Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 2893

Genetic Testing Summary

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

Last Updated: 03/26/24

Donor Reported Ancestry: Russian, Armenian Jewish Ancestry: Yes

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 108 mutations in the CFTR gene	1/270
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/350
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative for 37 mutations tested by genotyping in the HBB gene	<1/500 Beta Thalassemia <1/500 Sickle Cell Disease
ABCC8-Related Hyperinsulinism	Negative for 3 mutations in the ABCC8 gene	1/110
Bloom Syndrome	Negative for 2 mutations in the BLM gene	<1/500
Canavan Disease	Negative for 4 mutations in the ASPA gene	<1/500
Familial Dysautonomia	Negative for 3 mutations in the IKBKAP gene	<1/500
Fanconi Anemia Type C	Negative for 4 mutations in the FANCC gene	1/400

Gaucher Disease	Negative for 9 mutations in the GBA gene	1/390
Glycogen Storage Disease Type 1 A	Negative for 10 mutations in the G6PC gene	<1/500
Tay Sachs Disease	Negative for 4 mutations in the HEXA gene	<1/500
Lipoamide Dehydrogenase Deficiency	Negative for 2 mutations in the DLD gene	<1/500
Maple Syrup Urine Disease Type 1B	Negative for 3 mutations in the BCKDHB gene	1/420
Mucolipidosis IV	Negative for 2 mutations in the MCOLN1 gene	<1/500
Niemann-Pick Disease, SMPD1-Associated	Negative for 3 mutations in the SMPD1 gene	1/260
Usher Syndrome Type 1F	Negative for 1 mutations in the PCDH15 gene	1/200
Usher Syndrome Type 3	Negative for 1 mutations in the CLRN1 gene	<1/500
Special Testing		
Genes: CFTR, MEFV, MECR	Negative by gene sequencing	See attached results

*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]

Report Date: 12/06/2010

Ordering Healthcare Professional

Fairfax Cryobank [redacted]

Male Details

[redacted]

Name: Donor 2893

[redacted]

Ethnicity: Mixed or Other Caucasian
Sample Type: Saliva (OG-300)
Date of Collection: 11/19/2010
Barcode: [redacted]
Indication: Egg or Sperm Donor

Universal Genetic Test (Egg or Sperm Donor)

The Universal Genetic Test uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for a number of Mendelian 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 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 2893

Donor 2893's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The child risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Child Risk Summary

Your Universal Genetic Test indicates that your future children have a reduced risk for the diseases tested, including those listed below which are common in your ethnicity.

Cystic Fibrosis

Spinal Muscular Atrophy

* **Limitations:** In an unknown number of cases, nearby genetic variants may interfere with mutation detection. The child risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation. Individuals of African, Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and should also be offered carrier testing by CBC and hemoglobin electrophoresis or HPLC.

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.

Laboratory Director: Jessica Jacobson, MD
CLIA Number: 05D1102604

2200 Bridge Parkway, Suite 103, Redwood City, CA 94065
(888) COUNSYL | http://www.counsyl.com

Counsyl

Male
Name: Donor 2893
DOB: [redacted]

Female
Not tested

Full Results

Below are the full test results for all diseases on the panel. Noted are the specific genetic mutations for which the patient tested positive or negative. If there was insufficient data to determine the genotype for any variant, this will be noted as "no call." Also 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.

ABCC8-Related Hyperinsulinism

Your child's risk: 1 in 51,000 | **Risk before testing:** 1 in 50,000 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 110. <10% detection rate.

Gene: ABCC8. **Variants (3):** F1388del, V187D, 3992-9G>A.

Beta Thalassemia

Your child's risk: Less than 1 in 1,000,000 | **Risk before testing:** 1 in 250,000 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 80% detection rate.

Gene: HBB. **Variants (35):** K17X, Q39X, 619 bp deletion, 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), IVS-I-1(G>T), -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, Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, Pro5fs, Gly16fs, Glu6fs, IVS-II-705, IVS-II-844, -30T>A, CAP+1 A>C, Hb E, Hb O-Arab.

Bloom Syndrome

Your child's risk: Less than 1 in 1,000,000 | **Risk before testing:** less than 1 in 1,000,000 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. <10% detection rate.

Gene: BLM. **Variants (2):** 2281del6ins7, 2407insT.

