Donor 4210

Genetic Testing Summary

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

Last Updated: 03/21/22

Donor Reported Ancestry: English, Dutch, German, Native American 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 108 variants in the CFTR gene	1/270
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	< 1/500
Sickle Cell Disease and Beta Thalassemia (HBB)	Negative for 37 variants in the HBB gene	Sickle Cell Disease: <1/500 Beta Thalassemia <1/500
Special Testing		
Oculocutaneous Albinism Type 1	Negative for 27 variants in the TYR gene	1/137 (26% detection rate)

*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	Ordering Healthcare Professional	Male Details	Female Details
Fairfax Cryobank [redacted] [redacted] Report Date: 02/22/2011	Fairfax Cryobank - [redacted] [redacted] NPI: 1417048786	Name: Donor 4210 [redacted] Ethnicity: Mixed or Other Caucasian Sample Type: Saliva (OG-300) Date of Collection: 02/14/2011 [redacted] Indication: Egg or Sperm Donor	Not tested

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 4210

Donor 4210'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

3/7/11

RECEIVED 3/7/11

*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

Counsyl

Male
Name: Donor 4210
[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.

Beta Thalassemia

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

Donor 4210: 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.

Cystic Fibrosis

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

Donor 4210: No mutations detected. No call for 3199del6. 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).

Sickle Cell 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 4210: 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: 1 in 97,000
Risk before testing: 1 in 4,800
Reduced risk

Donor 4210: 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.

genzyme

Chromosome Analysis

Patient Name: Donor, 4210
Referring Physician: [redacted]
Specimen # [redacted] **Client #:** [redacted]
Patient ID: [redacted]

Fairfax Cryobank / [redacted]

DOB: Not Given
SSN:

Date Collected: 02/14/2011
Date Received: 02/16/2011
Lab ID: #4210-110214
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	500
Metaphases Karyotyped:	2			**Dept. Section:**	B1

RESULTS: 46,XY
Male karyotype

INTERPRETATION:

This analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. The standard cytogenetic methodology utilized in this analysis does not routinely detect subtle rearrangements or low-level mosaicism and cannot detect microdeletions. Also, it cannot detect molecular cytogenetic abnormalities (such as microdeletions and microduplications) that may be detectable by array comparative genomic hybridization (aCGH).

Signed:

