Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4570

Genetic Testing Summary

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

Last Updated: 06/16/23

Donor Reported Ancestry: German, French Canadian, Welsh 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 for 99 variants in the in the CFTR gene	1/160
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/570
Hb Beta Chain Related Hemoglobinopathies including Beta Thalassemia and Sickle Cell Disease	Negative for 28 variants in the HBB gene	1/930
Tay Sachs Enzyme Analysis	Non-Carrier by Hexosaminidase A testing	
Special Testing		
Gene: SERPINA1	Negative for 4 variants in the SERPINA1 gene	

*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

Result Recipient	Male	Female
[redacted]	Name: DONOR 4570 DOB: [redacted] Ethnicity: French Canadian or Cajun Sample Type: OG-510 Saliva Date of Collection: 10/31/2012 Date Received: 11/01/2012 Barcode: [redacted] Indication: Egg or Sperm Donor	Not tested

Report Date: 11/02/2012

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 **3 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 4570

DONOR 4570'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:

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

Lab Directors:

William Seltzer, PhD, FACMG

H. Peter Kang, MD

*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 4570
DOB: [redacted]

Female
Not tested

Mutations 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:** French Canadian or Cajun 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:** French Canadian or Cajun 83%.

Spinal Muscular Atrophy - Gene: SMN1. **Variants (1):** SMN1 copy number. **Detection rate:** French Canadian or Cajun 94%.

ENTERED 6/11/07/12

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 4570 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
Cystic Fibrosis	1 in 160	1 in 9,700	1 in 900
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 930	1 in 590,000	1 in 100,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 570	1 in 79,000	1 in 4,800

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
3015 Williams Drive, #110, Fairfax, VA, 22031, US
Physician: Suzanne Seitz
Report Generated: 2015-09-08
Report Updated: 2015-09-09

Donor 4570

DOB: 1986-09-01
Gender: Male
Ethnicity: European
Procedure ID: 28911
Kit Barcode: 20150820507184
Method: Genotyping
Specimen: Blood, #30598
Specimen Collection: 2015-08-28
Specimen Received: 2015-08-31
Specimen Analyzed: 2015-09-08

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4570 was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

♂ Male

Panel: Alpha-1-Antitrypsin Deficiency , Diseases Tested: 1, Mutations Tested: 4, Genes Tested: 1, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31D1054821
Lab Technician Bo Chu

Reviewed by Pere Colls, PhD, HCLD, Lab Director

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in >200 genes. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors.

The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

Diseases & Mutations Assayed

High Impact Treatment Benefits X-Linked Moderate Impact

H	T	X	M	Disease	#	Mutations
○	○	○	●	Alpha-1-Antitrypsin Deficiency	4	♂ Genotyping | c.226_228delTTC (p.76delF), c.A1131T (p.L377F), c.C187T (p.R63C), c.G1096A (p.E366K)

Integrated GENETICS
LabCorp Specialty Testing Group

Tay-Sachs Enzyme Analysis

Patient Name: Donor, 4570
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

[redacted]

DOB: [redacted]
SSN: ***-**-****

Date Collected: 11/09/2012
Date Received: 11/10/2012
Lab ID: 4570-121109
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 1868 nmol/mg protein**
Hexosaminidase Percent A: 58.2

ENTERED dw 11/27/12

		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: 11/15/2012

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

Integrated GENETICS
LabCorp Specialty Testing Group

Chromosome Analysis

Patient Name: Donor, 4570
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

[redacted]

DOB: [redacted]
SSN: ***-**-****

Date Collected: 11/09/2012
Date Received: 11/10/2012
Lab ID: 4570-121109
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: No family history / Gamete donor

ENTERED 11/27/12

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	625
Metaphases Karyotyped:	3			**Dept. Section:**	B1

RESULTS: 46,XY
Male karyotype

INTERPRETATION:

This analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. The standard cytogenetic methodology utilized in this analysis does not routinely detect subtle rearrangements or low-level mosaicism and cannot detect microdeletions. Also, it cannot detect molecular cytogenetic abnormalities (such as microdeletions and microduplications) that may be detectable by microarray analysis.

Signed: Jay C Leonard

Date: 11/16/2012

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Jay C Leonard, Ph.D. FACMG

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

Quest Diagnostics

Report Status: Final

ID4570, DONOR

Patient Information	Specimen Information	Client Information
ID4570, DONOR **DOB:** [redacted] **AGE:** [redacted] Gender: M Phone: NG Patient ID: Health ID: [redacted]	Specimen: Requisition: [redacted] Lab Ref #: Collected: 10/31/2012 / 11:30 CDT Received: 11/01/2012 / 03:13 CDT Reported: 11/02/2012 / 13:58 CDT	Client #: [redacted] 4195000 [redacted] ENTERED 11/2/12

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.79		4.20-5.80 Million/uL	CB
HEMOGLOBIN	15.5		13.2-17.1 g/dL	
HEMATOCRIT	45.0		38.5-50.0 %	
MCV	93.9		80.0-100.0 fL	
MCH	32.3		27.0-33.0 pg	
RDW	13.7		11.0-15.0 %	
HEMOGLOBIN A	97.8		>96.0 %	CB
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.2		1.8-3.5 %	
INTERPRETATION				
	Normal phenotype.			
	Normal hemoglobin distribution, no HgS, HgC or other abnormal hemoglobin observed.			
CHOLESTEROL, TOTAL	147		125-200 mg/dL	CB
AST	16		10-40 U/L	CB
ALT	17		9-60 U/L	CB
CBC (INCLUDES DIFF/PLT)				CB
WHITE BLOOD CELL COUNT	5.9		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.79		4.20-5.80 Million/uL	
HEMOGLOBIN	15.5		13.2-17.1 g/dL	
HEMATOCRIT	45.0		38.5-50.0 %	
MCV	93.9		80.0-100.0 fL	
MCH	32.3		27.0-33.0 pg	
MCHC	34.4		32.0-36.0 g/dL	
RDW	13.7		11.0-15.0 %	
PLATELET COUNT	245		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	3393		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1658		850-3900 cells/uL	
ABSOLUTE MONOCYTES	419		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	413		15-500 cells/uL	
ABSOLUTE BASOPHILS	18		0-200 cells/uL	
NEUTROPHILS	57.5		%	
LYMPHOCYTES	28.1		%	
MONOCYTES	7.1		%	
EOSINOPHILS	7.0		%	
BASOPHILS	0.3		%	
ABO GROUP AND RH TYPE				CB
ABO GROUP	B			
RH TYPE	RH(D) POSITIVE			

PERFORMING SITE:

CB QUEST DIAGNOSTICS WOOD DALE, 1355 MITTEL BOULEVARD, WOOD DALE, IL 60191-1024 Laboratory Director: ANTHONY V. THOMAS, MD, CLIA: 14D0417052