Donor 6565-PRS

Genetic Testing Summary

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

Last Updated: 08/27/18

Donor Reported Ancestry: Austrian, Peruvian, Spanish

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 97 mutations- in the CFTR gene	1/343
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Fragile X, PCR DNA Analysis	Normal Male	

*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.

genzyme

Cystic Fibrosis Mutation Analysis

Patient Name: Donor 6565, .
Referring Physician: Madelyn Kahn, MD
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

DOB: [redacted]
Sex: M
SSN:

Date Collected: 05/28/2010
Date Received: 06/01/2010
Lab ID:
Hospital ID:
Specimen Type: **BLDPER**

Pacific Reproductive Services
444 De Haro Street
Suite 222
San Francisco CA 94107

Ethnicity: Caucasian

Indication: Carrier test / Gamete donor

RESULTS: Negative for the 97 mutations analyzed 6/8/10

INTERPRETATION

This individual's risk to be a carrier is reduced from 1/25 (4%) to 1/343 (0.3%), based on these results and a negative family history.

COMMENTS:

Mutation Detection Rates among Ethnic Groups Detection rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild presentation (e.g. congenital absence of the vas deferens, pancreatitis) detection rates may vary from those provided here.

Ethnicity	Carrier risk reduction when no family history	Detection rate	References
African American	1/65 to 1/338	81%	Genet in Med 3:168, 2001
Ashkenazi Jewish	1/26 to 1/634	97%	Am J Hum Genet 51:951, 1994
Asian		Not Provided	Insufficient data
Caucasian	1/25 to 1/343	93%	Genet in Med 3:168, 2001; Genet in Med 4:90, 2002
Hispanic	1/46 to 1/205	78%	Genet in Med 3:168, 2001;www.dhs.ca.gov/pcfh/gdb/html/PDE/CFStudy.htm
Jewish, non-Ashkenazi		Varies by country of origin	Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997
Other or Mixed Ethnicity		Not Provided	Detection rate not determined and varies with ethnicity

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.

METHOD / LIMITATIONS:

DNA is isolated from the sample and tested for the 97 CF mutations listed. Regions of the *CFTR* gene are amplified enzymatically and subjected to a solution-phase multiplex allele-specific primer extension with subsequent hybridization to a bead array and fluorescence detection. Some mutations are then specifically identified by bi-directional dideoxysequencing. The assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V. 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.

Under the direction of: Ruth A Heim, PhD, FACMG Date: 06/08/2010

Ruth A. Heim, Ph.D., FACMG

Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357

genzyme
GENETICS

MUTATIONS ANALYZED				
ΔF311	3120+1G>A	712-1G>T	Q359K/T360K	S549N
ΔF508	3120G>A	935delA	Q493X	S549R T>G
ΔI507	3171delC	936delTA	Q552X	T338I
1078delT	3199del6	A455E	Q890X	V520F
1288insTA	3659delC	A559T	R1066C	W1089X
1677delTA	3667del4	C524X	R1158X	W1204X
1717-1G>A	3791delC	CFTRdele2,3	R1162X	W1282X
1812-1G>A	3849+10kbC>T	D1152H	R117C	Y1092X C>A
1898+1G>A	3876delA	E60X	R117H	Y1092X C>G
1898+5G>T	3905insT	E92X	R334W	Y122X
1949del84	394delTT	G178R	R347H	
2043delG	4016insT	G330X	R347P	
2055del9>A	405+1G>A	G480C	R352Q	
2105del13ins5	405+3A>C	G542X	R553X	
2108delA	406-1G>A	G551D	R560T	
2143delT	444delA	G85E	R709X	
2183delAA>G	457TAT>G	K710X	R75X	
2184delA	574delA	L206W	R764X	
2184insA	621+1G>T	M1101K	S1196X	
2307insA	663delT	N1303K	S1251N	
2789+5G>A	711+1G>T	P574H	S1255X	
2869insG	711+5G>A	Q1238X	S364P	

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.

