Pacific Reproductive Services

Donor 9389-PRS

Genetic Testing Summary

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

Last Updated: 02/13/24

Donor Reported Ancestry: Chinese 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	Not provided
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/806
Fragile X, PCR DNA Analysis	Normal Male	
Special Testing		
Genes: HBA1/HBA2, GJB2	Negative by gene sequencing	

*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.

Integrated GENETICS
LabCorp Specialty Testing Group

Client/Sending Facility:
Pacific Reproductive Svcs [redacted]

[redacted]

LCLS Specimen Number:	[redacted]	Account Number:	[redacted]
Patient Name:	DONOR 9389, X	Ordering Physician:	
Date of Birth:	[redacted]	Specimen Type:	BLOOD
Gender:	M	Date Collected:	08/19/2013
Patient ID:	PC9389	Date Received:	08/22/2013
Lab Number:	[redacted]	CoPath Number:	
Indications:	DONOR SCREENING	Client Reference:	
Test:	**Chromosome, Blood, Routine**	Date Reported:	08/29/2013
Cells Counted:	20	Cells Karyotyped:	2
Cells Analyzed:	20	Band Resolution:	500

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.

9/3/13

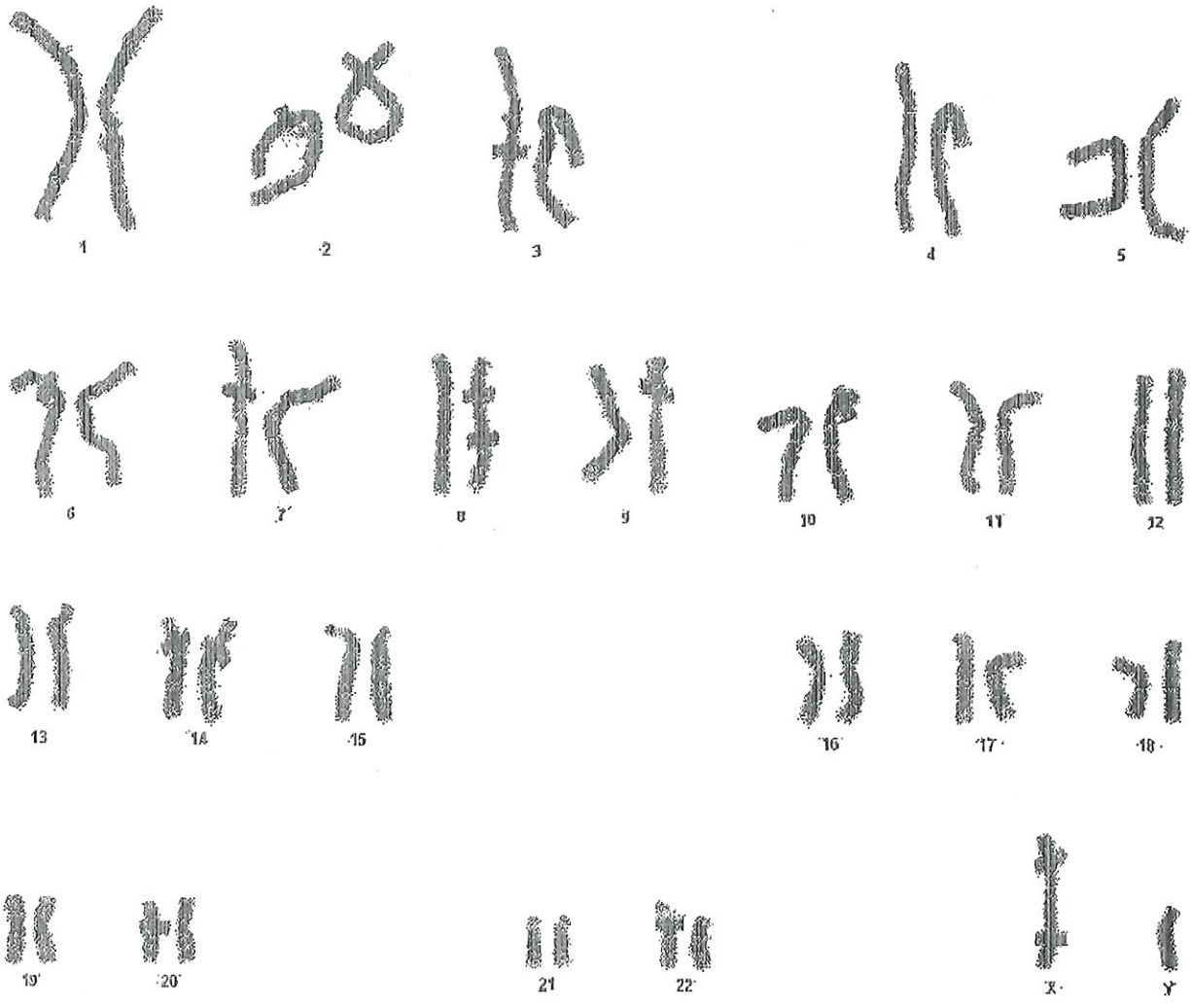
RESULTS REVIEW BY:
DISCUSSED WITH:
Recipient (Donor) N/A
OK TO FILE: (Y) N
DATE: 9/3/13

