Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4599

Genetic Testing Summary

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

Last Updated: 04/01/24

Donor Reported Ancestry: German, Norwegian, English 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
Special Testing		
Gene: PEX1	Negative by genotyping for 3 mutations	

*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]

Report Date: 05/16/2013

Male

Name: DONOR 4599
DOB: [redacted]
Ethnicity: Northern European
Sample Type: OG-510 Saliva
Date of Collection: 05/13/2013
Date Received: 05/14/2013
Barcode: [redacted]
Indication: Egg or Sperm Donor

Female

Not tested

Counsyl Test Results Summary (Egg or Sperm Donor)

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses copy number analysis and targeted genotyping as described in the methods section on page 2 to determine carrier status associated with **3 diseases**. Please refer to page 3 for a complete list of diseases and genes included in this panel.

DONOR 4599

DONOR 4599'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

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

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SEP 13 2013

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Methods and Limitations

DONOR 4599: 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. 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 Director:

Nyunsook Kang

H. Peter Kang, MD

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SEP 13 2013

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Counsyl

Male

Name: DONOR 4599
DOB: [redacted]

Female

Not tested

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Diseases 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:** Northern European 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:** Northern European 83%.

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

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Male

Name: DONOR 4599
DOB: [redacted]

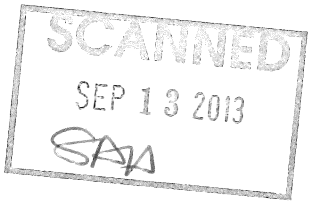
Female

Not tested

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 4599 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



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Integrated GENETICS
LabCorp Specialty Testing Group

Mutation-Specific Sequence Analysis

Patient Name: Donor 4599
Referring Physician: Harvey Stern, MD
Specimen #: [redacted] Client #: [redacted]
Patient ID#: [redacted] Case#: [redacted]

DOB: [redacted]
Sex: M

Date Collected: 9/12/2014
Date Received: [redacted]2014
Lab ID #: 4599-[redacted]
Hospital ID #:
Specimen Type: Peripheral Blood

[redacted]

ENTERED
CA 9/23/14

Ethnicity: Caucasian

Disease (Gene):	Zellweger Syndrome Spectrum, PEX1-Related, includes Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, and Zellweger Syndrome (PEX1)
Indication:	Gamete donor
Result:	Negative for the mutations analyzed
Interpretation:	These results reduce, but do not eliminate, the chance to be a carrier.
Mutation(s) Requested:	c.2097dupT (p.I700fs), c.2528G>A (p.G843D), c.2916delA (p.G973fs)

Unless otherwise noted, all interpretations are based on a negative family history and the absence of symptoms. These results may need further interpretations depending on the clinical presentation.

COMMENTS:

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of this result, as well as recommendations for testing other family members and, when applicable, this individual's partner.

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disease(s) tested. This analysis does not rule out the presence of disease-causing mutations in other regions of the gene(s) analyzed or in other genes, and does not rule out the presence of large deletion or duplication mutations or complex rearrangements.

METHOD/LIMITATIONS:

DNA is isolated and specific gene regions amplified by the polymerase chain reaction (PCR). Targeted mutations are detected by direct DNA sequencing using capillary gel electrophoresis and fluorescence detection. False positive or negative results may occur for reasons that include: genetic variants, technical handling, blood transfusions, bone marrow transplantation, mislabeling of samples, erroneous representation of family relationships, or contamination of a fetal sample with maternal cells.

This test was developed and its performance characteristics determined by Esoterix Genetic Laboratories, LLC. 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 and must be used in conjunction with clinical assessment, when available. It should not be regarded as investigational or for research.

Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

Signed by: Jane Thuo Ph.D., FACMG Jane Thuo Date: 9/23/2014

Testing Performed at Esoterix Genetic Laboratories, LLC 3400 Computer Drive Westborough, MA 01581
Bernice A. Allitto, PhD, FACMG, Laboratory Director 1-800-255-7357

GENETICS & IVF
Institute

CW 6/3/13

Cytogenetic Report

Client [redacted]

Address [redacted]

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

Patient name/Donor Alias Donor # 4599 **Patient DOB** N/A

Donor # 4599[redacted] **Specimen type** Peripheral Blood

Collection Date 05/13/2013 **Accession #** [redacted]