Frederick Elder

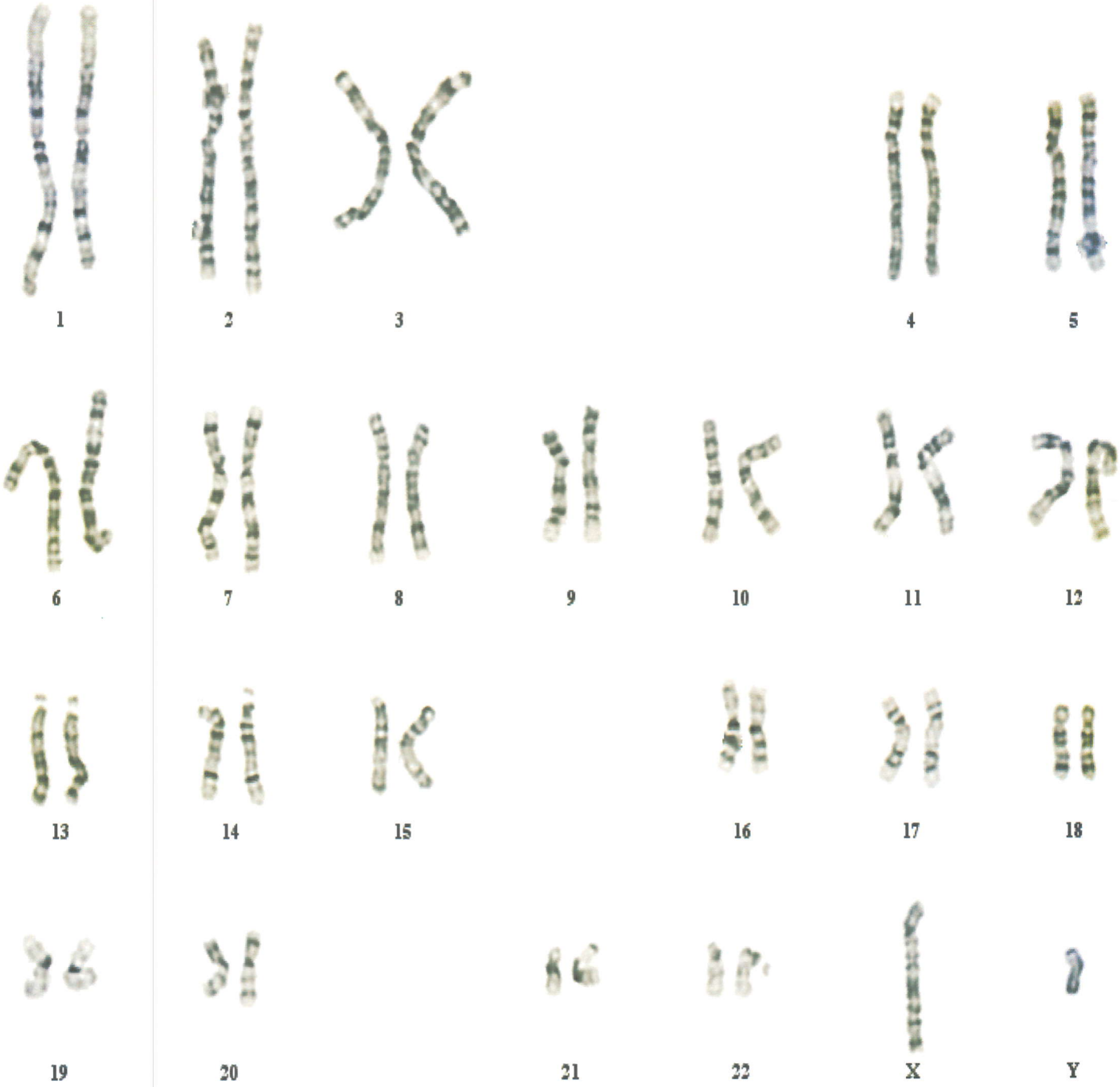
Frederick Elder, Ph.D. FFACMG

3/7/11

Date: 02/25/2011

Testing Performed At Genzyme Genetics 12906 Tampa Oaks Blvd Suite 300 Temple Terrace, FL 33637 1-800-966-4440

genzyme



Specimen #: **80331444 6**
Specimen Type: BLDPER
Patient Name: Donor, 4210
Image ID: CKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 02/16/2011
Date Reviewed: 02/25/2011
Reviewed By: FE

Quest Diagnostics

Report Status: Final

ID4210, DONOR

Patient Information	Specimen Information	Client Information
ID4210, DONOR **DOB:** [redacted] **AGE: 24** Gender: M Phone: NG Patient ID: 4210-110214	Specimen: IF479002G Requisition: 0000003 Collected: 02/14/2011 Received: 02/15/2011 / 07:58 CST Reported: 02/18/2011 / 02:31 CST	Client #: 41550 AUS0000 FAIRFAX CRYOBANK [redacted]

