Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4892

Genetic Testing Summary

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

Last Updated: 08/17/18

Donor Reported Ancestry: German, Polish, Irish, French, Norwegian, Swedish 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	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/610
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/290
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.

Counsyl

RESULTS RECIPIENT
CRYOGE ABORATORIES
[redacted]
Report Date: 05/27/2015

MALE
DONOR 4892
DOB: [redacted]
Ethnicity: Northern European
Sample Type: OG-510 Saliva
Date of Collection: 05/18/2015
Date Received: 05/20/2015
Date Tested: 05/27/2015
Barcode: [redacted]
Indication: Egg or sperm donor

FEMALE
N/A

ENTERED 5th 06/02/15

Family Prep Screen

NEGATIVE

ABOUT THIS TEST

The Counsyl Family Prep Screen (version 1.0) tests known mutations to help you learn about your chance to have a child with a genetic disease.

PANEL DETAILS

Fairfax Cryobank Fundamental Panel (3 conditions tested)

VERSION

DONOR 4892 (Family Prep Screen 1.0)

RESULTS SUMMARY

NEGATIVE

No known or potential disease-causing mutations were detected. A complete list of all conditions tested can be found on page 3.

CLINICAL NOTES

- None

NEXT STEPS

- If necessary, patients can discuss residual risks with their physician or a genetic counselor.
- To schedule a complimentary appointment to speak with a clinical expert about these results, please visit counsyl.com/my/consults/.

Methods and Limitations

DONOR 4892 (Family Prep Screen 1.0): targeted genotyping and copy number analysis.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 127 variants associated with 2 diseases. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

Copy number analysis: Targeted copy number analysis is used to determine the copy number of exon 7 of the SMN1 gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of SMN1 are carriers with two SMN1 genes on one chromosome and a SMN1 deletion on the other chromosome. In addition, a small percentage of SMA cases are caused by nondeletion mutations in the SMN1 gene. Thus, a test result of two SMN1 copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more SMN1 gene copies. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

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, bone marrow transplantation, blood transfusions and technical errors. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. 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 evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet. Gynecol. 2007;109:229-37*).

This test was developed and its performance characteristics determined by Counsyl, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: **#05D1102604**.

LAB DIRECTORS

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Rebecca Mar-Heyming, PhD, DABMG

Conditions Tested

Autosomal Recessive Disorders

TARGETED GENOTYPING

Cystic Fibrosis - Gene: CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542*, G551D, R553*, R560T, R1162*, W1282*, N1303K, F508del, I507del, c.2052delA, c.3528delC, c.489+1G>T, c.579+1G>T, c.1585-1G>A, c.1766+1G>A, 2789+5G>A, c.2988+1G>A, 3849+10kbC>T, E60*, R75*, E92*, Y122*, G178R, R347H, Q493*, V520F, S549N, P574H, M1101K, D1152H, c.2012delT, c.262_263delTT, c.313delA, c.948delT, c.3744delA, c.3773dupT, c.1680-1G>A, 3272-26A>G, c.2051_2052delAAinsG, S549R(c.1645A>C), R117C, L206W, G330*, T338I, R352Q, S364P, G480C, C524*, S549R(c.1647T>G), Q552*, A559T, G622D, R709*, K710*, R764*, Q890*, R1066C, W1089*, Y1092X, R1158*, S1196*, W1204*, Q1238*, S1251N, S1255*, 3199del6, c.442delA, c.531delT, c.803delA, c.805_806delAT, c.1545_1546delTA, M607_Q643del, c.1911delG, c.1923_1931del9ins1, c.1976delA, c.3039delC, c.3536_3539delCCAA, c.3659delC, c.1155_1156dupTA, c.2052dupA, c.2175dupA, c.2738insG, 296+12T>C, c.273+1G>A, 405+3A>C, c.274-1G>A, 711+5G>A, c.580-1G>T, c.1766+1G>T, 1898+5G>T, Q996, c.325_327delTATinsG, 3849+4A>G, c.1075_1079del5ins5. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection rate**: Northern European 91%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. **Variants (28):** E7V, K18*, Q40*, c.126_129delCTTT, c.27dupG, IVS-II-654, IVS-II-745, c.315+1G>A, IVS-I-6, IVS-I-110, IVS-I-5, c.92+1G>A, -88C>T, -28A>G, -29A>G, c.25_26delAA, c.217dupA, c.316-2A>C, c.316-2A>G, G25, -87C>G, E7K, W16*, c.51delC, c.20delA, Hb E, E122Q, E122K. **Detection rate**: Northern European 83%.

COPY NUMBER ANALYSIS

Spinal Muscular Atrophy - Gene: SMN1. **Variant (1):** SMN1 copy number. **Detection rate**: Northern European 95%.

Risk Calculations

Below are the risk calculations for all conditions tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Disease	DONOR 4892 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000

PATIENT INFORMATION
4892,DONOR

REPORT STATUS Final

Nichols Institute, Chantilly

ORDERING PHYSICIAN
[redacted]

DOB: [redacted] Age: [redacted]
SEX: M

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

ID: 4892-[redacted]

COLLECTED: 05/18/2015 00:00
RECEIVED: 05/19/2015 10:11
REPORTED: 05/29/2015 14:53

Test Name	In Range	Out of Range	Reference Range	Lab
Chromosome Analysis, Blood				AMD
Chromosome Analysis, Blood				

CYTOGENETIC RESULTS

ENTERED tm 06/02/15

Cytogenetic Reference #: CB-15-008863
Test Setup Date: 05/19/2015
Test Completion Date: 05/29/2015
Specimen Source: Peripheral Blood
Clinical History:Screening

Metaphases Counted:20 Analyzed:5 Karyotyped:2
Banding Level (G-bands):>=550

KARYOTYPE:
46,XY

INTERPRETATION and COMMENTS:
NORMAL MALE karyotype

Within the limits of standard cytogenetic methodologies, the chromosomes had normal G-banding patterns without apparent structural abnormality or rearrangement.

