Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4869

Genetic Testing Summary

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

Last Updated: 12/14/23

Donor Reported Ancestry: Swedish, Dutch, French Canadian 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	1/160
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/570
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/930
Tay Sachs Enzyme Analysis	Non-carrier by Hexosaminidase A testing	

*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
[redacted]
Attn: [redacted]
[redacted]
Report Date: 11/01/2014

MALE
DONOR 4869
DOB: [redacted]
Ethnicity: French Canadian or Cajun
Sample Type: OG-510 Saliva
Date of Collection: 10/24/2014
Date Received: 10/27/2014
Date Tested: 11/01/2014
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 4869 (Family Prep Screen 1.0)

RESULTS SUMMARY

NEGATIVE

No known or potential disease-causing mutations were detected.

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 4869 [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

Counsyl

RESULTS [redacted]

Report Date: 11/01/2014

MALE

DONOR 4869

DOB: [redacted]

Ethnicity: French Canadian or Cajun

Barcode: [redacted]

FEMALE

N/A

ENTERED 11/4/14

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, R117C, L206W, G330*, T338I, R352Q, S364P, G480C, C524*, S549R, 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**: French Canadian or Cajun 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**: French Canadian or Cajun 83%.

COPY NUMBER ANALYSIS

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

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 4869 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 160	1 in 9,700
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 930	1 in 590,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 570	1 in 79,000

GENETICS & IVF
Institute

Cytogenetic Report

ENTERED 11/6/14

Client [redacted]

Address [redacted]

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

Patient name/Donor Alias Donor # 4869 Patient DOB N/A

Donor # 4869-[redacted] Specimen type Peripheral Blood

Collection Date 10/24/2014 Accession # [redacted]

Date Received 10/25/2014

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	20	Type of banding	GTG
Cells analyzed	5	Band resolution	550
Cells karyotyped	2		
Modal chromosome #	46		

KARYOTYPE 46,XY

FISH

Probe(s)	N/A
Nuclei scored	N/A

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

W Wayne PhD

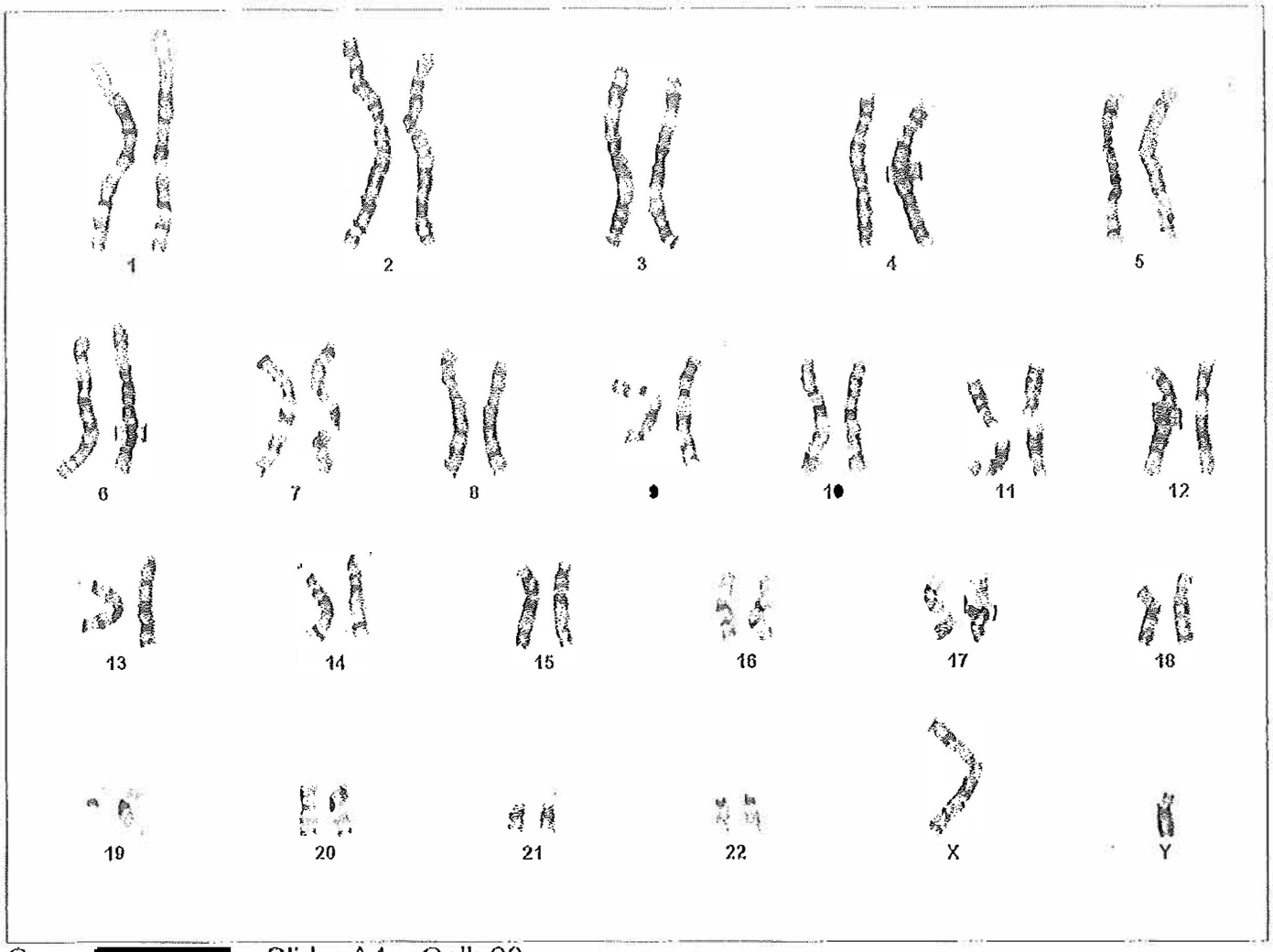
Wayne S. Stanley, Ph.D., FACMG
Clinical Cytogeneticist