LabCorp
Laboratory Corporation of America

June 8, 2010

Pacific Reproductive Svcs SF
444 De Haro St Ste 222
San Francisco, CA 94107

Test Results of: DONOR 6565, X
DOB: [redacted] Age: [redacted] Sex: M
Collected on: 05/28/2010
Received on: 05/29/2010
Reported on: 06/08/2010

Branch Number: [redacted]
Account Number: [redacted]
Specimen Number: [redacted]
Specimen Type: Blood

Patient ID#:

Physician:

Test: Fragile X, PCR DNA Analysis

Result:

NORMAL, Male **31 CGG repeats identified** ✓ CP 6/25/10

Interpretation:
DNA studies by PCR analysis identified one allele. These results do not provide evidence of the common CGG repeat expansion observed in patients with fragile X syndrome. Routine chromosome analysis is recommended in the diagnostic work-up for other causes of mental retardation. Due to the nature of the assay, small variations in reported repeat number may exist within and between laboratories.

Fragile X syndrome is one of the most common causes of inherited mental retardation. Some individuals with fragile X have characteristic physical features and behaviors. There can be wide variability in phenotypic expression. Fragile X is most often caused by an expansion in the number of the CGG repeats in the fragile X gene *(FMR1)*. People with fewer than 45 CGG repeats have alleles within the normal range. People with 45-54 repeats are considered normal but have alleles in the grey zone. Some increases and decreases in repeat number can occur in offspring of individuals with grey zone alleles, but the chance is small that grey zone alleles would expand to a full mutation in the next generation. Those with 55-200 repeats have alleles in the premutation range. These individuals are not expected to have fragile X, but are at increased risk to have children with fragile X syndrome. Individuals with more than 200 repeats have full mutations and are expected to be clinically affected. Exceptions can occur as there are rare forms of fragile X mental retardation protein (FMRP) deficiency not caused by CGG expansion, which may not be detected by this analysis.

Methodology:
DNA analysis of the *FMR1* gene was performed by PCR amplification followed by agarose gel, as well as capillary electrophoresis. Southern blot analysis was not indicated due to the presence of one normal allele by PCR. The detection rate of this test is >99% for the common fragile X expansion *(FRAXA)*. This test does not examine the *FRAXE* expansion. Molecular-based testing is highly accurate, but as in any laboratory test, rare diagnostic errors may occur. All test results must be combined with clinical information for the most accurate interpretation.

This test was developed and its performance characteristics determined by Laboratory Corporation of America Holdings (LabCorp). It has not been cleared or approved by the U.S. Food and Drug Administration.

The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or research.

References:

1. Park V, Howard-Peebles P, Sherman S, Taylor A, and Wulfsberg E. (1994). Am J Med Genet 53:380-381.
2. Maddalena A, et al. (2001). Genet Med 3:200-205.
3. Jacquemont S, Hagerman RJ, Lechey MA, Hall DA, Levine RA, Brunberg JA, Zhang L, Jardini T, Gane LW, Harris SW, Herman K, Grigsby J, Greco CM, Berry-Kravis E, Tassone F, and Hagerman PJ. (2004) J Amer Med Assoc 291:460-469.
4. Hagerman PJ and Hagerman RJ. (2004) Am J Hum Genet 74:805-816.

Results Released By: Kenneth J. Friedman, Ph.D., Director
Report Released By: Dagny M. Patton, MS, CGC, Genetic Counselor

Arundhati Chatterjee, M.D.
Medical Director

LabCorp
1912 Alexander Drive, RTP, NC, 27709 (800) 345-GENE

genzyme

SMN1 Copy Number Analysis

Patient Name: X Donor 6565
DOB [redacted] Age: [redacted]
SSN #: Gender: Male

Genzyme Specimen #: [redacted]
Case #: [redacted] Patient ID #: [redacted]
Date Collected: 05/28/2010 Date Received: 06/01/2010

163934 / 163935
Lab Corp
13112 Evening Creek Drive - South
San Diego, CA 92128

Referring Physician: Madelyn Kahn
Genetic Counselor:

Specimen Type: Peripheral Blood

Clinical Data: Carrier Test/Screening

Client Lab ID #:
Hospital ID #:
Specimen ID #:
Specimen(s) Received: 1 - Lavender 10 ml round bottom tube(s)
Ethnicity: Not Provided

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: Lynne S. Rosenblum, Ph.D., FACMG, on 06/04/2010
Reported by: /

