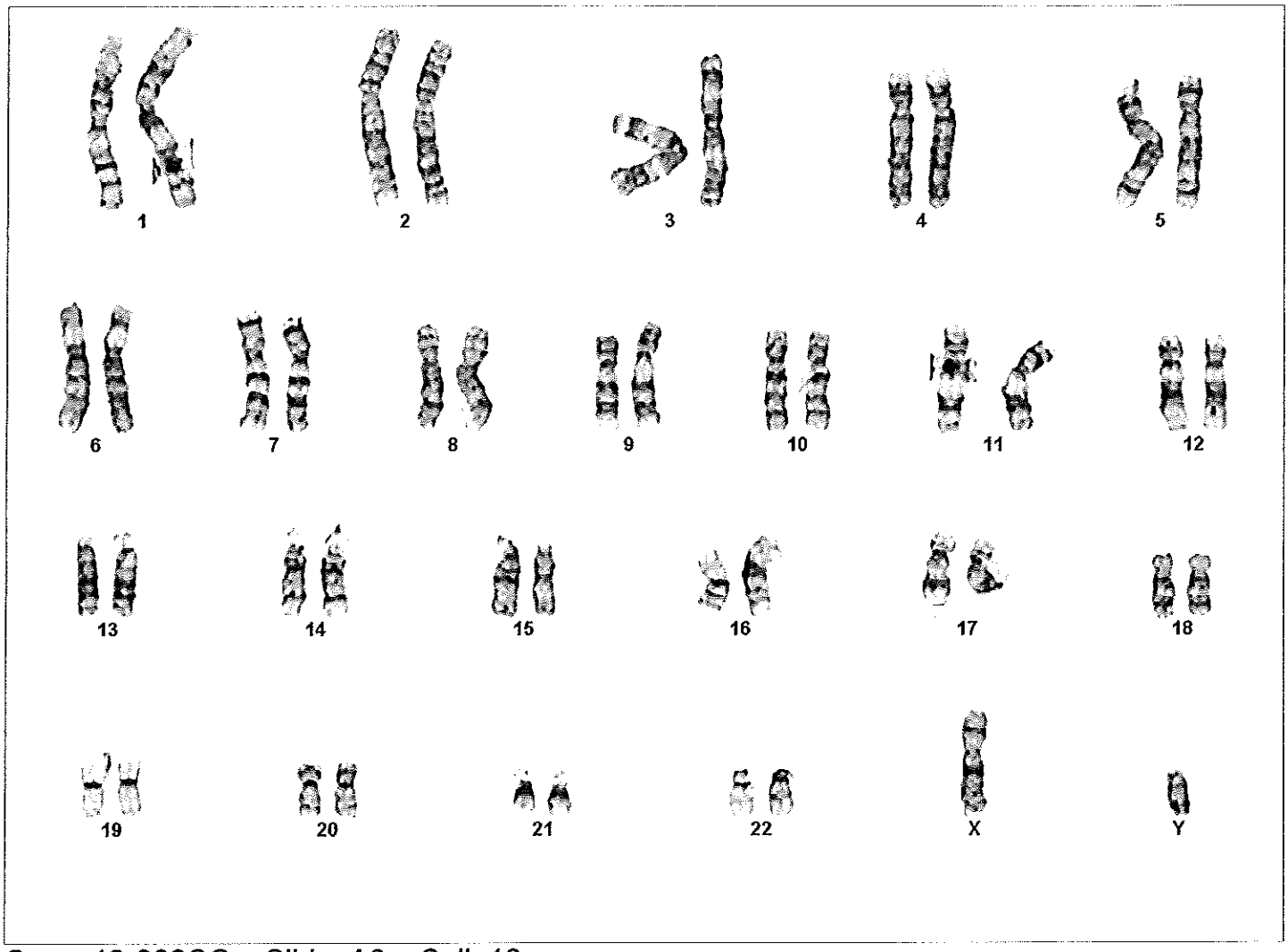
6/19/12
Date

Genetics & IVF Preimplantation Genetics Laboratory

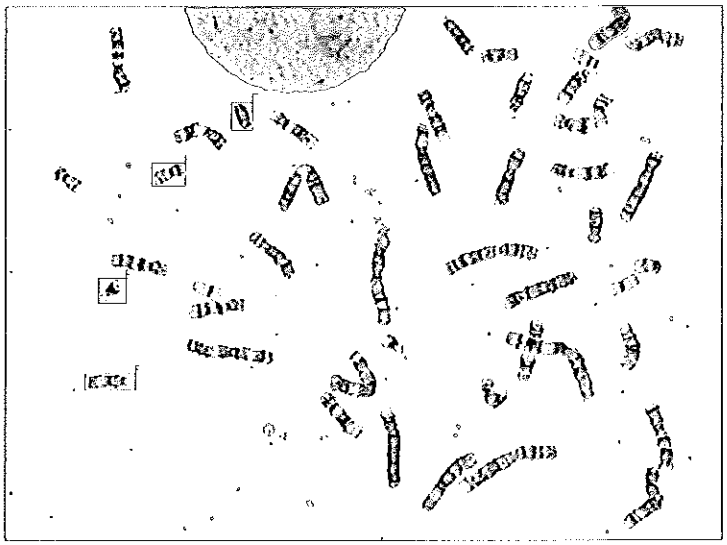
Patient name: DONOR #4552

Case name: 12-066CG

46,XY



Case: 12-066CG Slide: A3 Cell: 18



Quest Diagnostics

Report Status: Final

ID 4552, DONOR Y

Patient Information	Specimen Information	Client Information
ID 4552, DONOR Y DOB [redacted] Gender: M Phone: NG Patient ID: 4552	Specimen: [redacted] Requisition: Collected: 05/23/2012 / 13:00 CDT Received: 05/24/2012 / 05:25 CDT Reported: 05/24/2012 / 17:03 CDT	Client #: [redacted] 4195000 STERN, HARVEY J [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.04		4.20-5.80 Million/uL	CB
HEMOGLOBIN	16.5		13.2-17.1 g/dL	
HEMATOCRIT	48.3		38.5-50.0 %	
MCV	95.8		80.0-100.0 fL	
MCH	32.6		27.0-33.0 pg	
RDW	13.3		11.0-15.0 %	
HEMOGLOBIN A		95.7 L	>96.0 %	CB
HEMOGLOBIN F	1.5		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.8		1.8-3.5 %	
INTERPRETATION				

Normal phenotype.

Normal hemoglobin distribution, no HgS, HgC or other abnormal hemoglobin observed.

[redacted]

CLIENT SERVICES: 800.323.5917 SPECIMEN: WX684282L PAGE 1 OF 1

Counsyl

Results Recipient	Male	Female
[redacted] Attn: Dr. Harvey Stern [redacted] Report Date: 05/30/2012	Name: DONOR 4552 DOB: [redacted] Ethnicity: Northern European Sample Type: OG-500 Saliva Date of Collection: 05/23/2012 Date Received: 05/25/2012 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 **4 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 4552

DONOR 4552'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:

- 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 free appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