Integrated GENETICS
LabCorp Specialty Testing Group

Client/Sending Facility:
Pacific Reproductive Svcs [redacted]

[redacted]

LCLS Specimen Number:	[redacted]	Account Number:	[redacted]
Patient Name:	**DONOR 9389, X**	Ordering Physician:	
Date of Birth:	[redacted]	Specimen Type:	**BLOOD**
Gender:	M	Date Collected:	08/19/2013
Patient ID:	PC9389	Date Received:	08/22/2013
Lab Number:	[redacted]	CoPath Number:	
Indications:	DONOR SCREENING	Client Reference:	



Integrated GENETICS
LabCorp Specialty Testing Group

Client/Sending Facility:
Pacific Reproductive Svcs [redacted]

[redacted]

LCLS Specimen Number:	[redacted]	Account Number:	[redacted]
Patient Name:	DONOR 9389, X	Ordering Physician:	
Date of Birth:	[redacted]	Specimen Type:	BLOOD
Gender:	M	Date Collected:	08/19/2013
Patient ID:	PC9389	Date Received:	08/22/2013
Lab Number:	[redacted]	CoPath Number:	
Indications:	DONOR SCREENING	Client Reference:	

Karen K. Phillips

Karen Phillips, PhD, FACMG
Board Certified Cytogeneticist

Arundhati Chatterjee, MD
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
1904 Alexander Drive , RTP , NC , 27709-0153 (800) 345-4363
Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr. Research Triangle Park, NC 27709. Medical Director, Arundhati Chatterjee, MD.
Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly owned subsidiary of Laboratory Corporation of America Holdings.

LabCorp
Laboratory Corporation of America

LabCorp San Diego
13112 Evening Creek Dr So Ste 200
San Diego, CA 92128-4108
Phone: **858-668-3700**

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
[redacted]	PD9389	CYF04262552	04262552	626-432-1681	00

Patient Last Name	Account Address
DONOR 9389	Pacific Reproductive Svcs [redacted]
Patient First Name: **X** / Patient Middle Name	[redacted]
Patient SS# / Patient Phone / Total Volume	
[redacted] / Date of Birth [redacted] / Sex: M / Fasting: No	
Patient Address	Additional Information: 1770985749

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
06/24/13 12:00	06/25/13	07/03/13 04:07ET			

Tests Ordered
CBC With Differential/Platelet; Hgb Frac. w/o Solubility; Cytomegalovirus (CMV) Culture

