Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4912

Genetic Testing Summary

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

Last Updated: 10/26/23

Donor Reported Ancestry: Danish, Scandinavian, Italian, Irish, Welsh 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
Tay Sachs Enzyme Analysis	Non-carrier by hexosaminidase A testing	
Special Testing		
Genes: BTD, MEFV, GBA, NEB, BCHE, HBA1/HBA2	Non-carrier by genotyping in 2015	

*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	Male	Female
Fairfax Cryobank - [redacted] [redacted] Report Date: 09/30/2013	Name: 4912 [redacted] DOB: [redacted] Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 09/25/2013 Date Received: 09/27/2013 Barcode: [redacted] Indication: Egg or Sperm Donor Test Type: The Counsyl Test	Not tested

Counsyl Test Results Summary (Egg or Sperm Donor)

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.

4912 4912

4912 4912'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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Counsyl

Male	Female
Name: 4912 [redacted]	Not tested
DOB: [redacted]	

Methods and Limitations

4912 4912: 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: 4912[redacted]	Not tested
DOB: [redacted]	

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%.

Male	Female
Name: 4912 [redacted] DOB: [redacted]	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	4912 4912 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

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted]7 Fax # [redacted] Email [redacted]

Patient name/Donor Alias Donor # 4912 Patient DOB N/A

Donor # 4912[redacted] Specimen type Peripheral Blood

Collection Date 09/25/2013 Accession # [redacted]

Date Received 09/26/2013

RESULTS

CYTOGENETIC ANALYSIS

Cells counted 21

Cells analyzed 5

Cells karyotyped 2

Modal chromosome # 46

Type of banding GTG

Band resolution 550

FISH

Probe(s) N/A

Nuclei scored N/A

KARYOTYPE 46,XY

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

10/10/13
Date

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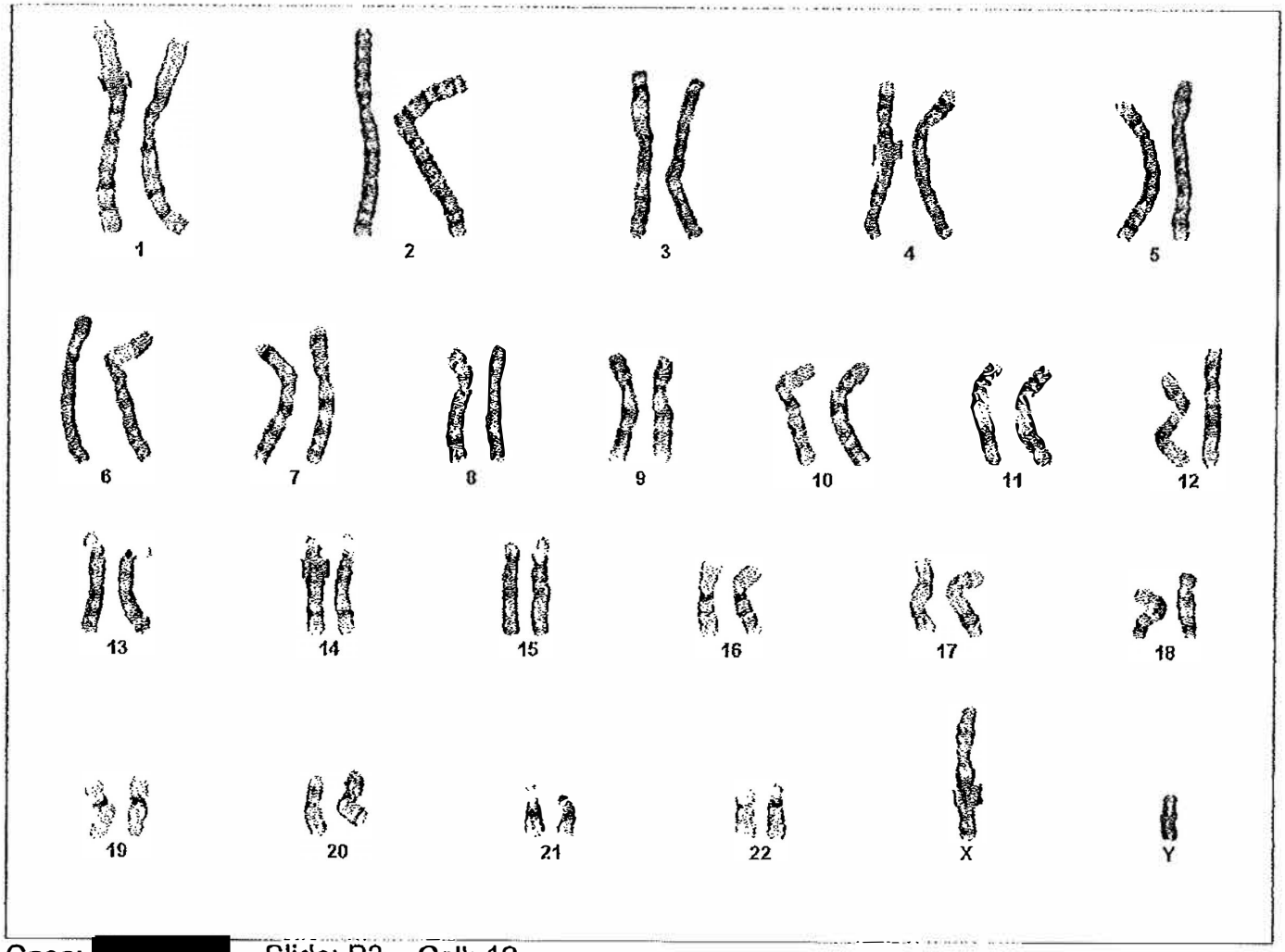
Genetics and IVF Preimplantation Genetics Laboratory