Canavan Disease

Your child's risk: Less than 1 in 1,000,000 | **Risk before testing:** less than 1 in 1,000,000 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 53% detection rate.

Gene: ASPA. **Variants (4):** E285A, Y231X, A305E, IVS2-2A>G.

Cystic Fibrosis

Your child's risk: 1 in 30,000 | **Risk before testing:** 1 in 3,100 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 270. 90% detection rate.

Gene: CFTR. **Variants (108):** 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, S1235R, 394delTT, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), G91R, R117C, I148T, L206W, G330X, T338I, R352Q, S364P, G480C, I506V, F508C, C524X, S549I, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, Q890X, R1066C, R1070Q, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, R1283M, dele2-3 21kb, 3199del6, F311del, 574delA, 663delT, 935delA, 936delTA, 1161delC, 1609delCA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2105-2117del13insAGAAA, 3171delC, 3667del4, 3821delT, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1811+1.6kbA>G, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, W1204X(c.3612G>A).

Familial Dysautonomia

Your child's risk: Less than 1 in 1,000,000 | **Risk before testing:** less than 1 in 1,000,000 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. <10% detection rate.

Gene: IKBKAP. **Variants (3):** IVS20+6T>C, R696P, P914L.

Fanconi Anemia Type C

Your child's risk: 1 in 250,000 | **Risk before testing:** 1 in 100,000 | Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 400. 60% detection rate.

Gene: FANCC. **Variants (4):** IVS4+4A>T, 322delG, Q13X, R548X.

Counsyl

Male	Female
Name: Donor 2893 DOB: [redacted]	Not tested

Gaucher Disease

Your child's risk:	Risk before testing:	
1 in 170,000	1 in 50,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 390. 71% detection rate.

Gene: GBA. **Variants (9):** N370S, L444P, 1035insG, IVS2+1G>A, V394L, R496H, D409V, R463C, R463H.

Glycogen Storage Disease Type Ia

Your child's risk:	Risk before testing:	
1 in 520,000	1 in 130,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 76% detection rate.

Gene: G6PC. **Variants (10):** R83C, Q347X, 727G>T, F327del, Q27fsdelC, 459insTA, R83H, G188R, Q242X, G270V.

Maple Syrup Urine Disease Type 1B

Your child's risk:	Risk before testing:	
1 in 420,000	1 in 250,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 420. 40% detection rate.

Gene: BCKDHB. **Variants (3):** R183P, G278S, E322X.

Maple Syrup Urine Disease Type 3

Your child's risk:	Risk before testing:	
Less than 1 in 1,000,000	less than 1 in 1,000,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. <10% detection rate.

Gene: DLD. **Variants (2):** 105insA, G229C.

Mucolipidosis IV

Your child's risk:	Risk before testing:	
Less than 1 in 1,000,000	less than 1 in 1,000,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. <10% detection rate.

Gene: MCOLN1. **Variants (2):** 511_6944del, IVS3-2A>G.

Niemann-Pick Disease Type A

Your child's risk:	Risk before testing:	
1 in 260,000	1 in 250,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 260. <10% detection rate.

Gene: SMPD1. **Variants (3):** fsP330, L302P, R496L.

Sickle Cell Disease

Your child's risk:	Risk before testing:	
Less than 1 in 1,000,000	less than 1 in 1,000,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. >99% detection rate.

Gene: HBB. **Variants (37):** Hb S, K17X, Q39X, 619 bp deletion, 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), IVS-I-1(G>T), -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, Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, Pro5fs, Gly16fs, Glu6fs, IVS-II-705, IVS-II-844, -30T>A, CAP+1 A>C, Hb E, Hb D-Punjab, Hb O-Arab.

Spinal Muscular Atrophy

Your child's risk:	Risk before testing:	
1 in 97,000	1 in 4,800	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 95% detection rate.

Gene: SMN1. **Variants (1):** Exon 7 deletion.

Tay-Sachs Disease

Your child's risk:	Risk before testing:	
1 in 690,000	1 in 360,000	Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. 48% detection rate.

Gene: HEXA. **Variants (4):** 1278insTATC, IVS12+1G>C, IVS9+1G>A, IVS7+1G>A.

Counsyl

Male
Name: Donor 2893
DOB: [redacted]

Female
Not tested

Usher Syndrome Type 1F

Your child's risk: 1 in 160,000
Risk before testing: 1 in 150,000
Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is 1 in 200. <10% detection rate.

