Donor 4253

Genetic Testing Summary

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

Last Updated: 09/11/23

Donor Reported Ancestry: German, Irish, Syrian, Sicilian 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	<1/500
Tay Sachs Disease	Non- Carrier by Hexosaminidase A analysis	

*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
Fairfax Cryobank - [redacted]

Report Date: 03/15/2012

Male
Name: DONOR 4253
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Sample Type: OG-500 Saliva
Date of Collection: 03/06/2012
Date Received: 03/08/2012
Barcode: [redacted]
Indication: Egg or Sperm Donor

Female
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 4253

DONOR 4253'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.*
- Genetic counseling is recommended. To schedule a free appointment to speak with a genetic counselor about your results, please visit www.counsyl.com/appointment.

Lab Directors:

Jessica Jacobson, MD

William Seltzer, PhD, FACMG

4/12/12

*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
Name: DONOR 4253
DOB: [redacted]

Female
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:** Mixed or Other Caucasian 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:** Mixed or Other Caucasian 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:** Mixed or Other Caucasian 70%.

Spinal Muscular Atrophy - Gene: SMN1. **Variants (1):** Exon 7 deletion. **Detection rate:** Mixed or Other Caucasian 95%.

Counsyl

Male

Name: DONOR 4253
DOB: [redacted]

Female

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 4253 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

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LabCorp Specialty Testing Group

Chromosome Analysis

atient Name: Donor #4253, .
eferring Physician: [redacted]
pecimen #: [redacted]
atient ID: [redacted]
Client #: [redacted]

Fairfax Cryobank / Genetics and IVF Institute
[redacted]

OB: Not Given
SN:

Date Collected: 03/06/2012
Date Received: 03/07/2012
Lab ID:
Hospital ID:
Specimen Type: **Peripheral Blood**

dication: Gamete Donor

etaphases Counted:	20			**Banding Technique:**	GTW
etaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	575
etaphases Karyotyped:	3			**Dept. Section:**	B1

RESULTS: 46,XY
Male karyotype

ITERPRETATION:

his analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities.

he standard cytogenetic methodology utilized in this analysis does not routinely detect subtle arrangements or low-level mosaicism and cannot detect microdeletions. Also, it cannot detect molecular togenetic abnormalities (such as microdeletions and microduplications) that may be detectable by microarray alysis.

REVIEWED 4/12/12

gned: James T. Mascarello

Date: 03/14/2012

James T. Mascarello, Ph.D.

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

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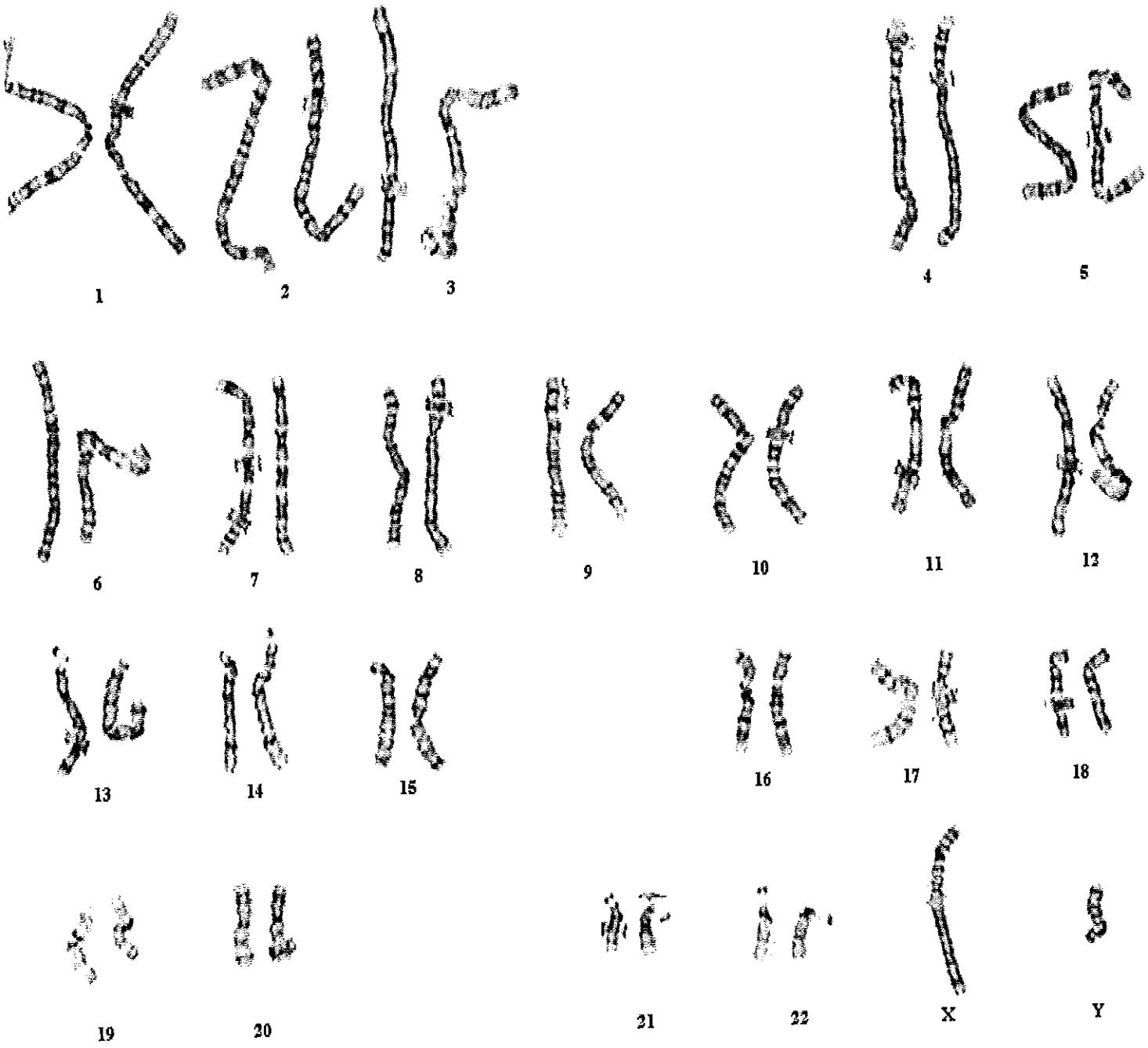