Date Received 05/14/2013

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

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

5/28/13
Date

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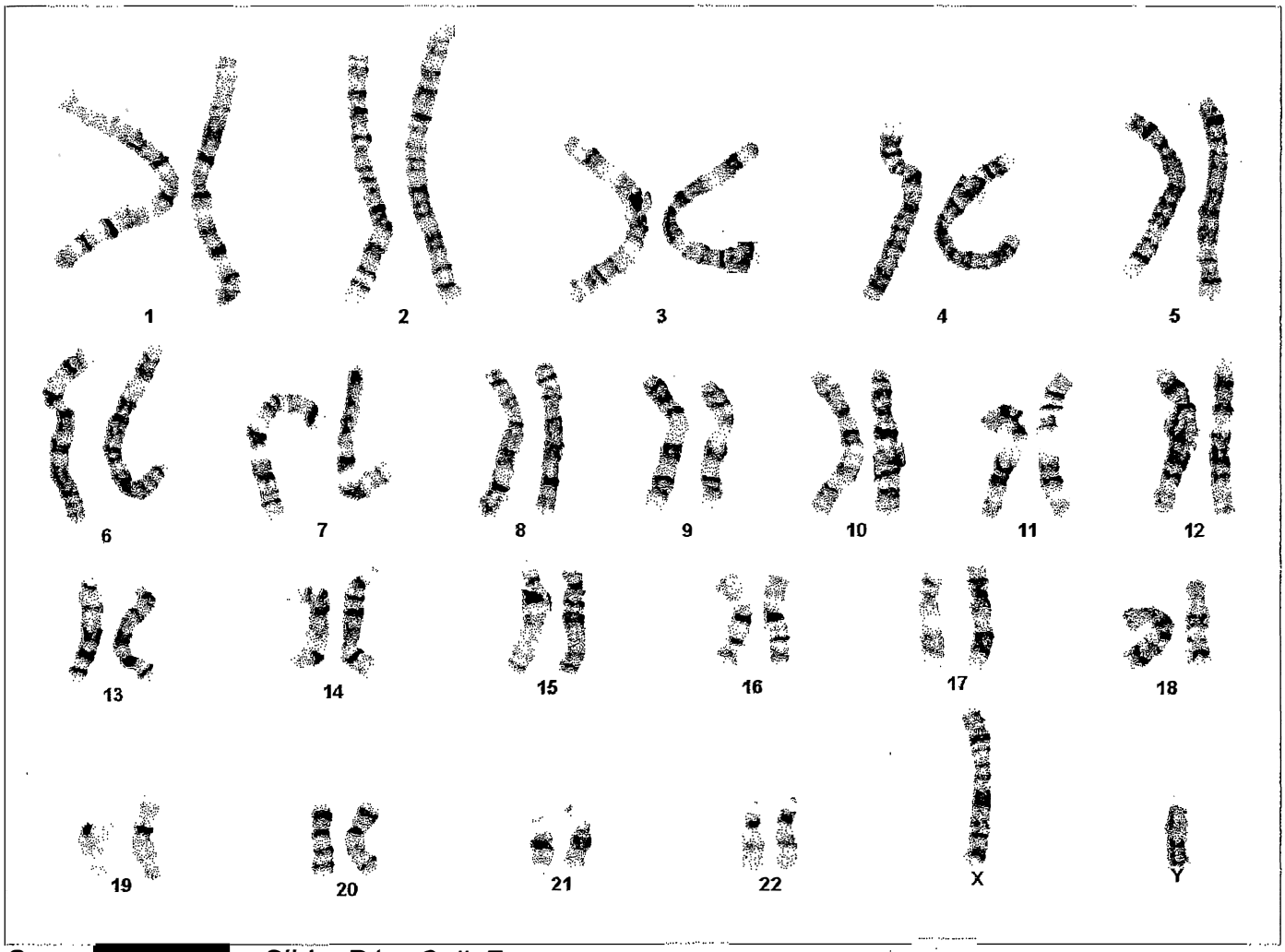
Genetics a. IVF Preimplantation Genetics Lat. atory

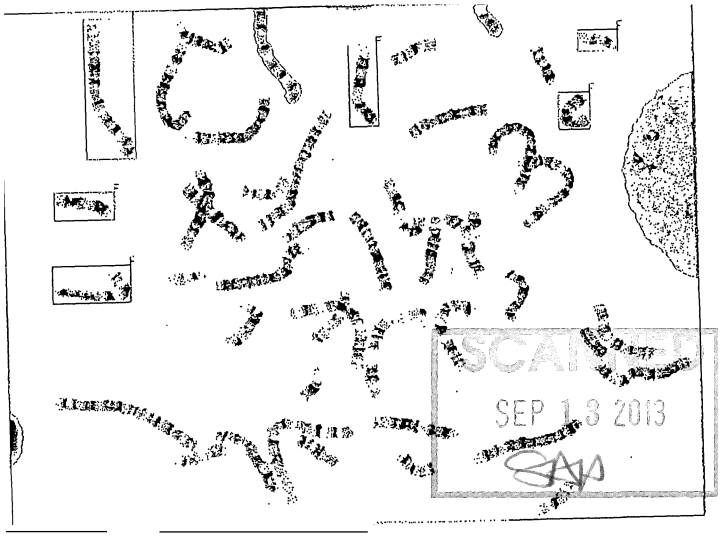
Patient name: DONOR # 4599

Case name: [redacted]

46,XY



Case: [redacted] Slide: B1 Cell: 7



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Quest Diagnostics

Report Status: Final

ID 4599, DONOR

Patient Information	Specimen Information	Client Information
ID 4599, DONOR [redacted] M Patient ID: [redacted] Health ID: [redacted]	Specimen: [redacted] Collected: 05/13/2013 Received: 05/14/2013 / 01:18 CDT Reported: 05/15/2013 / 16:01 CDT	[redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.76		4.20-5.80 Million/uL	CB
HEMOGLOBIN	14.1		13.2-17.1 g/dL	
HEMATOCRIT	42.5		38.5-50.0 %	
MCV	89.2		80.0-100.0 fL	
MCH	29.6		27.0-33.0 pg	
RDW	13.1		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.

ENTERED 6/4/3/13

PERFORMING SITE:

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

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