Donor 2776

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: Canadian, Irish, Romanian 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 by for 99 variants 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 Globin-Related Hemoglobinopathy screening	Negative for 28 variants in the HBB gene	Sickle Cell Disease <1/500 Beta Thalassemia 1/500
Tay Sachs Enzyme Analysis	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 [redacted]bank - Fairfax
[redacted]
Report Date: 01/23/2012

Male
Name: DONOR 2776
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Sample Type: EDTA Blood
Date of Collection: 01/13/2012
Barcode: [redacted]
Indication: Egg or Sperm Donor

Female
Not tested

ENTERED
DH 3-12-12

Counsyl Test Results (Egg or Sperm Donor)

Panel: Fairfax Cryobank Fundamental Panel

The Counsyl 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 2776

DONOR 2776'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.*

To schedule a free appointment to speak with a genetic counselor about your results, please visit www.counsyl.com/appointment.

*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, 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.

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

Reproductive risk: Less than 1 in 1,000,000
Risk before testing: 1 in 250,000
Reduced risk

DONOR 2776: 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 1,500. 83% detection rate.

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.

Cystic Fibrosis

Reproductive risk: 1 in 34,000
Risk before testing: 1 in 3,000
Reduced risk

DONOR 2776: 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 310. 91% detection rate.

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.

Sickle Cell Disease

Reproductive risk: Less than 1 in 1,000,000
Risk before testing: less than 1 in 1,000,000
Reduced risk

DONOR 2776: 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. 70% detection rate.

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.

Spinal Muscular Atrophy

Reproductive risk: 1 in 97,000
Risk before testing: 1 in 4,800
Reduced risk

DONOR 2776: 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 700. 95% detection rate.

Gene: SMN1. **Variants (1):** Exon 7 deletion.

GENETICS & IVF
Institute

ENTERED
PH 3.12.12

Cytogenetic Report

Client Fairfax Cryobank - Fairfax

Address 3015 Williams Drive
Fairfax, VA 22031

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

Patient name/Donor Alias Donor # 2776 **Patient DOB** N/A

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

Collection Date 01/13/2012 **Accession #** 12-004CG

Date Received 01/13/2012

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 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