Gene: PCDH15. **Variants (1):** R245X.

Usher Syndrome Type 3

Your child's risk: Less than 1 in 1,000,000
Risk before testing: less than 1 in 1,000,000
Reduced risk

Donor 2893: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier, assuming a negative family history, is < 1 in 500. <10% detection rate.

Gene: CLRN1. **Variants (1):** N48K.

Laboratory Director: Jessica Jacobson, MD
CLIA Number: 05D1102604

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genzyme

SMN1 Copy N[redacted]nber Analysis

Patient Name: . 2893
DOB: Age:
SSN #: Gender: Male

606452 / 366220
Fairfax Cryobank[redacted] Institute
[redacted]

Genzyme Specimen [redacted]
Case #[redacted] Patient ID #[redacted]2
Date Collected: 05/10/2010 Date Received: 05/11/2010

Referring Physician: [redacted]
Genetic Counselor:

Client Lab ID #: [redacted]
Hospital ID #:
Specimen ID #:

Specimen Type: Peripheral Blood

Specimen(s) Received: 2 - Yellow (ACD) 10 ml round bottom tube(s)

Clinical Data: Carrier Test/Gamete donor

Ethnicity: Armenian

RESULTS: SMN1 copy number: 2 (Reduced Carrier Risk)

INTERPRETATION:

This individual has an SMN1 copy number of two. This result reduces but does not eliminate the risk to be a carrier of SMA. Ethnic specific risk reductions based on a negative family history and an SMN1 copy number of two are provided in the Comments section of this report.

COMMENT:

Spinal muscular atrophy (SMA) is an autosomal recessive disease of variable age of onset and severity caused by mutations (most often deletions or gene conversions) in the survival motor neuron (SMN1) gene. Molecular testing assesses the number of copies of the SMN1 gene. Individuals with one copy of the SMN1 gene are predicted to be carriers of SMA. Individuals with two or more copies have a reduced risk to be carriers. (Affected individuals have 0 copies of the SMN1 gene.)

This copy number analysis cannot detect individuals who are carriers of SMA as a result of either 2 (or very rarely 3) copies of the SMN1 gene on one chromosome and the absence of the SMN1 gene on the other chromosome or small intragenic mutations within the SMN1 gene. This analysis also will not detect germline mosaicism or mutations in genes other than SMN1. Additionally, de novo mutations have been reported in approximately 2% of SMA patients.

Carrier Frequency and Risk Reductions for Individuals with No Family History of SMA				
Ethnicity	**Detection Rate[1]**	**A priori Carrier Risk[1]**	**Reduced Carrier Risk for 2 copy result**	**Reduced Carrier Risk for 3 copy result**
Caucasian	94.9%	1:35	1:632	1:3,500
Ashkenazi Jewish	90.2%	1:41	1:350	1:4,000
Asian	92.6%	1:53	1:628	1:5,000
Hispanic	90.6%	1:117	1:1061	1:11,000
African American	71.1%	1:66	1:121	1:3,000
Mixed Ethnicities	For counseling purposes, consider using the ethnic background with the most conservative risk estimates.			

METHOD/LIMITATIONS:
Specimen DNA is isolated and amplified by real-time polymerase chain reaction (PCR) for exon 7 of the SMN1 gene and two reference genes. A mathematical algorithm is used to calculate the number of copies of SMN1. Sequencing of the primer and probe binding sites for the SMN1 real-time PCR reaction is performed on all fetal samples, and on samples from individuals with 1 copy of SMN1 on carrier testing, to rule out the presence of sequence variants which could interfere with analysis and interpretation. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

REFERENCES:
1. Carrier frequency and detection rate are calculated based on analysis of allele frequencies among > 1000 individuals from each ethnic group noted (Genzyme Genetics, data submitted for publication). 2. Online review of SMA: http://www.genereviews.org/profiles/sma

The test was developed and its performance characteristics have been determined by Genzyme. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjunction with clinical assessment, when available.