Testing performed at Genzyme Genetics 3400 Computer Drive, Westborough, MA 01581 1-800-255-7357

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Pacific Reproductive Svcs SF

444 De Haro St Ste 222
San Francisco, CA 94107
Ph: (415)487-2288
Fax: (415) 863-4358 CAB-60

LCLS Specimen Number: [redacted]
Patient Name: **DONOR 6565, X**
Date of Birth: [redacted]
Gender: M
Patient ID:
Lab Number: [redacted]
Indications: DONOR

Account Number: [redacted]
Ordering Physician:
Specimen Type: **BLOOD**
Date Collected: 05/28/2010
Date Received: 05/29/2010
CoPath Number:
Client Reference:

Test: **Chromosome, Blood**

Date Reported: **06/15/2010**

Cells Counted: 15
Cells Analyzed: 5

Cells Karyotyped: 2
Band Resolution: 650

CYTOGENETIC RESULT: 46,XY

INTERPRETATION: NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Molecular results of fragile X testing will be reported separately.

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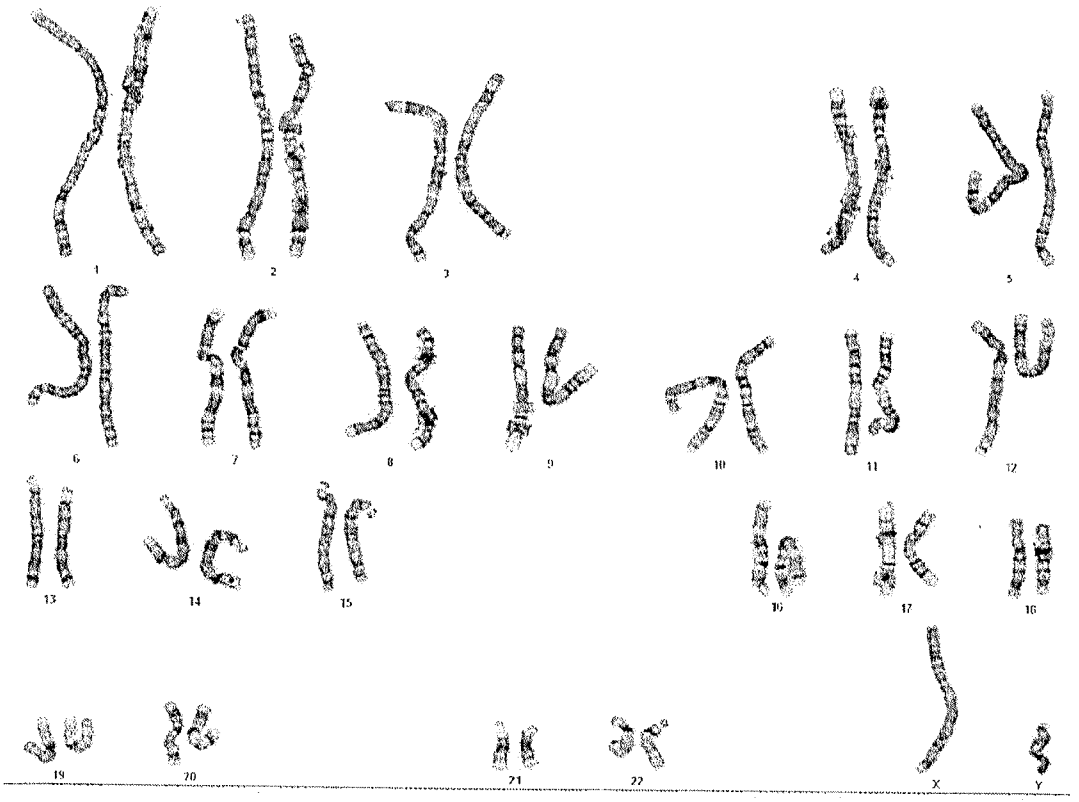
Pacific Reproductive Svcs SF

444 De Haro St Ste 222
San Francisco, CA 94107
Ph: (415)487-2288
Fax: (415) 863-4358 CAB-60

LCLS Specimen Number: [redacted]
Patient Name: **DONOR 6565, X**
Date of Birth: [redacted]
Gender: M
Patient ID:
Lab Number: [redacted]
Indications: DONOR

