Donor 5972-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: East Indian

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	No residual risk available for those of Asian ethnicity
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/628
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.

3015 Williams Drive #110, Fairfax, VA 22031 info@fairfaxcryobank.com 703-698-3976 800-338-8407

genzyme

Cystic Fibrosis Mutation Analysis

Patient Name: Donor, 5972
Referring Physician: Madelyn Kahn, MD
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted] **Case #:** [redacted]

DOB [redacted]
Sex: M
SSN:

Date Collected: 03/25/2009
Date Received: 03/26/2009
Lab ID:
Hospital ID:
Specimen Type: **BLDPER**

Pacific Reproductive Services
65 North Madison Avenue
Suite 610
Pasadena CA 91101

Ethnicity: East Indian

Indication: Carrier test / Gamete donor

RESULTS: Negative for the 97 mutations analyzed ✓

INTERPRETATION

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

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 1/834	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

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 fluorescent detection. The assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V. In some cases, specific allele identification requires enzymatic amplification followed by hybridization to oligonucleotide probes.

RESULTS REVIEWED BY [redacted]

DISCUSSED WITH:
RECIPIENT (DONOR) NA

OK TO FILE (Y) N

DATE: 4/22/09

Under the direction of: Zhuhui

Hui Zhu, PhD FACMG

Date: 04/03/2009

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

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	

False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation or somatic heterogeneity of the tissue sample. This test was developed and its performance characteristics determined by Genzyme. 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. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.

LabCorp
Laboratory Corporation of America

ViroMed
6101 Blue Circle Drive
Minnetonka, MN 55343-9018

Phone: **800-457-1177**

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
[redacted]	DONOR 5972	[redacted]	[redacted]	626-432-1681	00

Patient Last Name	Account Address
DONOR	Pacific Reproductive Svcs Pasa
Patient First Name: 5972 / Patient Middle Name:	65 N Madison Ave Ste 610
Patient SS# / Patient Phone / Total Volume:	Pasadena CA 91101
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
07/27/09 14:15	07/29/09	08/05/09 17:06ET			

Tests Ordered
S.Muscular Atrophy Carrier; Fragile X, PCR Reflex Southern

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB

S.Muscular Atrophy Carrier

Specimen Status 01

Reference lab report to follow via mail.

Specimen Type: Comment: 02

Peripheral Blood

Clinical Data: Comment: 02

Carrier Test / Screening

Results: Comment: 02

SMN1 copy number: 2 (Reduced Carrier Risk) ✓

8/12/09 on to file 8

Interpretation Comment: 02

Comment: Comment: 02

Spinal muscular atrophy (SMA) is an autosomal recessive disease of variable age of onset and severity caused by mutation (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 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 prior Carrier	Reduced Carrier	Reduced Carrier

Patient Name					Specimen Number		
DONOR, 5972					[redacted]		
Account Number	Patient ID	Control Number	Date and Time Collected	Date Reported	Sex	Age(Y/M/D)	Date of Birth
[redacted]	DONOR 5972	[redacted]	07/27/09 14:15	08/05/09	M	[redacted]	[redacted]

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
	Risk(1)	Risk for 2 copy result	Risk for 3 copy result		
Caucasian	94.9%	1:35	1:632	1:3,600	
Ashkenzai 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: Comment: 02

Specimen DNA is isolated and ampilified 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 interpretaion. 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: Comment: 02

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 Amendment of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjuction with clinical assessment, when available.

LabCorp
Laboratory Corporation of America

ViroMed
6101 Blue Circle Drive
Minnetonka, MN 55343–9018

Phone: **800–457–1177**

Patient Name					Specimen Number		
DONOR, 5972					[redacted]		
Account Number	Patient ID	Control Number	Date and Time Collected	Date Reported	Sex	Age(Y/M/D)	Date of Birth
[redacted]	DONOR 5972	[redacted]	07/27/09 14:15	08/05/09	M	[redacted]	[redacted]

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
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	KC	LabCorp Kansas City 1706 N Corrington Avenue, Kansas City, MO 64120	Dir: Nancy Litton, MD
02	G$	Genzyme Genetics 3400 Computer Drive, Westborough, MA 01581	Dir: Bernice Allitto, PhD
03	TG	LabCorp RTP 1912 Alexander Drive, RTP, NC 27709-9998	Dir: Arundhati Chatterjee, MD

For inquiries, the physician may contact **Branch: 800-859-6046 Lab: 800-457-1177**

LabCorp
Laboratory Corporation of America

August 5, 2009

Pacific Reproductive Svcs Pasa
65 N Madison Ave Ste 610
Pasadena, CA 91101

Test Results of: DONOR, 5972
DOB: [redacted] Age: [redacted] Sex: M
Collected on: 07/27/2009
Received on: 07/29/2009
Reported on: 08/05/2009

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

Patient ID#: DONOR 5972

Physician:

Test: Fragile X, PCR DNA Analysis

Result:

NORMAL, Male **28 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 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: Val V. Zvereff, M.D., Ph.D., Director
Report Released By: Lori A. Carpenter, MS, Genetic Counselor