Electronically Signed by: Hui Zhu, Ph.D. FACMG, on 05/20/2010
Reported by: JH/jh

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted] **Fax #** [redacted] **Email** N/A

Patient name/Donor Alias Donor 2893 **Patient DOB** N/A

Donor # 2893-100510 **Specimen type** Peripheral blood

Collection Date 05/10/2010 **Accession #** 10-046CG

Date Received 05/11/2010

RESULTS

CYTOGENETIC ANALYSIS

Cells counted 20

Cells analyzed 5

Cells karyotyped 3

Modal chromosome # 46

Type of banding GTG

Band resolution 500

FISH

Probe(s) N/A

Nuclei scored N/A

KARYOTYPE 46,XY

INTERPRETATION

Normal male karyotype
No 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

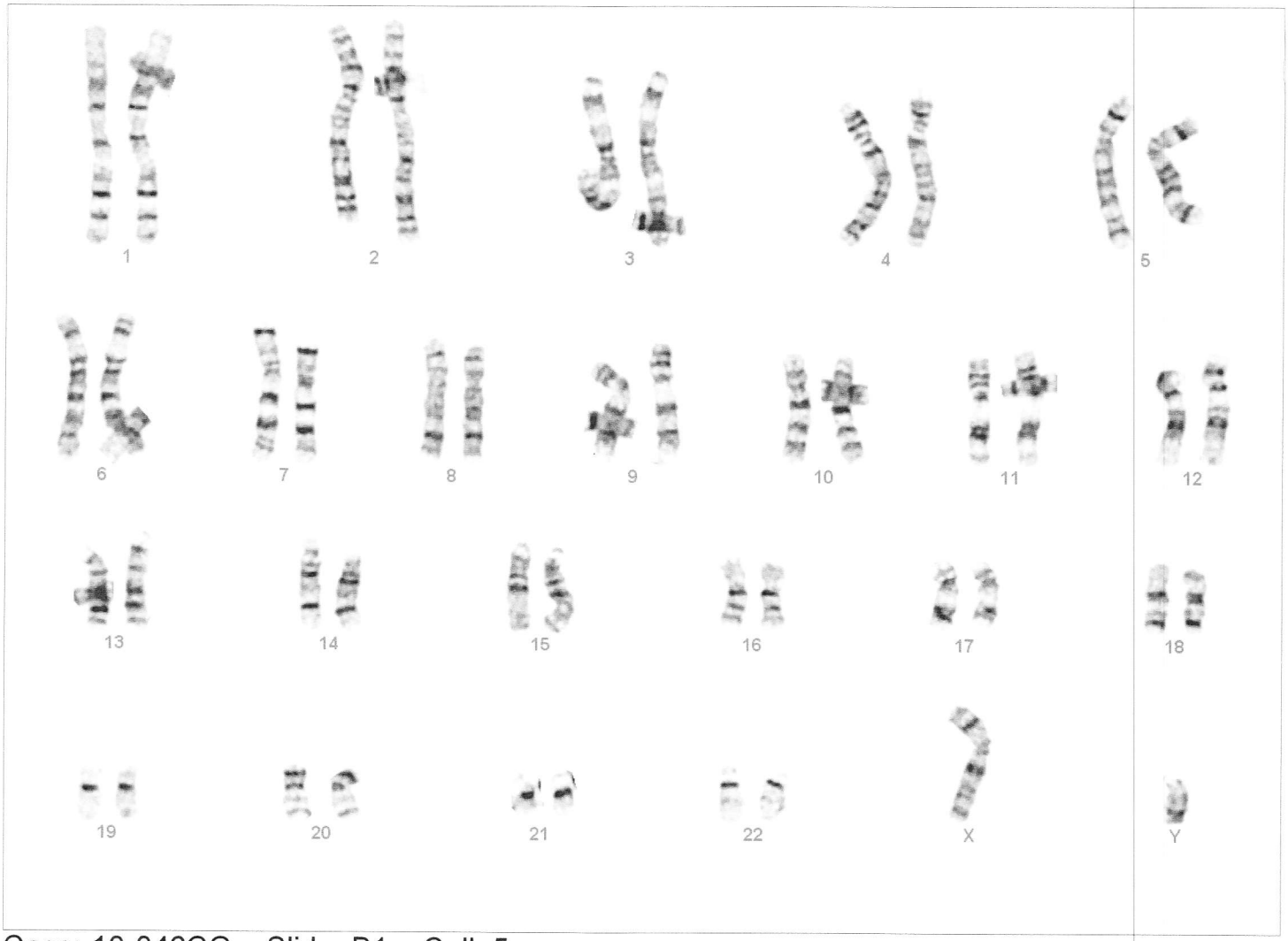
5/24/10
Date

Genetics and IVF Preimplantation Genetics Laboratory

Patient name: DONOR 2893

Case name: 10-046CG

46,XY



Case: 10-046CG Slide: B1 Cell: 5

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info@fulgentgenetics.com
www.fulgentgenetics.com