11/6/14
Date

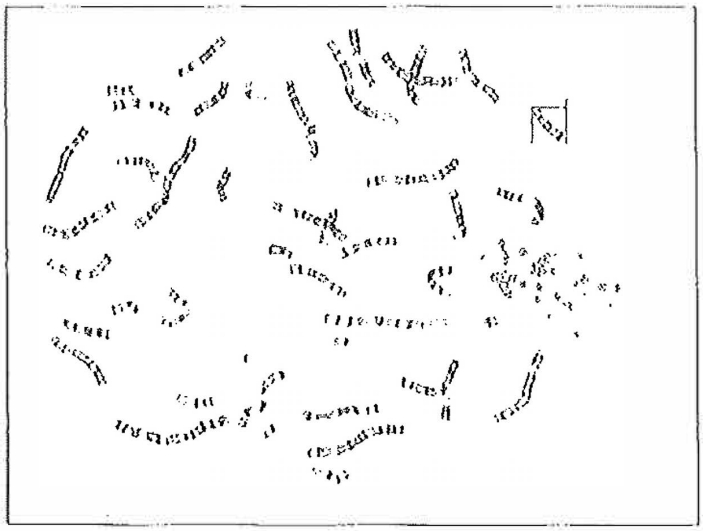
Patient name: DONOR # 4869

Case name: [redacted]

46,XY



Case: [redacted] Slide: A4 Cell: 20



Quest Diagnostics

Report Status: Final

ID4869, DONOR

Patient Information	Specimen Information	Client Information
ID4869, DONOR DOB: [redacted] AGE: [redacted] Gender: M Phone: NG Patient ID: 4869[redacted] Health ID: [redacted]	Specimen: [redacted] Requisition: [redacted] Lab Ref #: 4869-[redacted] Collected: 10/24/2014 Received: 10/25/2014 / 01:45 CDT Reported: 10/27/2014 / 12:30 CDT	Client #: 22663146 4195000 [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.14		4.20-5.80 Million/uL	CB
HEMOGLOBIN	15.1		13.2-17.1 g/dL	
HEMATOCRIT	43.6		38.5-50.0 %	
MCV	84.7		80.0-100.0 fL	
MCH	29.3		27.0-33.0 pg	
RDW	14.3		11.0-15.0 %	
HEMOGLOBIN A	97.5		>96.0 %	CB
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.5		1.8-3.5 %	
INTERPRETATION				
	Normal phenotype.			
	Normal hemoglobin distribution, no HgS, HgC or other abnormal hemoglobin observed.			

CHD 4/4/14

11-4-14

PERFORMING SITE:

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

Integrated GENETICS

Tay-Sachs Enzyme Analysis

Patient Name: 4869, Donor
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** 606452
Patient ID: [redacted]

[redacted]

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

Date Collected: 10/30/2014
Date Received: 10/31/2014
Lab ID: 4869-[redacted]
Hospital ID:
Specimen Type: **White Blood Cells**

ENTERED 6~11/6/14

RESULTS: **Hexosaminidase Activity : 1286 nmol/mg protein**
Hexosaminidase Percent A: 55.2

		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: 11/06/2014

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