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
CBC With Differential/Platelet					
WBC	7.0		x10E3/uL	4.0 - 10.5	01
RBC	4.65		x10E6/uL	4.14 - 5.80	01
Hemoglobin	14.7		g/dL	12.6 - 17.7	01
Hematocrit	45.9		%	37.5 - 51.0	01
MCV	**99**	**High**	fL	79 - 97	01
MCH	31.6		pg	26.6 - 33.0	01
MCHC	32.0		g/dL	31.5 - 35.7	01
RDW	12.9		%	12.3 - 15.4	01
Platelets	213		x10E3/uL	140 - 415	01
Neutrophils	69		%	40 - 74	01
Lymphs	23		%	14 - 46	01
Monocytes	7		%	4 - 13	01
Eos	0		%	0 - 7	01
Basos	1		%	0 - 3	01
Neutrophils (Absolute)	4.9		x10E3/uL	1.8 - 7.8	01
Lymphs (Absolute)	1.6		x10E3/uL	0.7 - 4.5	01
Monocytes(Absolute)	0.5		x10E3/uL	0.1 - 1.0	01
Eos (Absolute)	0.0		x10E3/uL	0.0 - 0.4	01
Baso (Absolute)	0.0		x10E3/uL	0.0 - 0.2	01
Immature Granulocytes	0		%	0 - 2	01
Immature Grans (Abs)	0.0		x10E3/uL	0.0 - 0.1	01
Hgb Frac. w/o Solubility					
Hgb F	0.0		%	0.0 - 2.0	02
Hgb A	97.4		%	94.0 - 98.0	02
Hgb S	0.0		%	0.0	02
Hgb C	0.0		%	0.0	02
Hgb A2	2.6		%	0.7 - 3.1	02
Interpretation					
Normal adult hemoglobin present.					02

7/8/13 * will Repeat after 3 mths. for further assessment. 3 m return to normal parameters. dh

DONOR 9389, X	PD9389	[redacted]	Seq # 2184

07/03/13 04:07 ET **FINAL REPORT** Page 1 of 2

LabCorp
Laboratory Corporation of America

LabCorp San Diego
13112 Evening Creek Dr So Ste 200
San Diego, CA 92128-4108
Phone: 858-668-3700

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
[redacted]	PC9389	[redacted]	[redacted]	[redacted]	00

Patient Last Name	Patient First Name	Patient Middle Name	Patient SS#	Patient Phone	Total Volume	Age (Y/M/D)	Date of Birth	Sex	Fasting
DONOR 9389	X					[redacted]	[redacted]	M	No

Account Address: Pacific Reproductive Svcs [redacted]

Patient Address:

Additional Information:

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
07/25/13 10:00	07/26/13	07/26/13 09:31ET			

Tests Ordered: CBC With Differential/Platelet

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
CBC With Differential/Platelet					
WBC	5.2		x10E3/uL	4.0 - 10.5	01
RBC	4.70		x10E6/uL	4.14 - 5.80	01
Hemoglobin	14.6		g/dL	12.6 - 17.7	01
Hematocrit	45.2		%	37.5 - 51.0	01
MCV	96		fL	79 - 97	01
MCH	31.1		pg	26.6 - 33.0	01
MCHC	32.3		g/dL	31.5 - 35.7	01
RDW	12.9		%	12.3 - 15.4	01
Platelets	233		x10E3/uL	140 - 415	01
Neutrophils	53		%	40 - 74	01
Lymphs	37		%	14 - 46	01
Monocytes	8		%	4 - 13	01
Eos	1		%	0 - 7	01
Basos	1		%	0 - 3	01
Neutrophils (Absolute)	2.8		x10E3/uL	1.8 - 7.8	01
Lymphs (Absolute)	1.9		x10E3/uL	0.7 - 4.5	01
Monocytes(Absolute)	0.4		x10E3/uL	0.1 - 1.0	01
Eos (Absolute)	0.0		x10E3/uL	0.0 - 0.4	01
Baso (Absolute)	0.0		x10E3/uL	0.0 - 0.2	01
Immature Granulocytes	0		%	0 - 2	01
Immature Grans (Abs)	0.0		x10E3/uL	0.0 - 0.1	01

01 SO LabCorp San Diego Dir: Kelli Chase, MD
13112 Evening Creek Dr So Ste 200, San Diego, CA 92128-4108
For inquiries, the physician may contact **Branch: 800-859-6046 Lab: 858-668-3700**

RESULTS REVIEW BY:
DISCUSSED WITH:
Recipient Donor N/A
OK TO FILE: Y N
DATE: 8/1/13

DONOR 9389, X	PC9389	[redacted]	Seq # 2199

07/26/13 09:31 ET **FINAL REPORT** Page 1 of 1

LabCorp
Laboratory Corporation of America

LabCorp San Diego
13112 Evening Creek Dr So Ste 200
San Diego, CA 92128-4108
Phone: 858-668-3700

