Donor 2782

Genetic Testing Summary

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

Last Updated: 03/21/24

Donor Reported Ancestry: English, French, German 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	Beta Thalassenmia: 1/500 Sickle Cell Disease: <1/500

*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 Cry[illegible]k - Fairfax
[redacted]
Report Date: 12/08/2011

Male
Name: DONOR 2782
DOB: [redacted]
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 12/02/2011
Barcode [redacted]
Indication: Egg or Sperm Donor

Female
Not tested

ENTERED DH 2-13-12

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 2782

DONOR 2782'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.

Counsyl

Male
Name: DONOR 2782
DOB: [redacted]

Female
Not tested

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

DONOR 2782: 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:	Risk before testing:	
1 in 34,000	1 in 3,000	Reduced risk

DONOR 2782: 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:	Risk before testing:	
Less than 1 in 1,000,000	less than 1 in 1,000,000	Reduced risk

DONOR 2782: 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:	Risk before testing:	
1 in 97,000	1 in 4,800	Reduced risk

DONOR 2782: 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.

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. Lab Directors: Jessica Jacobson, MD, William K. Seltzer, PhD, FACMG.

GENETICS & IVF
Institute

Cytogenetic Report

ENTERED DH 2-13-12

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted] **Fax #** [redacted]4 **Email** [redacted]

Patient name/Donor Alias Donor # 2782 **Patient DOB** N/A

Donor # 2782 **Specimen type** Peripheral Blood

Collection Date 12/02/2011 **Accession #** [redacted]

Date Received 12/02/2011

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	50	**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