[redacted]

Patient Information:
2893, Donor
DOB: [redacted]
Sex: M
MR#: 2893
Patient#: FT-[redacted]

Partner Information:
Not Tested

Physician:
Seitz, Suzanne
ATTN: Seitz, Suzanne
Fairfax Cryobank
3015 Williams Drive
Fairfax, VA 22031

Laboratory:
Fulgent Therapeutics, LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Mar 20,2024**

Accession:
[redacted]
Test#: [redacted]
Specimen Type: DNA
Collected: Mar 04,2024

Accession:
N/A

FINAL RESULTS

No carrier mutations identified

TEST PERFORMED

Custom Beacon Carrier Screening Panel

(2 Gene Panel: *CFTR and MEFV*; gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- No carrier mutations were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; https://www.nsgc.org)

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GENES TESTED:

Custom Beacon Carrier Screening Panel - 2 Genes

This analysis was run using the Custom Beacon Carrier Screening Panel gene list. 2 genes were tested with 100.0% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

CFTR, MEFV

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution

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of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes and Limitations

CFTR: Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. CFTR variants primarily associated with CFTR-related isolated congenital bilateral absence of the vas deferens and CFTR-related pancreatitis are not included in this analysis. CFTR variants with insufficient evidence of being cystic fibrosis mutations will not be reported either.

SIGNATURE:

Yan Meng, Ph.D., CGMB, FACMG on **3/20/2024**
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed and its performance characteristics determined by **Fulgent Therapeutics, LLC**. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or **info@fulgentgenetics.com**. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

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[redacted]

To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes on this test please visit the following link:
Beacon Expanded Carrier Screening Supplemental Table

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[redacted]

Patient Information:
2893, Donor
DOB: [redacted]
Sex: M
MR#: 2893
Patient#: [redacted]

Accession:
[redacted]
Test#: [redacted]
Order#: [redacted]
Ext Test#: [redacted]
Ext Order#: [redacted]
Specimen Type: DNA
Collected: Mar 04,2024
Received Date: Mar 12,2024
Authorized Date: Mar 14,2024

Physician:
Seitz, Suzanne
ATTN: Seitz, Suzanne
Fairfax Cryobank
3015 Williams Drive
Fairfax, VA 22031
Phone:
Fax:

Laboratory:
Fulgent Therapeutics, LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Mar 25,2024**

Final Report

TEST PERFORMED

MECR Single Gene
(1 Gene Panel: *MECR*; gene sequencing with deletion and duplication analysis)

RESULTS:

No clinically significant sequence or copy-number variants were identified in the submitted specimen.

A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations of the sort not queried by this test or in areas not reliably assessed by this test.

INTERPRETATION:

Notes and Recommendations:

- As requested, this report only includes variants classified as Pathogenic, Likely Pathogenic, or Risk Allele at the time of analysis. If detected, this report does not include variants classified as of uncertain significance.
- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; https://www.nsgc.org)
- Guide to Interpreting Genomic Reports: A Genomics Toolkit (CSER Consortium; February 2017) (https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources#hep)

GENES TESTED:

MECR Single Gene
1 genes tested (100.00% at >20x).

MECR

Gene Specific Notes and Limitations

No gene specific limitations apply to the genes on the tested panel.

METHODS:

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Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. New York patients: diagnostic findings are confirmed by Sanger, MLPA, or qPCR; exception SNV variants in genes for which confirmation of NGS results has been performed >=10 times may not be confirmed if identified with high quality by NGS. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to this individual's phenotype, and negative results do not rule out a genetic cause for the indication for testing. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is designed and validated for detection of germline variants only. It is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions (eg. trinucleotide or hexanucleotide repeat expansion). DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm for copy number variants, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which are two or more contiguous exons in size; single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

SIGNATURE:

Yan Meng, Ph.D., CGMB, FACMG on **3/25/2024**
Laboratory Director, Fulgent

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