Patient Name					Specimen Number		
DONOR 9389, X					175-229-7216-0		
Account Number	Patient ID	Control Number	Date and Time Collected	Date Reported	Sex	Age(Y/M/D)	Date of Birth
04262552	PD9389	CYF04262552	06/24/13 12:00	07/03/13	M	[redacted]	[redacted]

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Cytomegalovirus (CMV) Culture					
	No Cytomegalovirus isolated. ✓				02

01	SO	LabCorp San Diego Dir: Kelli Chase, MD 13112 Evening Creek Dr So Ste 200, San Diego, CA 92128-4108
02	BN	LabCorp Burlington Dir: William F Hancock, MD 1447 York Court, Burlington, NC 27215-3361

For inquiries, the physician may contact **Branch: 800-859-6046 Lab: 858-668-3700**

7/8/13

RESULTS REVIEW BY:
DISCUSSED WITH:
Recipient Donor N/A
OK TO FILE: Y N
DATE: 7/8/13

Integrated GENETICS

Cystic Fibrosis Mutation Analysis

Patient Name: Donor 9389, .
Referring Physician: Madelyn Kahn, MD
Specimen #: [redacted]
Patient ID: [redacted]
Client #: [redacted]
Case #: [redacted]

880107 / 155522
Pacific Reproductive Services
[redacted]

DOB: [redacted]
Sex: M
SSN: ***-**-****

Date Collected: 08/19/2013
Date Received: 08/20/2013
LAB ID:
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Asian

Indication: Carrier Test / Gamete donor

RESULTS: Negative for the 97 mutations analyzed ✓ sh 8/26/13

INTERPRETATION:

This individual is negative for the mutations analyzed. This result reduces but does not eliminate the risk to be a CF carrier.

COMMENTS:

Mutations 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**
Caucasian	1/25 to 1/343	93%	Genet in Med 3:168, 2001; Genet in Med 4:90, 2002
African American	1/65 to 1/338	81%	Genet in Med 3:168, 2001
Hispanic	1/46 to 1/205	76%	Genet in Med 3:168, 2001;www.dhs.ca.gov/pcfh/gdb/html/PDE/CFStudy.htm
Ashkenazi Jewish	1/26 to 1/834	97%	Am J Hum Genet 51:951, 1994
Jewish, non-Ashkenazi		Varies by country of origin	Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997
Asian		Not Provided	Insufficient data
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.

RESULTS REVIEW BY: sh
DISCUSSED WITH:
Recipient Donor N/A
OK TO FILE:
DATE: 8/26/13

Electronically Signed By: Ruth A. Heim, Ph.D., FACMG, on 08/23/2013

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 Esoterix Genetic Laboratories, LLC. 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.

Integrated GENETICS
LabCorp Specialty Testing Group

SMN1 Copy Number Analysis

Patient Name: X Donor 9389
DOB: [redacted] Age [redacted]
SSN #: Gender: Male

Specimen #: [redacted]
Case #: [redacted] Patient ID #: [redacted]
Date Collected: 08/19/2013 Date Received: 08/21/2013

163934 / 163935
LabCorp
13112 Evening Creek Drive - South
San Diego, CA 92128
USA

Referring Physician: . 1 N/A
Genetic Counselor:

Specimen Type: Peripheral blood

Clinical Data: Not Provided

Client Lab ID #: [redacted]
Hospital ID #: SO^EGLMA
Specimen ID #:
Specimen(s) Received: 1 - Lavender 10 ml round bottom tube(s)

Ethnicity: Not Provided

RESULTS: SMN1 copy number: 2 (Reduced Carrier Risk) ✓ 9/3/13

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. Information regarding clinical indication may provide a more detailed interpretation.

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]**	**Prior Carrier Risk[1]**	**Reduced Carrier Risk for 2 copy result**	**Reduced Carrier Risk for 3 copy result**
Caucasian	94.8%	1:47	1:834	1:5,600
Ashkenazi Jewish	90.5%	1:67	1:611	1:5,400
Asian	93.3%	1:59	1:806	1:5,600
Hispanic	90.0%	1:68	1:579	1:5,400
African American	70.5%	1:72	1:130	1:4,200
Asian Indian	90.2%	1:52	1:443	1:5,400
Mixed or Other Ethnic Background	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 the internal standard reference genes. A mathematical algorithm is used to calculate and report SMN1 copy numbers of 0, 1, 2 and 3. Based upon this analysis, an upper limit of 3 represents the highest degree of accuracy in reporting SMN1 copy number with statistical confidence. Sequencing of the primer and probe binding sites is performed on all fetal samples and samples with one copy of SMN1 by real-time PCR 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. Sugarman EA, Nagan N, Zhu H, et al. Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur J Hum Genet 2012; 20:27-32. 2. Prior TW, et al. Technical standards and guidelines for spinal muscular atrophy testing. Genet Med 2011; 13(7): 686-694.

