September 25, 2012

Seattle Sperm Bank
4915 25th Ave Ne Ste 204
SEATTLE, WA 98105

Test Results of: 9617, DONOR
[redacted] Age: [redacted] Sex: M
Collected on: 09/20/2012
Received on: 09/20/2012
Reported on: 09/25/2012

Branch Number: WAB55
Account Number: 46857540
Specimen Number: 264-129-0744-0
Specimen Type: Blood

Patient ID#: 9617

Physician: OLLIFFE,JE

Test: Cystic Fibrosis, DNA Analysis

Result:

Negative for 32 mutations

Interpretation:
This individual is negative for the 32 most common cystic fibrosis (CF) mutations. This includes the mutations recommended by ACOG/ACMG for routine carrier screening. The detection rate varies with ethnicity and is listed below. In the absence of a family history, the remaining risk that a person with a negative result could be a carrier is listed in the table. If there is a family history of CF, these risk figures do not apply. Please contact LabCorp- Esoterix at 1(888) 690-3935 for a revised report. Diagnosis of cystic fibrosis should not rely on DNA testing alone, but should take into consideration clinical symptoms and other test results, such as sweat chloride analysis. The presence of a rare mutation cannot be ruled out. The diagnostic criteria for cystic fibrosis are:

At least one characteristic clinical feature, *or*		Positive sweat chloride on 2 separate occasions, *or*
Family history of CF, *or*	**AND**	Presence of 2 *CFTR* mutations, *or*
Positive neonatal screening test		Positive nasal transmembrane potential

Cystic fibrosis is a common genetic disorder resulting in chronic pulmonary and gastrointestinal/pancreatic disease. There is wide variability in clinical symptoms. CF is inherited in a recessive manner, which means that both parents must be carriers to have an affected child. When both parents are carriers, there is a 25% chance with each pregnancy that the child will be affected. Genetic counseling and CF molecular testing are recommended for the reproductive partners and at-risk family members of CF carriers.

Ethnicity	Detection Rate	Carrier Risk	Remaining carrier risk given a negative result
Ashkenazi Jewish	97%	1/25	1/800
Caucasian (non-Hispanic)	90%	1/25	1/240
African-American	69%	1/65	1/207
Hispanic	73%	1/46	1/168
Asian	55%	1/90	1/198

Mutations:

G85E	A455E	S549N	R1162X	711+1 G→T	2184delA	3876delA
R117H	ΔI507	S549R	W1282X	1078delT	2789+5 G→A	3905insT
R334W	ΔF508	G551D	N1303K	1717-1 G→A	3120+1 G→A	
R347H	V520F	R553X	394delTT	1898+1 G→A	3659delC	
R347P	G542X	R560T	621+1 G→T	2183AA→G	3849+10kb C→T	

Methodology:
DNA analysis of the *CFTR* gene was performed by the oligonucleotide ligation assay. Molecular-based testing is highly accurate, but as in any laboratory test, rare diagnostic errors may occur. When R117H is positive, reflex testing for 5T is performed. Reflex testing for the F508C, I506V and I507V polymorphisms is performed to rule out false positive ΔF508 homozygotes, using Tm Biosciencе/Luminex primer extension chemistry. The assay provides information intended to be used for carrier screening in adults of reproductive age, as an aid in newborn screening, and as a confirmatory test for another medically established diagnosis in newborns and children. The test is not indicated for use in fetal diagnostic testing, pre-implantation screening, or for any stand-alone diagnostic purposes without confirmation by another medically established diagnostic product or procedure.

References:

1. Watson, et al. (2004) *Genet Med* 6:387-91
2. Richards, et al. (2002) *Genet Med* 4:379-391
3. Preconception and prenatal carrier screening for cystic fibrosis: (2001)ACOG.ACMG publication

Results Released By: Samuel H. Pepkowitz, M.D., Medical Director
Report Released By: Samuel H. Pepkowitz, M.D., Medical Director

Samuel H. Pepkowitz, MD
Medical Director, Esoterix

LabCorp - Esoterix
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Integrated GENETICS
Laboratory Specialty Testing Service

SMN1 Copy Number Analysis

Patient Name: Donor 9617 [redacted]

SSN #: [redacted] Gender: Male

803037 / 803038
Seattle Sperm Bank
4915 25th Avenue East
Suite 204W
Seattle, WA 98105
USA

Specimen #: 62215516-1
Case #: 62105414 Patient ID #: 61831109
Date Collected: 09/20/2012 Date Received: 09/21/2012

Referring Physician: Jeffrey Olliffe
Genetic Counselor:

Client Lab ID #:
Hospital ID #:
Specimen ID #:

Specimen Type: Peripheral blood

Specimen(s) Received: 1 - Yellow (ACD) 10 ml round bottom tube(s)

Clinical Data: Carrier Test/Gamete donor

Ethnicity: Caucasian

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]	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.