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods, or rare events such as low level mosaicism or very subtle rearrangements.

Electronic Signature on File

Steven A. Schonberg, Ph.D., FACMG
Technical Director, Cytogenetics, 703-802-7156

Results Received 05/29/15
Reference lab accession: CB15008863EC

For more information on this test, go to
http://education.questdiagnostics.com/faq/chromsblood

PATIENT INFORMATION
4892,DONOR

REPORT STATUS Final

Nichols Institute, Chantilly

ORDERING PHYSICIAN
STERN

DOB: [redacted] Age: 28Y
SEX: M
ID: 4892[redacted]

COLLECTED: 05/18/2015 00:00
REPORTED: 05/29/2015 14:53

Performing Laboratory Information:

AMD QUEST DIAGNOSTICS INCORPORATED NICHOLS INSTITUTE 14225 NEWBROOK DR CHANTILLY VA 20151
Laboratory Director: PATRICK W. MASON, MD, PHD

Quest Diagnostics®

PATIENT INFORMATION:
4892, DONOR

DOB: [redacted]
GENDER: M
PATIENT ID: 4892-[redacted]

Cytogenetics Karyotype Report

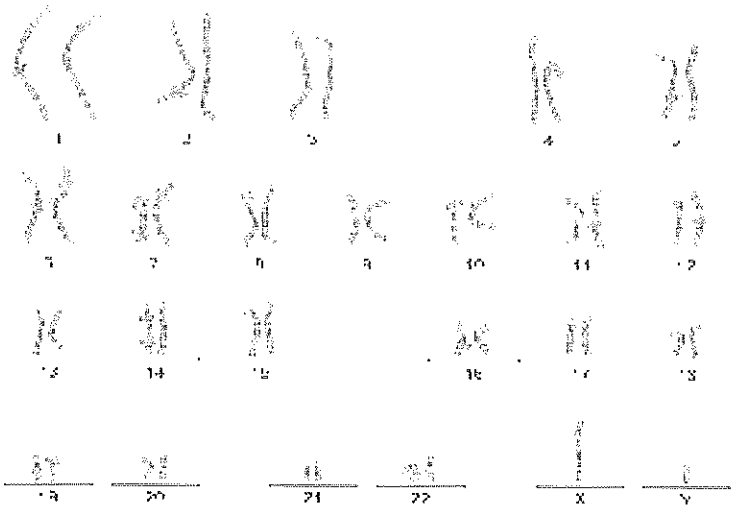
REPORT STATUS: FINAL

ORDERING PHYSICIAN:
[redacted]

SPECIMEN ID:
REQUISITION: [redacted]
COLLECTED: 05/18/2015 00:00
RECEIVED: 05/19/2015 10:11
REPORTED: 05/29/2015 11:53

CYTOGENETICS - Karyotype (14596 Chromosome Analysis, Blood)

Case Number: [redacted]
Specimen Source: Peripheral Blood
Clinical History: Screening
Metaphases Counted: 20
Metaphases Analyzed: 5
Metaphases Karyotyped: 2
Banding Level: >=550



Interpretation / Comments

NORMAL MALE karyotype

Within the limits of standard cytogenetic methodologies, the chromosomes had normal G-banding patterns without apparent structural abnormality or rearrangement.

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods, or rare events such as low level mosaicism or very subtle rearrangements.

Reviewed by

Electronic Signature on File

Steven A. Schonberg, Ph.D., FACMG
Technical Director, Cytogenetics, 703-802-7156

Quest Diagnostics

Report Status: Final

ID, 4892 D

Patient Information	Specimen Information	Client Information
ID, 4892 D DOB [redacted] AGE: [redacted] Gender: M Phone: NG Patient ID: [redacted] Health ID: [redacted]	Specimen: [redacted] Requisition: Lab Ref #: Collected: 05/21/2015 Received: 05/22/2015 / 02:54 CDT Reported: 05/24/2015 / 14:07 CDT	Client #: 22663146 4195000 [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.45		4.20-5.80 Million/uL	CB
HEMOGLOBIN	16.6		13.2-17.1 g/dL	
HEMATOCRIT	49.7		38.5-50.0 %	
MCV	91.2		80.0-100.0 fL	
MCH	30.5		27.0-33.0 pg	
RDW	13.6		11.0-15.0 %	
HEMOGLOBIN A	97.4		>96.0 %	CB
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.6		1.8-3.5 %	
INTERPRETATION				

Normal phenotype.

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

ENTERED 05/02/15

PERFORMING SITE:

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

CLIENT SERVICES: 866.697.8378

SPECIMEN: [redacted]

C 5/27/15

Integrated GENETICS

Tay-Sachs Enzyme Analysis

Patient Name: Donor, 4892
Referring Physician: [redacted]
Specimen # [redacted] **Client #:** 606452
Patient ID: [redacted]

[redacted]

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

Date Collected: 05/21/2015
Date Received: 05/22/2015
Lab ID: 4892-150521
Hospital ID:
Specimen Type: **White Blood Cells**

ENTERED 5/26/2015

RESULTS: **Hexosaminidase Activity : 1603 nmol/mg protein**
Hexosaminidase Percent A: 64.8

		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, MDCC

Stanford Marenberg, Ph.D.

Date: 05/29/2015

Testing Performed At Esoterix Genetic Laboratories, LLC 2000 Vivigen Way Santa Fe, NM 87505 Philip Wyatt, MD, PhD, Laboratory Director 1-800-848-4436