Account Number: [redacted]
Ordering Physician:
Specimen Type: **BLOOD**
Date Collected: 05/28/2010
Date Received: 05/29/2010
CoPath Number:
Client Reference:



Frederick W. Luthardt PhD, FACMG
Board Certified Cytogeneticist

David Corwin, M.D.
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Test Site: Dynacare Laboratories
550 17th Ave. Suite 200 , SEATTLE , WA , 98122-5789 (800) 676-8033

LabCorp
Laboratory Corporation of America

LabCorp Kansas City
1706 N Corrington Avenue
Kansas City, MO 64120
Phone: 800-457-1177

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
[redacted]		[redacted]	[redacted]	[redacted]	00

Patient Last Name: DONOR 6565
Patient First Name: DONOR 6565
Patient Middle Name:
Patient SS#:
Patient Phone:
Total Volume:
Age (Y/M/D):
Date of Birth:
Sex: M
Fasting:
Patient Address:

Account Address:
Pacific Reproductive Svcs SF
444 De Haro St Ste 222
San Francisco CA 94107

Additional Information:

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
04/22/10 15:30	04/23/10	04/26/10 15:14ET			

Tests Ordered: CBC With Differential/Platelet; Hgb Frac. w/o Solubility

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
CBC With Differential/Platelet					
WBC	5.7		x10E3/uL	4.0 - 10.5	01
RBC	4.69		x10E6/uL	4.10 - 5.60	01
Hemoglobin	15.0		g/dL	12.5 - 17.0	01
Hematocrit	43.0		%	36.0 - 50.0	01
MCV	92		fL	80 - 98	01
MCH	32.0		pg	27.0 - 34.0	01
MCHC	34.9		g/dL	32.0 - 36.0	01
RDW	13.1		%	11.7 - 15.0	01
Platelets	197		x10E3/uL	140 - 415	01
Neutrophils	66		%	40 - 74	01
Lymphs	29		%	14 - 46	01
Monocytes	**3**	**Low**	%	4 - 13	01
Eos	2		%	0 - 7	01
Basos	0		%	0 - 3	01
Neutrophils (Absolute)	3.8		x10E3/uL	1.8 - 7.8	01
Lymphs (Absolute)	1.7		x10E3/uL	0.7 - 4.5	01
Monocytes(Absolute)	0.2		x10E3/uL	0.1 - 1.0	01
Eos (Absolute)	0.1		x10E3/uL	0.0 - 0.4	01
Baso (Absolute)	0.0		x10E3/uL	0.0 - 0.2	01
Hgb Frac. w/o Solubility					
Hgb A	97.7		%	94.0 - 98.0	02
Hgb S	0.0		%	0.0	02
Hgb C	0.0		%	0.0	02
Hgb A2	2.3		%	0.7 - 3.1	02
Hgb F	0.0		%		02
			1 day	69.7 - 84.3	
			5 days	71.0 - 82.6	
			3 weeks	62.7 - 77.3	
			6- 9 weeks	41.9 - 63.9	
			3- 4 months	7.2 - 39.2	
			6 months	2.5 - 6.9	
			8-12 months	0.6 - 2.6	
			> 1 year	0.0 - 2.0	

DONOR 6565, DONOR 6565		[redacted]	Seq # 2011

FINAL REPORT

LabCorp
Laboratory Corporation of America

LabCorp Kansas City
1706 N Corrington Avenue
Kansas City, MO 64120
Phone: 800-457-1177

Patient Name							
DONOR 6565, DONOR 6565						Specimen Number: [redacted]	
Account Number	Patient ID	Control Number	Date and Time Collected	Date Reported	Sex	Age(Y/M/D)	Date of Birth
[redacted]		60111024737	04/22/10 15:30	04/26/10	M		

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Interpretation					
Normal adult hemoglobin present. CP 4/29/10					02

01	KC	LabCorp Kansas City 1706 N Corrington Avenue, Kansas City, MO 64120	Dir: Dugald Taylor, MD
02	BN	LabCorp Burlington 1447 York Court, Burlington, NC 27215-3361	Dir: William F Hancock, MD

For inquiries, the physician may contact **Branch: 800-888-1113 Lab: 800-457-1177**