Specimen #: [redacted]
Specimen Type: BLDPER
Patient Name: Donor #4253, .
Image ID: AKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 03/07/2012
Date Reviewed: 03/14/2012
Reviewed By: JTM

Quest Diagnostics

Report Status: Final

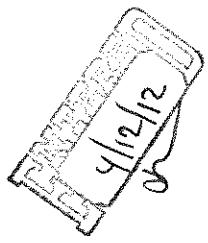
DONOR, 4253

Patient Information	Specimen Information	Client Information
DONOR, 4253 **DOB:** [redacted] **AGE:** [redacted] Gender: M Phone: NG Patient ID: [redacted]	Specimen: [redacted] Requisition: Collected: 03/06/2012 Received: 03/07/2012 / 07:18 CST Reported: 03/09/2012 / 02:22 CST	Client #: 41550 [redacted] FAIRFAX CRYOBANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.88		4.20-5.80 Million/uL	IG
HEMOGLOBIN	14.6		13.2-17.1 g/dL	
HEMATOCRIT	43.1		38.5-50.0 %	
MCV	88.4		80.0-100.0 fL	
MCH	29.9		27.0-33.0 pg	
RDW	12.8		11.0-15.0 %	
HEMOGLOBIN A	97.6		>96.0 %	IG
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.4		1.8-3.5 %	
INTERPRETATION				
Normal phenotype.				
CHOLESTEROL, TOTAL	151		125-200 mg/dL	IG
AST	24		10-40 U/L	IG
ALT	26		9-60 U/L	IG
CBC (INCLUDES DIFF/PLT)				IG
WHITE BLOOD CELL COUNT	5.2		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.88		4.20-5.80 Million/uL	
HEMOGLOBIN	14.6		13.2-17.1 g/dL	
HEMATOCRIT	43.1		38.5-50.0 %	
MCV	88.4		80.0-100.0 fL	
MCH	29.9		27.0-33.0 pg	
MCHC	33.8		32.0-36.0 g/dL	
RDW	12.8		11.0-15.0 %	
PLATELET COUNT	208		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	2683		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1602		850-3900 cells/uL	
ABSOLUTE MONOCYTES	619		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	286		15-500 cells/uL	
ABSOLUTE BASOPHILS	10		0-200 cells/uL	
NEUTROPHILS	51.6		%	
LYMPHOCYTES	30.8		%	
MONOCYTES	11.9		%	
EOSINOPHILS	5.5		%	
BASOPHILS	0.2		%	
ABO GROUP AND RH TYPE				IG
ABO GROUP	B			
RH TYPE	RH (D) NEGATIVE			

PERFORMING SITE:

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



4/10/2012

Last name: Donor
First name: #4253
DOB: 9/17/1986

COPY

HUMAN GENETICS LABORATORY
at JACOBI MEDICAL CENTER
DEPARTMENT OF PATHOLOGY
1400 PELHAM PKWY, BRONX, NY 10461
PHONE: (718) 918-7514 FAX: (718) 918-7935
CLIA #33D0668554

TAY-SACHS ENZYME ASSAY REPORT

PHYSICIAN INFORMATION:

[redacted]

PATIENT INFORMATION:
Last Name: Donor
First Name: #4253
DOB: [redacted]
Reported ethnicity: Other

Lab Specimen: Blood
Lab Specimen # [redacted]

Date Collected: 4/3/2012
Date Received: 4/4/2012
Date of Report: 4/10/2012

Indication: gamete donor
Test type: Tay-Sachs enzyme; platelet assay

RESULTS:

Platelet Hexosaminidase A (HexA) %	62%, 61%

Methods: %HexA levels were determined on platelet samples using enzymatic assay after chromatographic isoenzyme separation.
Reference: HexA %: Non-carrier ≥ 57%
Carrier ≤ 48%
Inconclusive 49% – 56%

4/26/12

INTERPRETATION:

The patient has been identified as a Tay-Sachs disease **NON-CARRIER**. No further testing is necessary.

Note: Report for Genetic Testing, if ordered, is sent under separate cover.

Disclaimers:
The Tay-Sachs enzyme assay was developed and its performance characteristics determined by Human Genetics Laboratory at Jacobi Medical Center, Department of Pathology. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. Pursuant to the requirements of CLIA '88, this laboratory has established the test's accuracy and precision. Carrier detection by enzyme assay is highly reliable, with a detection rate >98%. False positive results are rare, although pseudodeficient alleles cannot be discriminated from disease-causing alleles by this method alone.

REPORTED BY: Sachiko Nakagawa
Sachiko Nakagawa, Ph.D.

Date: 4/10/2012

ENTERED 4/12/12