RESULTS REVIEW BY:
DISCUSSED WITH:
Recipient Donor N/A
OK TO FILE: Y N
DATE: 9/3/13

Electronically Signed by: Zhaoqing Zhou, Ph.D., FACMG, on 08/26/2013
Reported by: /

LabCorp
Laboratory Corporation of America

LabCorp San Diego
13112 Evening Creek Dr So Ste 200
San Diego, CA 92128-4108
Phone: [redacted]

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
[redacted]	PC9389	[redacted]	[redacted]	[redacted]	00

Patient Last Name	Account Address
DONOR 9389	Pacific Reproductive Svcs [redacted]
Patient First Name: X / Patient Middle Name:	[redacted]
Patient SS# / Patient Phone / Total Volume:	
Age (Y/M/D): [redacted] / Date of Birth: [redacted] / Sex: M / Fasting: No	
Patient Address	Additional Information

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
08/19/13 13:00	08/20/13	08/29/13 20:14ET			

Tests Ordered
SMN1 Copy Number Analysis; Chromosome, Blood, Routine; Fragile X, PCR Reflex Southern

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB

SMN1 Copy Number Analysis

Genetic Counselor:	Not applicable				01
Client Specimen ID:	Not applicable				01
Specimen Type:					01
Peripheral blood					
Specimen(s) Received:					01
1 - Lavender 10 ml round bottom tube(s)					
Clinical Data:					01
Not Provided					
Ethnicity:					01
Not Provided					
Results:					01
SMN1 copy number:	2 (Reduced Carrier Risk)				
Interpretation:	Note				01

This individual has an SMN1 copy number of two. This result reduces but does not eliminate the risk to be a carrier of SMA. Information regarding clinical indication may provide a more detailed interpretation.

Comments: Note 01

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

RESULTS REVIEW BY: 9/3/13
DISCUSSED WITH:
Recipient Donor N/A
OK TO FILE: Y N
DATE: 9/3/13

patients.

Carrier Detection Rate: Note 01

Carrier Frequency and Risk Reductions for Individuals with No Family History of SMA

Ethnicity	Detection rate(1)	Prior Carrier Risk(1)	Reduced Carrier Risk for 2 copy result	Reduced Carrier Risk for 3 copy result
Caucasian	94.8%	1:47	1:834	1:5,600
Ashkenazi Jewish	90.5%	1:67	1:611	1:5,400
Asian	93.3%	1:59	1:806	1:5,600
Hispanic	90.0%	1:68	1:579	1:5,400
African American	70.5%	1:72	1:130	1:4,200
Asian Indian	90.2%	1:52	1:443	1:5,400

Method/Limitations: Note 01

METHOD/LIMITATIONS: Specimen DNA is isolated and amplified by real-time polymerase chain reaction (PCR) for exon 7 of the SMN1 gene and the internal standard reference genes. A mathematical algorithm is used to calculate and report SMN1 copy numbers of 0, 1, 2 and 3. Based upon this analysis, an upper limit of 3 represents the highest degree of accuracy in reporting SMN1 copy number with statistical confidence. Sequencing of the primer and probe binding sites is performed on all fetal samples and samples with one copy of SMN1 by real-time PCR 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: Note 01

REFERENCES: 1. Sugarman EA, Nagan N, Zhu H, et al. Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur J Hum Genet 2012; 20:27-32. 2. Prior TW, et al. Technical standards and guidelines for spinal muscular atrophy testing. Genet Med 2011; 13(7): 686-694.