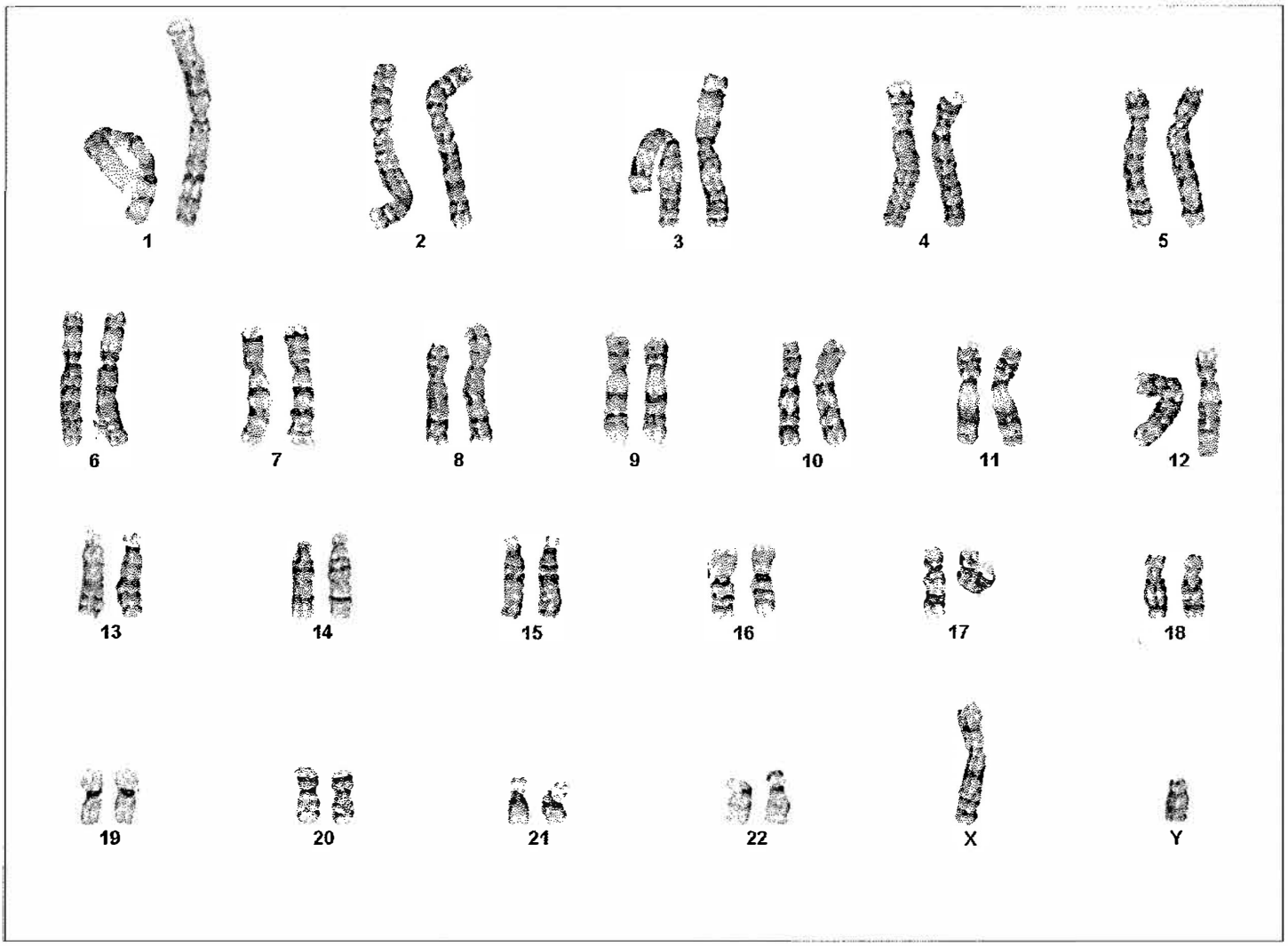
12/15/11
Date

Genetics a IVF Preimplantation Genetics L(oratory

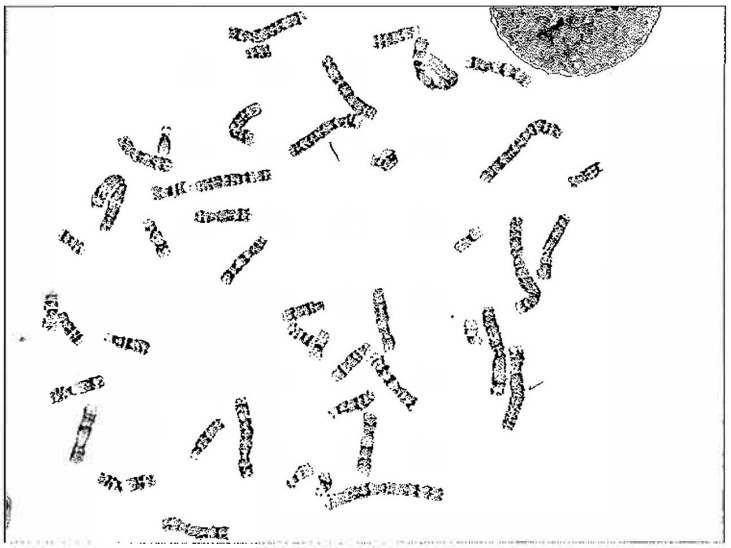
Patient name: DONOR #2782

Case name: [redacted]

46,XY



Case: 11-132CG Slide: A1 Cell: 4



QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 410.247.9100

PATIENT INFORMATION
2782,DONOR

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

REPORT STATUS **Final**

ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
[redacted]
FAIRFAX CRYOBANK
[redacted]

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

ID: 2782[redacted]

ENTERED 12-13-12

COLLECTED: 12/02/2011 00:00
RECEIVED: 12/02/2011 22:36
REPORTED: 12/06/2011 10:33

Test Name	In Range	Out of Range	Reference Range	Lab
CHOLESTEROL,TOTAL*				QBA
CHOLESTEROL	185		125-200 MG/DL	
AST				QBA
AST	21		10-40 U/L	
ALT				QBA
ALT	23		9-60 U/L	
CBC (INCLUDES DIFF-PLT)				QBA
WHITE BLOOD CELL COUNT	4.9		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.74		4.20-5.80 Million/uL	
HEMOGLOBIN	14.7		13.2-17.1 g/dL	
HEMATOCRIT	43.9		38.5-50.0 %	
MCV	93		80-100 fL	
MCH	30.9		27-33 pg	
MCHC	33.4		32-36 g/dL	
PLATELET COUNT	178		140-400 Thousand/uL	
RDW	13.2		11.0-15.0 %	
MPV	8.6		7.5-11.5 fL	
ABSOLUTE NEUTROPHILS	2935		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1308		850-3900 cells/uL	
ABSOLUTE MONOCYTES	446		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	176		15-500 cells/uL	
ABSOLUTE BASOPHILS	34		0-200 cells/uL	
NEUTROPHILS	59.9		%	
LYMPHOCYTES	26.7		%	
REACTIVE LYMPHOCYTES	0.0		%	
MONOCYTES	9.1		%	
EOSINOPHILS	3.6		%	
BASOPHILS	0.7		%	
COMMENT				
HEMOGLOBINOPATHY EVALUATION				QBA
RED BLOOD CELL COUNT	4.74		4.20-5.80 Million/uL	
HEMOGLOBIN	14.7		13.2-17.1 g/dL	
HEMATOCRIT	43.9		38.5-50.0 %	
MCV	93		80-100 fL	
MCH	30.9		27-33 pg	
RDW	13.2		11.0-15.0 %	
HEMOGLOBIN A	97.7		>96.0 %	
HEMOGLOBIN F	NONE DETECTED		0.0-1.9	
HEMOGLOBIN A2	2.3		1.8-3.5 %	
HGB SCREEN INTERPRETATION				
	THE HEMOGLOBINOPATHY SCREEN IS NORMAL.			

PATIENT INFORMATION
2782,DONOR

REPORT STATUS **Final**

QUEST DIAGNOSTICS INCORPORATED

ORDERING PHYSICIAN [redacted]

DOB: [redacted] Age: [redacted]
GENDER: M
ID: 2782[redacted]

COLLECTED: 12/02/2011 00:00
REPORTED: 12/06/2011 10:33

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION (Continued)				
ABNORMAL HEMOGLOBIN #1 %:	0.0		%	

Performing Laboratory Information:

QBA Quest Diagnostics Incorporated 1901 Sulphur Spring Road Baltimore MD 21227 Laboratory Director: Robert R. L. Smith, M.D.