Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4951

Genetic Testing Summary

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

Last Updated: 03/17/22

Donor Reported Ancestry: German, Irish, Polish Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype (attached)	46,XY,inv(9)(p12q13). Normal chromosome variant. 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
Special testing		
Genes: GJB2, ACADS	Negative by genotyping	See attached result for residual risks

*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]
Attn: Dr. Harvey Stern
[redacted]
Report Date: 02/11/2015

MALE
DONOR 4951
DOB: [redacted]
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 02/02/2015
Date Received: 02/04/2015
Date Tested: 02/11/2015
Barcode: [redacted]
Indication: Egg or Sperm Donor

FEMALE
N/A

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 diseases tested)

VERSION

DONOR 4951 (Family Prep Screen 1.0)

RESULTS SUMMARY

NEGATIVE
No known or potential disease-causing mutations were detected.

ENTERED
NP 02.13.15

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/.

180 Kimball Way, South San Francisco, CA 94080
(888) COUNSYL | http://www.counsyl.com

Methods and Limitations

DONOR 4951 (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 workup. CLIA Number: **#05D1102604.**

LAB DIRECTORS

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Rebecca Mar-Heyming, PhD, DABMG

Diseases Tested

Autosomal Recessive Disorders

TARGETED GENOTYPING

Cystic Fibrosis - Gene: CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542*, G551D, R553*, R560T, R1162*, W1282*, N1303K, c.1521_1523delCTT, c.1519_1521delATC, 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*, c.3067_3072del6, 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, E27K, 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 diseases 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 4951 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	Specimen Information	Client Information
ID, 4951 **DOB: Not Given** **AGE: Not Given** Gender: M Fasting: N Phone: NG Patient ID: NG	Specimen: [redacted] Requisition Collected: 02/02/2015 / 08:45 EST Received: 02/02/2015 / 23:52 EST Faxed: 02/03/2015 / 19:01 EST	Client #: 19104437 HO130000 STERN, HARVEY J FAIRFAX CRYO BANK [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	QHO
HEMOGLOBIN	15.1		13.2-17.1 g/dL	
HEMATOCRIT	45.0		38.5-50.0 %	
MCV	92.1		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
RDW	12.5		11.0-15.0 %	
HEMOGLOBIN A	97.5		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.5		1.8-3.5 %	
INTERPRETATION				
	Normal phenotype.			
SPECIMEN ID NOTIFICATION				QHO
MISSING SECOND ID				
Only one form of patient ID was present on one or more sample(s); two forms of patient ID are required by the College of American Pathologists (CAP).				

PERFORMING SITE:

QHO QUEST DIAGNOSTICS HORSHAM, 900 BUSINESS CENTER DRIVE, HORSHAM, PA 19044-3432 Laboratory Director: ANDREW S EDELMAN, MD PHD, CLIA: 39D0204404

NE 02.17.15

Integrated GENETICS

Chromosome Analysis

Patient Name: 4951, [redacted]
Referring Physician: [redacted]
Specimen # [redacted] **Client #:** 606452
Patient ID: 16618612-1-B1

Fairfax Cryobank [redacted] Genetics and IVF Institute
Genetics and IVF Institute
[redacted]

DOB: Not Given
SSN: ***-**-****

Date Collected: 02/02/2015
Date Received: 02/03/2015
Lab ID:
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Sperm donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	550
Metaphases Karyotyped:	3			**Dept. Section:**	B1

RESULTS: 46,XY,inv(9)(p12q13)
Variant male karyotype

INTERPRETATION:

Cytogenetic analysis shows an apparently balanced pericentric inversion of one chromosome 9. This inversion 9 is considered a normal chromosome variant, and has not been associated with any phenotypic abnormality (Gardner, RJ, Sutherland GR, and Shaffer LG; Chromosome Abnormalities and Genetic Counseling, Fourth Edition, Oxford University Press, 2012. pp. 163, 174, 258).

COMMENT:
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.

Considered normal variant
2/19/15

ENTERED
No 02.17.15

Signed: Jay C Leonard

Date: 02/12/2015

TE

Jay C Leonard, Ph.D. FACMG

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 1170
Fairfax Cryobank - [redacted]
[redacted]
Physician: [redacted]
Report Generated: 2016-07-28

4951 [redacted]

DOB: [redacted]
Gender: Male
Ethnicity:
Procedure ID: 60209
Kit Barcode: [redacted]
Specimen: Blood, #63274
Specimen Collection: 2016-07-20
Specimen Received: 2016-07-21
Specimen Analyzed: 2016-07-28

Partner Not Tested

TEST INFORMATION

Test: CarrierMap^{GEN} (Genotyping)
Panel: Custom Panel
Diseases Tested: 2
Genes Tested: 2
Mutations Tested: 35

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

4951 [redacted] was not identified to carry any of the mutation(s) tested.

No pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.

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

Assay performed by Reprogenetics
CLIA ID: 31D1054821
3 Regent Street, Livingston, NJ 07039
Lab Technician: Bo Chu

Recombine CLIA # 31D2100763
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 the genes tested. 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.

This test was developed and its performance determined by Recombine, Inc., and it has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

Diseases & Mutations Assayed

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutations (30): ♂
Genotyping | c.167delT, c.235delC, c.312_325delGAAGTTCATCAAGG, c.358delGAG (p.120delE), c.35delG, c.370C>T (p.Q124X), c.427C>T (p.R143W), c.109G>A (p.V37I), c.231G>A (p.W77X), c.551G>C (p.R184P), c.71G>A (p.W24X), c.101T>C (p.M34T), c.229T>C (p.W77R), c.269T>C (p.L90P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T, c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147K), c.-23+1G>A, c.550C>T (p.R184W), c.-259C>T

Short-Chain Acyl-CoA Dehydrogenase Deficiency (ACADS): Mutations (5): ♂
Genotyping | c.1058C>T (p.S353L), c.1138C>T (p.R380W), c.1147C>T (p.R383C), c.319C>T (p.R107C), c.575C>T (p.A192V)

Residual Risk Information

Detection rates are calculated from the primary literature and may not be available for all ethnic populations. The values listed below are for genotyping. Sequencing provides higher detection rates and lower residual risks for each disease. More precise values for sequencing may become available in the future.

Disease	Carrier Rate	Detection Rate	Residual Risk
Nonsyndromic Hearing Loss and Deafness: GJB2 Related	♂ Ashkenazi Jewish: 1/20	95.83%	1/480
	♂ Chinese: 1/100	82.26%	1/564
	♂ European: 1/53	83.98%	1/331
	♂ Indian: Unknown	66.98%	Unknown
	♂ Israeli: 1/16	93.10%	1/232
	♂ Japanese: 1/75	75.00%	1/300
	♂ Roma: Unknown	>99%	Unknown
	♂ United States: 1/34	46.50%	1/64
Short-Chain Acyl-CoA Dehydrogenase Deficiency	♂ Ashkenazi Jewish: 1/15	65.00%	1/43