Disclaimer: Note 01

The test was developed and its performance characteristics have been determined by Esoterix Genetic Laboratories, LLC. 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. Integrated Genetics is a business unit of

LabCorp
Laboratory Corporation of America

LabCorp San Diego
13112 Evening Creek Dr So Ste 200
San Diego, CA 92128-4108
Phone: [redacted]

Patient Name					Specimen Number		
DONOR 9389, X					[redacted]		
Account Number	Patient ID	Control Number	Date and Time Collected	Date Reported	Sex	Age(Y/M/D)	Date of Birth
04262552	PC9389	[redacted]	08/19/13 13:00	08/29/13	M	[redacted]	[redacted]

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB

Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

Electronically Signed by: 01
Zhaoqing Zhou, Ph.D., FACMG,
Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LL
a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

SMN1 Copy Number Analysis PDF
. 01

Chromosome, Blood, Routine

Chromosome-Routine 02
The test has been resulted. The testing report will be sent by fax, mail and/or electronic interface reporting, based on client result delivery set up.

Director Review: Comment: 02
Karen Phillips, PhD, FACMG

Fragile X, PCR Reflex Southern

Fragile X DNA 03
Molecular analysis report has been mailed.
This test was developed and its performance characteristics determined by 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 for research.

01	EGLMA	Esoterix Genetic Laboratories 3400 Computer Drive, Westborough, MA 01581-1771	Dir: Bernice Allitto, PhD
02	YU	LabCorp RTP 1904 Alexander Drive Ste C, RTP, NC 27709-0153	Dir: Arundhati Chatterjee, MD
03	TG	LabCorp RTP 1912 Alexander Drive, RTP, NC 27709-0150	Dir: Arundhati Chatterjee, MD

For inquiries, the physician may contact **Branch: 800-859-6046 Lab: 508-898-9001**

RESULTS REVIEW BY: [signature]
DISCUSSED WITH:
Recipient (Donor) N/A
OK TO FILE: (Y) N
DATE: 09/3/13

LabCorp
Laboratory Corporation of America

August 23, 2013

Pacific Reproductive Svcs
[redacted]

Test Results of: DONOR 9389, X
DOB: [redacted] Sex: M
Collected on: 08/19/2013
Received on: 08/20/2013
Reported on: 08/23/2013

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

Patient ID#: PC9389

Physician:

Test: Fragile X, PCR DNA Analysis

Result:

NORMAL, Male
30 CGG repeats identified ✓

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: Alecia S. Willis, Ph.D., Director
Report Released By: Emily Walsh, MS, CGC, Genetic Counselor

Arundhati Chatterjee, M.D.
Medical Director

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

9/3/13

RESULTS REVIEW BY:
DISCUSSED WITH:
Recipient (Donor) N/A
OK TO FILE: (Y) N
DATE: 9/3/13

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[redacted]

Patient Information:
9389 PRS, Donor
DOB: [redacted]
Sex: M
MR#: 9389 PRS
Patient#: [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: **Feb 07,2024**

Accession:
[redacted]
Test#: [redacted]
Specimen Type: DNA
Collected: Jan 23,2024

Accession:
N/A

FINAL RESULTS

No carrier mutations identified

TEST PERFORMED

Custom Beacon Carrier Screening Panel
(3 Gene Panel: *GJB2, HBA1, and HBA2*; 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.
- 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 - 3 Genes

This analysis was run using the Custom Beacon Carrier Screening Panel gene list. 3 genes were tested with 97.0% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

GJB2, HBA1, HBA2

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, 98.90% and 96.96% 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 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.

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Gene Specific Notes and Limitations

HBA1: The phase of heterozygous alterations in the *HBA1* gene cannot be determined, but can be confirmed through parental testing.
HBA2: The phase of heterozygous alterations in the *HBA2* gene cannot be determined, but can be confirmed through parental testing.

SIGNATURE:

Jianbo Song, Ph.D., ABMGG, CGMB, CCS, FACMG on **2/7/2024 09:08 PM PST**
Electronically signed

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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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
GJB2	Nonsyndromic hearing loss 1A	AR	General Population	1 in 42	99%	1 in 4,101	1 in 688,968
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 240,100
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 168,084
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Latino Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 8,702,992
HBA1	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
HBA2	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000

* For genes that have tested negative
† The carrier frequency for heterozygous alpha thalassemia carriers (αα/α-) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis (αα/- -) is 1 in 1000.
Abbreviations: AR, autosomal recessive; XL, X-linked