	Test Name	In Range	Out Of Range	Reference Range	Lab
	HEMOGLOBINOPATHY EVALUATION				
	RED BLOOD CELL COUNT	4.97		4.20-5.80 Million/uL	IG
	HEMOGLOBIN	15.4		13.2-17.1 g/dL	
	HEMATOCRIT	45.4		38.5-50.0 %	
	MCV	91.3		80.0-100.0 fL	
	MCH	31.0		27.0-33.0 pg	
	RDW	14.1		11.0-15.0 %	
*	HEMOGLOBIN A	97.9		>96.0 %	IG
*	HEMOGLOBIN F	<1.0		<2.0 %	
*	HEMOGLOBIN A2 (QUANT)	2.1		1.8-3.5 %	
*	INTERPRETATION				
	Normal phenotype.				
	CHOLESTEROL, TOTAL	153		125-200 mg/dL	IG
	AST	31		10-40 U/L	IG
	ALT	39		9-60 U/L	IG
	CBC (INCLUDES DIFF/PLT)				IG
	WHITE BLOOD CELL COUNT	9.9		3.8-10.8 Thousand/uL	
	RED BLOOD CELL COUNT	4.97		4.20-5.80 Million/uL	
	HEMOGLOBIN	15.4		13.2-17.1 g/dL	
	HEMATOCRIT	45.4		38.5-50.0 %	
	MCV	91.3		80.0-100.0 fL	
	MCH	31.0		27.0-33.0 pg	
	MCHC	34.0		32.0-36.0 g/dL	
	RDW	14.1		11.0-15.0 %	
	PLATELET COUNT	281		140-400 Thousand/uL	
	ABSOLUTE NEUTROPHILS	6960		1500-7800 cells/uL	
	ABSOLUTE LYMPHOCYTES	2356		850-3900 cells/uL	
	ABSOLUTE MONOCYTES	446		200-950 cells/uL	
	ABSOLUTE EOSINOPHILS	99		15-500 cells/uL	
	ABSOLUTE BASOPHILS	40		0-200 cells/uL	
	NEUTROPHILS	70.3		%	
	LYMPHOCYTES	23.8		%	
	MONOCYTES	4.5		%	
	EOSINOPHILS	1.0		%	
	BASOPHILS	0.4		%	
	ABO GROUP AND RH TYPE				IG
	ABO GROUP	A			
	RH TYPE	RH (D) NEGATIVE			

PERFORMING SITE:

IG QUEST DIAGNOSTICS-IRVING, 4770 REGENT BLVD., IRVING, TX 75063 Laboratory Director: SUZANNE H. KREISBERG, MD, CLIA: 45D0697943

ENTERED 3/7/11

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
[redacted]
Report Generated: 2016-05-26

Donor # 4210

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 53615
Kit Barcode: [redacted]
Specimen: Sperm, #56415
Specimen Collection: 2011-09-02
Specimen Received: 2016-05-16
Specimen Analyzed: 2016-05-26

TEST INFORMATION

Test: CarrierMap^{GEN} (Genotyping)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Mutations Tested: 27

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor # 4210 was not identified to carry any of the mutation(s) tested.

No pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.

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

Assay performed by Reprogenetics
CLIA ID: 31D1054821
3 Regent Street, Livingston, NJ 07039
Lab Technician: Bo Chu

Recombine CLIA # 31D2100763
Reviewed by Pere Colls, PhD, HCLD, Lab Director

Methods and Limitations

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

This test was developed and its performance determined by Recombine, Inc., and it has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

Diseases & Mutations Assayed

Oculocutaneous Albinism: Type 1 : Mutations (27): ♂ Genotyping | c.272G>A (p.C91Y), c.242C>T (p.P81L), c.265T>C (p.C89R), c.1A>G (p.M1V), c.140G>A (p.G47D), c.325G>A (p.G109R), c.568delG (p.G191Dfs), c.707G>A (p.W236X), c.832C>T (p.R278X), c.1118C>A (p.T373K), c.229C>T (p.R77W), c.823G>T (p.V275F), c.32G>A (p.W11X), c.149C>T (p.S50L), c.1467_1468insT (p.A490Cfs), c.820-2A>G, c.892C>T (p.R298W), c.1064C>T (p.A355V), c.1090A>C (p.N364H), c.1150C>G (p.P384A), c.1184+1G>A, c.1309G>A (p.D437N), c.1469C>A (p.A490D), c.133_134insC (p.P45fs), c.710delA (p.D237fs), c.978delA (p.Q326fs), c.1138_1158delTCTGCCAACGATCCTATCTTC (p.S380_F386del)

Residual Risk Information

Detection rates are calculated from the primary literature and may not be available for all ethnic populations. The values listed below are for genotyping. Sequencing provides higher detection rates and lower residual risks for each disease. More precise values for sequencing may become available in the future.

Disease	Carrier Rate	Detection Rate	Residual Risk
Oculocutaneous Albinism: Type 1	♂ European: 1/101	26.32%	1/137
	♂ Hutterite: 1/7	>99%	<1/700
	♂ Moroccan Jewish: 1/30	71.88%	1/107
	♂ Puerto Rican: Unknown	91.67%	Unknown