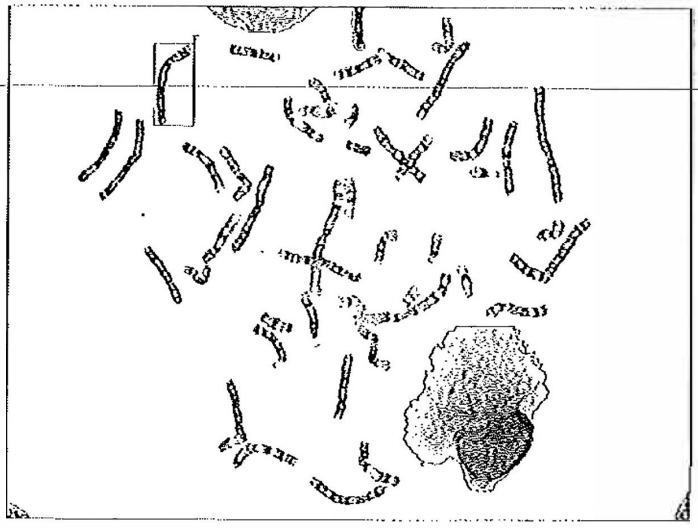
Patient name: DONOR # 4912

Case name: [redacted]

46,XY



Case: [redacted] Slide: B3 Cell: 12



Integrated GENETICS
LabCorp Specialty Testing Group

Tay-Sachs Enzyme Analysis

Patient Name: 4912, .
Referring Physician: [redacted]
Specimen #: 1[redacted] **Client #:** [redacted]
Patient ID: [redacted]

Fairfax Cryobank [redacted]

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

Date Collected: 09/25/2013
Date Received: 09/26/2013
Lab ID: 4912[redacted]
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 1194 nmol/mg protein**
Hexosaminidase Percent A: 61.2

		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.

ENTERED 10-23-13

Under the direction of: Stanford Marenberg, PHD, ABCC

Stanford Marenberg, Ph.D.