Electronically Signed by: Ruth Heim, Ph.D., FACMG, on 09/24/2012
Reported by: /

Integrated GENETICS
LabCorp Specialty Testing Group

Seattle Sperm Bank

4915 25th Ave Ne Ste 204
SEATTLE, WA 98105
Ph: (206)588-1484
Fax: (206) 588-1485 WAB-55

LCLS Specimen Number:	**264-129-0744-0**	Account Number:	46857540
Patient Name:	**9617, DONOR**	Ordering Physician:	**Dr. OLLIFFE**
[redacted]		Specimen Type:	**BLOOD**
Gender:	M	Date Collected:	09/20/2012
Patient ID:	9617	Date Received:	09/21/2012
Lab Number:	(J12-3138 L	CoPath Number:	
Indications:	DONOR	Client Reference:	
Test:	**Chromosome, Blood, Routine**	Date Reported:	**09/28/2012**
Cells Counted:	15	Cells Karyotyped:	2
Cells Analyzed:	5	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.

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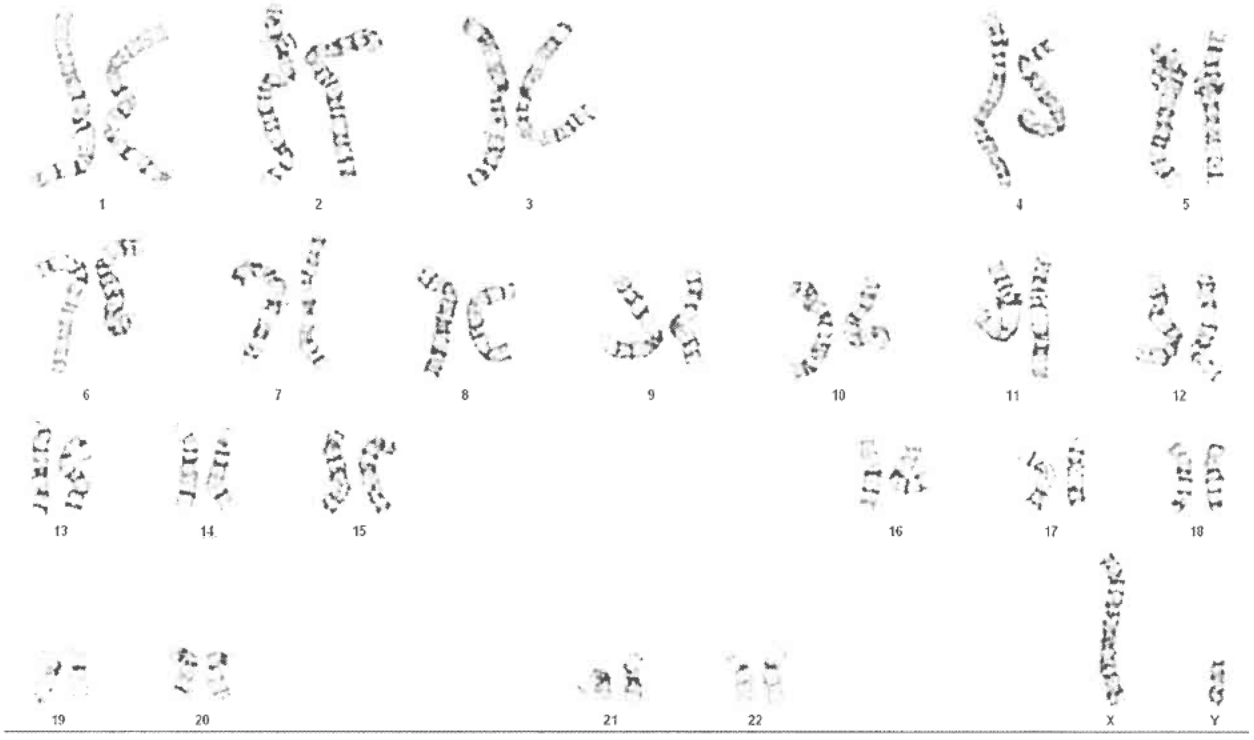

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Elisabeth Keitges PhD, FACMG
Board Certified Cytogeneticist

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 (206) 861-7050