Donor 4835

Genetic Testing Summary

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

Last Updated: 04/13/22

Donor Reported Ancestry: Irish, German, Swiss 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 for 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 Globin Chain Hemoglobinopathy	Negative for 28 mutations in the HBB gene	1/290
Tay Sachs Enzyme Analysis	Non-Carrier by Hexosaminidase A testing	
Special Testing		
Non-syndromic Hearing Loss (GJB2)	Negative for 14 mutations in the GJB2 gene	Residual risk not provided

*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.

GENETICS & IVF
Institute

Cytogenetic Report

Client [redacted]

Address [redacted]

ENTERED 1/7/14

Reporting Phone # [redacted]

Patient name/Donor Alias	Donor # 4835	**Patient DOB**	N/A
Donor #	4835-131220	**Specimen type**	Peripheral Blood
Collection Date	12/23/2013	**Accession #**	13-198CG
Date Received	12/24/2013		

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	21	**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

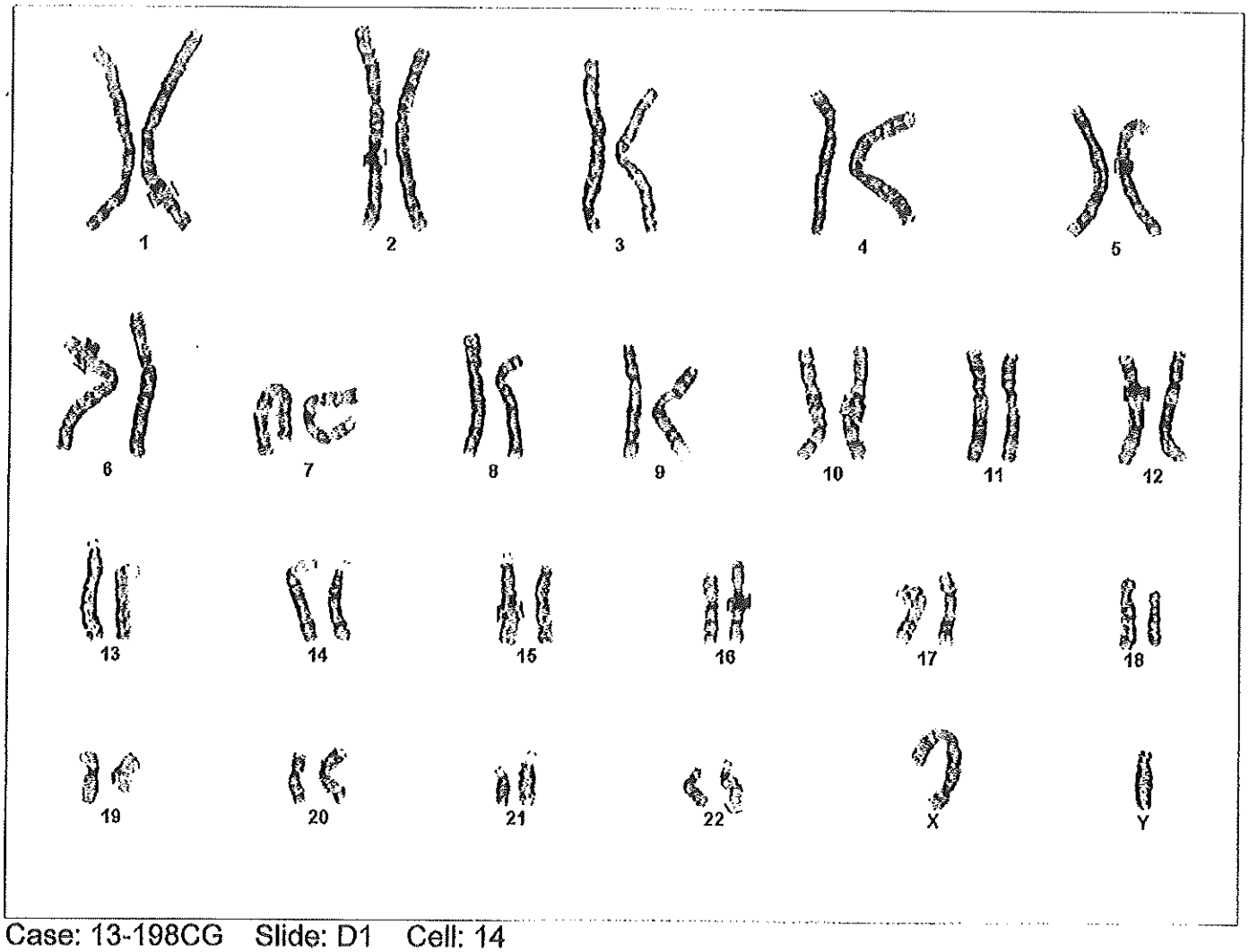
1/6/14
Date

Genetics and IVF Preimplantation Genetics Laboratory

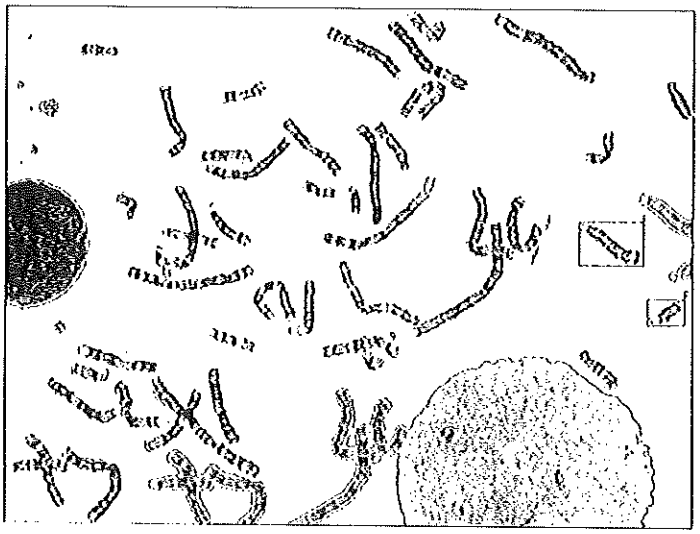
Patient name: DONOR # 4835

Case name: 13-198CG

46,XY



Case: 13-198CG Slide: D1 Cell: 14



Counsyl

Results Recipient
[redacted]
Attn: Dr. Harvey Stern
[redacted]
Report Date: 12/28/2013

Male
Name: DONOR 4835
DOB: [redacted]
Ethnicity: Northern European
Sample Type: OG-510 Saliva
Date of Collection: 12/23/2013
Date Received: 12/26/2013
Barcode: [redacted]
Indication: No family history (screening)
Test Type: The Counsyl Test

Female
Not tested

Counsyl Test Results Summary

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses targeted genotyping and copy number analysis 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 4835

DONOR 4835'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/.

Methods and Limitations

DONOR 4835: The Counsyl Test - 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 Directors:

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Jelena Brezo

Jelena Brezo, PhD, FACMG

Counsyl

Male	Female
Name: DONOR 4835 DOB: [redacted]	Not tested

ENTERED 6/17/14

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%.

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 4835 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

Integrated GENETICS
LabCorp Specialty Testing Group

Tay-Sachs Enzyme Analysis

Patient Name: 4835, Donor
Referring Physician: Harvey Stern, MD
Specimen #: 21135784 **Client #:** 606452
Patient ID: 15897998-6

[redacted]

DOB: 09/01/1986
SSN: ***-**-****

Date Collected: 12/23/2013
Date Received: 12/24/2013
Lab ID: 4835-131223
Hospital ID:
Specimen Type: **White Blood Cells**

ENTERED 1/7/14

RESULTS: **Hexosaminidase Activity : 1874 nmol/mg protein**
Hexosaminidase Percent A: 57.2