Date: 09/30/2013

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

Report Status: Final

ID, 4912

Patient Information	Specimen Information	Client Information
ID, 4912 **DOB: Not Given** **AGE: Not Given** Gender: M Fasting: N Phone: NG Patient ID: NG	Specimen: [redacted] Requisition: [redacted] Collected: 09/25/2013 / 10:00 EDT Received: 09/25/2013 / 20:46 EDT Reported: 09/27/2013 / 07:31 EDT	Client #[redacted] HO130000 [redacted] FAIRFAX CRYO BANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	4.81		4.20-5.80 Million/uL	QHO
HEMOGLOBIN	15.0		13.2-17.1 g/dL	
HEMATOCRIT	44.9		38.5-50.0 %	
MCV	93.3		80.0-100.0 fL	
MCH	31.2		27.0-33.0 pg	
RDW	12.9		11.0-15.0 %	
HEMOGLOBIN A	96.5		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.5		1.8-3.5 %	
INTERPRETATION				

Normal phenotype.

PERFORMING SITE:

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

ENTERED 10.23.13

Recombine

CarrierMap™

Ordering Practice	Donor 4912 Donor 4912	Partner Not Tested
Practice Code: 926 Fairfax Cryobank [redacted] Physician: Suzanne Seitz Report Generated: 04/22/2015	DOB [redacted] Gender: Male Ethnicity: European Procedure ID: 19358 Kit Barcode: [redacted] Method: Genotyping Specimen: Blood, #20818 Specimen Collection: 04/09/2015 Specimen Received: 04/10/2015 Specimen Analyzed: 04/22/2015	

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4912 Donor 4912 was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

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

♂ Male

Panel: Fairfax 6 Panel Disease , Diseases Tested: 6, Mutations Tested: 45, Genes Tested: 7, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31D1054821
Lab Technician: Bo Chu

Reviewed by: Pere Colls, PhD, HCLD, Lab Director

[redacted]
WP 04.27.15

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in >200 genes. 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.

Diseases & Mutations Assayed

High Impact | Treatment Benefits | X-Linked | Moderate Impact

H	T	X	M	Disease	#	Mutations
●	○	○	○	Alpha Thalassemia	15	♂ Genotyping | SEA deletion, THAI deletion, 11.1kb deletion, c.207C>A (p.N69K), c.223G>C (p.D75G), c.-5092_95+60del5247bp (Alpha 5.2), c.2T>C (p.M1T), c.94_95delAG (p.R32DfsX24), c.207C>G (p.N69K), c.339C>G (p.H113Q), c.340_351delCTCCCCGCCGAG (p.L114_ E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G, c.175C>T (p.His59Tyr)
●	●	○	○	Biotinidase Deficiency	10	♂ Genotyping | c.98_104delGCGGCTGinsTCC (p.C33FfsX68), c.A1368C (p.Q456H), c.A755G (p.D252G), c.C1612T (p.R538C), c.C235T (p.R79C), c.G100A (p.G34S), c.G1330C (p.D444H), c.G511A (p.A171T), c.T1207G (p.F403V), c.A1466C (p.N489T)
●	●	○	○	Familial Mediterranean Fever	12	♂ Genotyping | c.2076_2078delAAT (p.692dell), c.A2080G (p.M694V), c.A2084G (p.K695R), c.C1437G (p.F479L), c.C800T (p.T267I), c.G1958A (p.R653H), c.G2040A (p.M680I), c.G2040C (p.M680I), c.G2082A (p.M694I), c.G2230T (p.A744S), c.G2282A (p.R761H), c.T2177C (p.V726A)
●	●	○	○	Gaucher Disease	6	♂ Genotyping | c.84_85insG, c.A1226G (p.N409S), c.A1343T (p.D448V), c.C1504T (p.R502C), c.G1297T (p.V433L), c.G1604A (p.R535H)
●	○	○	○	Nemaline Myopathy: NEB Related	1	♂ Genotyping | c.7434_7536del2502bp
○	●	○	●	Pseudocholinesterase Deficiency	1	♂ Genotyping | c.A293G (p.D98G)