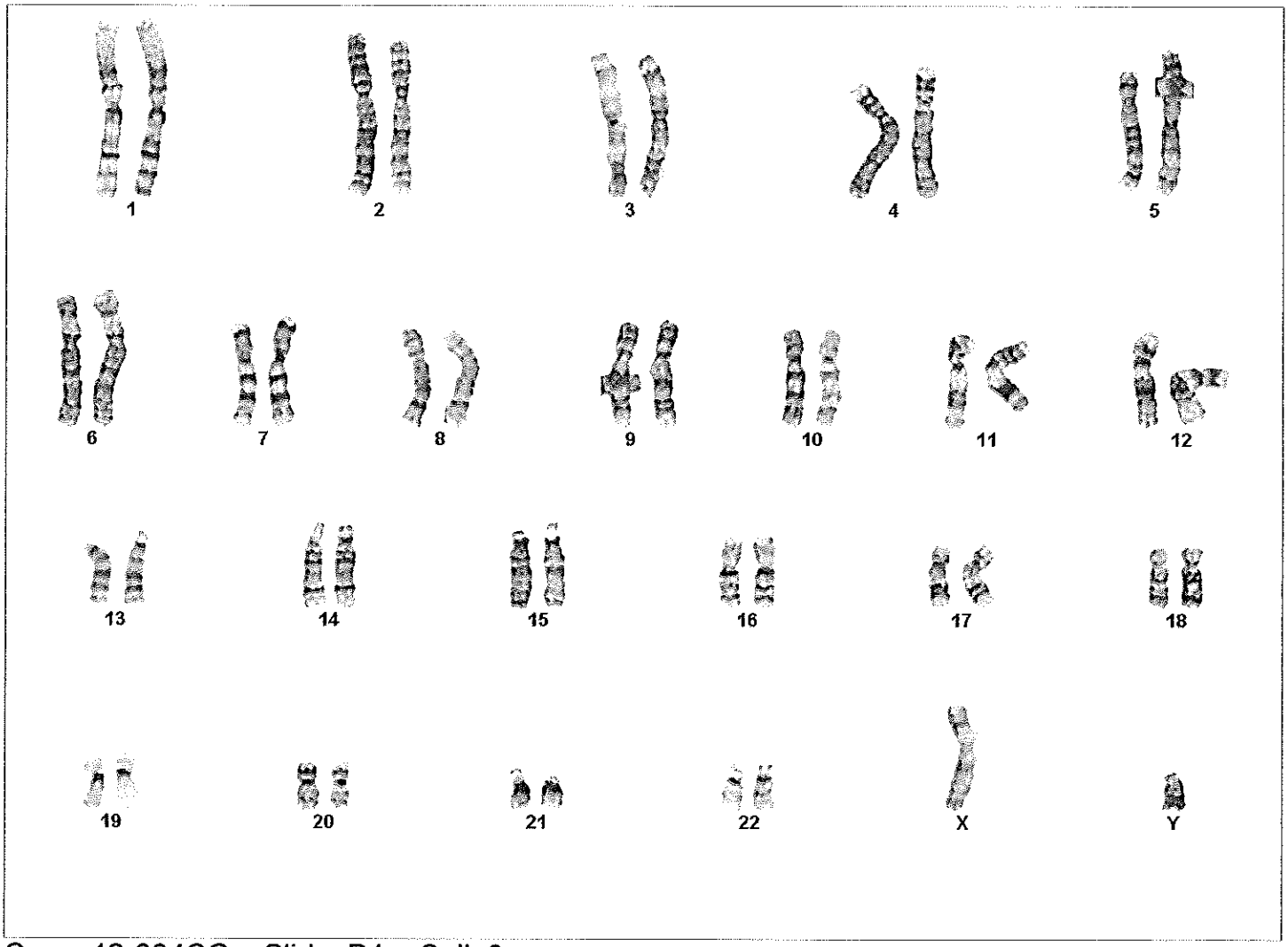
1/24/12
Date

Genetics [illegible] IVF Preimplantation Genetics L[illegible]oratory

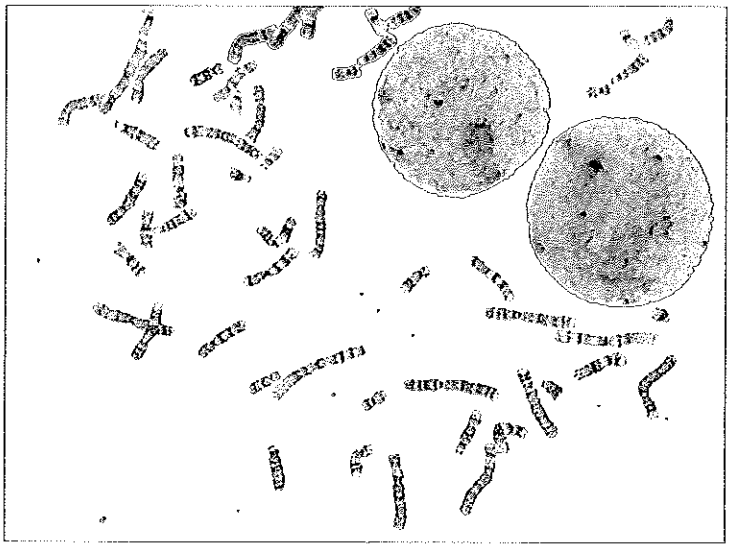
Patient name: DONOR #2776

Case name: [redacted]

46,XY



Case: 12-004CG Slide: B1 Cell: 9



QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 410.247.9100

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION:
LAB REF NO:

COLLECTED: 01/13/2012 00:00
RECEIVED: 01/13/2012 22:23
REPORTED: 01/16/2012 16:34

PATIENT INFORMATION
2776,DONOR

DOB: [redacted] Age: [redacted]
GENDER: M

ID: [redacted]

ENTERED DH 3.12.12

REPORT STATUS **Final**

ORDERING PHYSICIAN
FAIRFAX CRYOBANK
CLIENT INFORMATION
507059
FAIRFAX CRYOBANK
3015 WILLIAMS DR STE 110
FAIRFAX, VA 22031

Test Name	In Range	Out of Range	Reference Range	Lab
CHOLESTEROL,TOTAL*				QBA
CHOLESTEROL		216 H	125-200 MG/DL	
AST				QBA
AST	16		10-40 U/L	
ALT				QBA
ALT	21		9-60 U/L	
CBC (INCLUDES DIFF-PLT)				QBA
WHITE BLOOD CELL COUNT	4.4		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.59		4.20-5.80 Million/uL	
HEMOGLOBIN	14.4		13.2-17.1 g/dL	
HEMATOCRIT	41.9		38.5-50.0 %	
MCV	91		80-100 fL	
MCH	31.4		27-33 pg	
MCHC	34.4		32-36 g/dL	
PLATELET COUNT	235		140-400 Thousand/uL	
RDW	14.2		11.0-15.0 %	
MPV	8.5		7.5-11.5 fL	
ABSOLUTE NEUTROPHILS	2270		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1694		850-3900 cells/uL	
ABSOLUTE MONOCYTES	334		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	79		15-500 cells/uL	
ABSOLUTE BASOPHILS	22		0-200 cells/uL	
NEUTROPHILS	51.6		%	
LYMPHOCYTES	38.5		%	
REACTIVE LYMPHOCYTES	0.0		%	
MONOCYTES	7.6		%	
EOSINOPHILS	1.8		%	
BASOPHILS	0.5		%	
COMMENT				
HEMOGLOBINOPATHY EVALUATION				QBA
RED BLOOD CELL COUNT	4.59		4.20-5.80 Million/uL	
HEMOGLOBIN	14.4		13.2-17.1 g/dL	
HEMATOCRIT	41.9		38.5-50.0 %	
MCV	91		80-100 fL	
MCH	31.4		27-33 pg	
RDW	14.2		11.0-15.0 %	
HEMOGLOBIN A	97.6		>96.0 %	
HEMOGLOBIN F	NONE DETECTED		0.0-1.9	
HEMOGLOBIN A2	2.4		1.8-3.5 %	
HGB SCREEN INTERPRETATION				
	THE HEMOGLOBINOPATHY SCREEN IS NORMAL.			

genzyme
Genetics

Tay-Sachs Enzyme Analysis

Patient Name: Donor, #2776
Referring Physician: Steve Pool, MD
Specimen #: 18146334 **Client #:** 606452
Patient ID: 14553684-1

DOB: Not Given
SSN:

Date Collected: 01/13/2012
Date Received: 01/16/2012
Lab ID:
Hospital ID:
Specimen Type: **White Blood Cells**

Fairfax Cryobank / Genetics and IVF Institute
Genetics and IVF Institute
3015 Williams Drive
Suite 110
Fairfax VA 22031

ENTERED DH 3.12.12

RESULTS: **Hexosaminidase Activity : 1051 nmol/mg protein**
Hexosaminidase Percent A: 63.1

		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, FACC

Stanford Marenberg, Ph.D.

Date: 01/21/2012

Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436