		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, FACC
Stanford Marenberg, Ph.D.

Date: 12/29/2013

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

Recombine

CarrierMap™

A COMPREHENSIVE GENETIC CARRIER SCREEN

Assay performed by Reprogenetics
CLIA ID: 31D1054821

Patient Information

Patient: DONOR 4835
DOB: [redacted]
Gender: Male
Ethnicity: EUROPEAN
Procedure ID: 14931
Report Date: 01/20/2015
Report Updated: 01/23/2015

Sample Information

Specimen Type: Blood
Specimen Number: 16460
[redacted]
Date Specimen Collected: 01/09/2015
Date Specimen Received: 01/12/2015
Date Specimen Analyzed: 01/20/2015

Ordering Practice

Practice Code: 926
Fairfax Cryobank
3015 Williams Drive #110
Fairfax, VA 22031
Physician: [redacted]

Summary of Genetic Testing Results

You did not test positive for any of the mutations assayed.

Disease Groups

High Impact	Treatment Benefits	X-Linked	Moderate Impact
These diseases have a significant impact on life expectancy and quality of life.	Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.	These diseases are passed down by female carriers. Carriers may have symptoms.	These diseases typically do not affect life expectancy but can affect quality of life.

Summary of Testing Performed

Diseases Tested: 1	Mutations Tested: 14	Genes Tested: 1

All other mutations analyzed by Recombine were not detected. This reduces but does not eliminate your chance to be a carrier for the associated genetic diseases. Recombine does not screen for every possible genetic disease.

Learn More

A list of all the genetic diseases and mutations you were screened for is included in the latter pages of this report. For disease information, please visit **www.recombine.com/our-test**. To speak with a Genetic Counselor, call **855.OUR.GENES**

Lab Technician: Bo Chu

Reviewed by: Pere Colls, PhD, HCLD

**Methods and Limitations: Recombine developed this genetic assay using the Illumina Infinium Custom HD Genotyping Assay. The test is intended for clinical preconception and/or prenatal screening purposes and is not validated for detection of homozygous mutations. False positive or negative results may occur for reasons that include: genetic variants, assay limitations, sample mix-up, sample contamination, and molecular and technical errors. Recombine tests for Spinal Muscular Atrophy via an Identity-by-State shared haplotype comparison algorithm. Detection is limited to haplotypes within our library of known carriers of the most common mutation (deletion of Exon 7).*

Diseases & Mutations Assayed

Groups	Disease	#	Mutations
●●○○	Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2)	15	c.167delT, c.235delC, c.312_325delGAAGTTCATCAAGG, c.358delGAG (p.120delE), c.C370T (p.Q124X), c.C427T (p.R143W), c.G231A (p.W77X), c.G551C (p.R184P), c.G71A (p.W24X), c.T101C (p.M34T), c.T229C (p.W77R), c.T269C (p.L90P), c.G109A (p.V37I), c.35delG, c.35G>T