Lab Directors:

Jessica Jacobson, MD

William Seltzer, PhD, FACMG

*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.

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. CLIA Number: **#05D1102604.**

Counsyl

Male	Female
Name: DONOR 4552 DOB: [redacted]	Not tested

Mutations Tested

Beta Thalassemia - Gene: HBB. **Variants (27):** 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%.

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%.

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 70%.

Spinal Muscular Atrophy - Gene: SMN1. **Variants (1):** Exon 7 deletion. **Detection rate:** Northern European 95%.

Counsyl

Male	Female
Name: DONOR 4552 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 4552 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
Beta Thalassemia	1 in 1,500	< 1 in 1,000,000	1 in 250,000
Cystic Fibrosis	1 in 310	1 in 34,000	1 in 3,000
Sickle Cell Disease	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Spinal Muscular Atrophy	1 in 700	1 in 97,000	1 in 4,800

Integrated GENETICS
LabCorp Specialty Testing Group

Tay-Sachs Enzyme Analysis

Patient Name: Donor, 4552
Referring Physician: [redacted]
Specimen [redacted] **Client #:** [redacted]
Patient ID: [redacted]

[redacted]

DOB [redacted]
SSN:

Date Collected: 05/23/2012
Date Received: 05/25/2012
Lab ID: [redacted]
Hospital ID:
Specimen Type: **White Blood Cells**

ENTERED
CW 6/21/12

RESULTS: **Hexosaminidase Activity : 1904 nmol/mg protein**
Hexosaminidase Percent A: 59

		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.

Under the direction of: Stanford Marenberg, PHD, ABCC

Stanford Marenberg, Ph.D.

Date: 06/02/2012

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