Arundhati Chatterjee, M.D.
Medical Director

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

RESULTS REVIEWED BY [signature]

DISCUSSED WITH:
RECIPIENT DONOR (NA)

OK TO FILE (Y) N

DATE: 8/12/09

genzyme

Chromosome Analysis

Patient Name: Donor, 5972
Referring Physician: Madelyn Kahn, MD
Specimen #: [redacted]
Patient ID: [redacted]
Client #: 880107

DOB [redacted]
SSN:

Date Collected: 03/25/2009
Date Received: 03/26/2009
Lab ID:
Hospital ID:
Specimen Type: **Peripheral Blood**

Pacific Reproductive Services
65 North Madison Avenue
Suite 610
Pasadena CA 91101

Indication: Gamete donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	550
Metaphases Karyotyped:	3			**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 small rearrangements and low level mosaicism, and cannot detect microdeletions. ✓

RESULTS REVIEWED BY JM

DISCUSSED WITH:
RECIPIENT (DONOR) NA

OK TO FILE (Y) N

DATE: 4/22/09

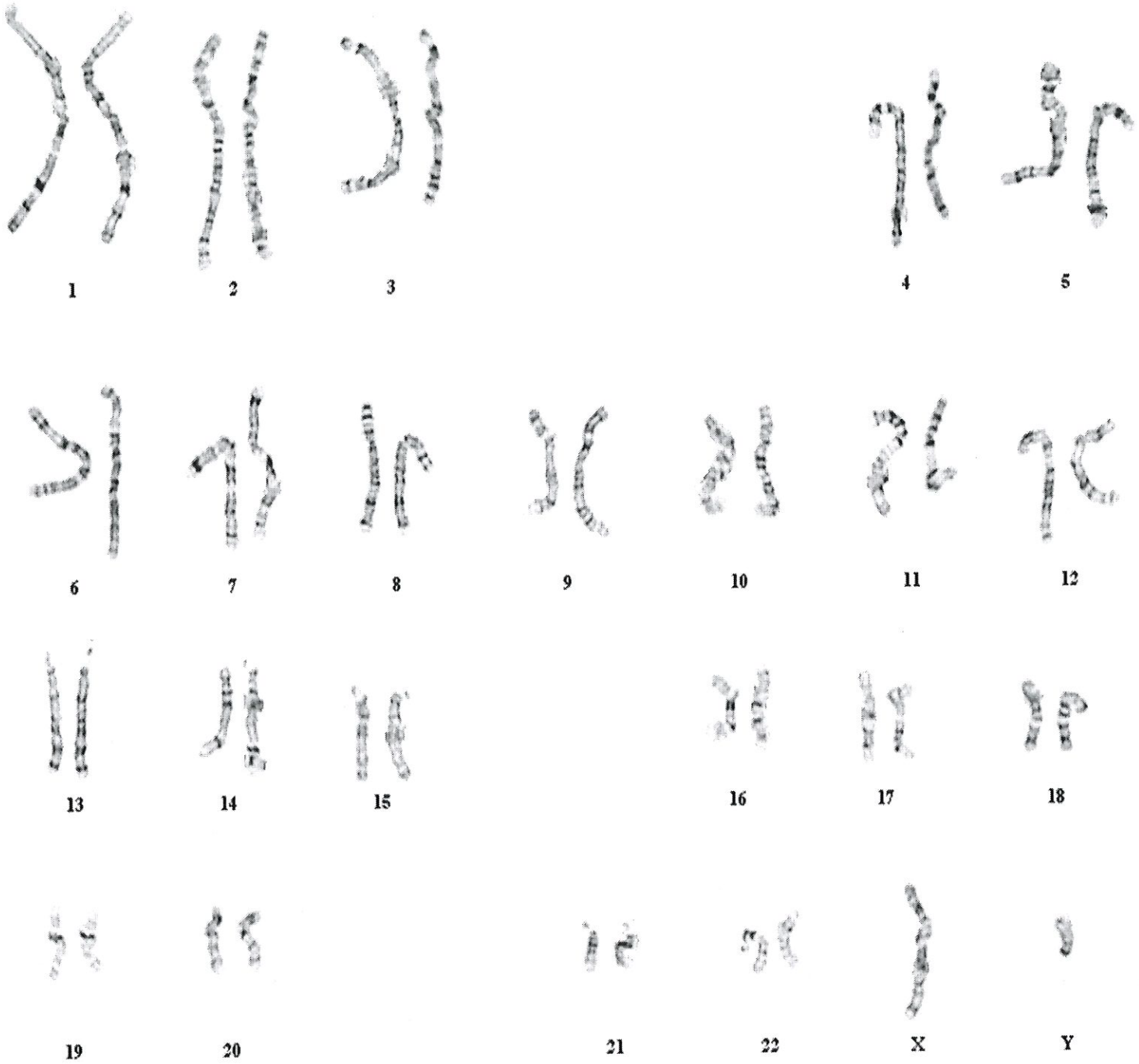
Signed: Joyce L. Murata-Collins

Joyce L. Murata-Collins, Ph.D.

Date: 04/03/2009

Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

genzyme



Specimen #: [redacted]
Specimen Type: BLDPER
Patient Name: Donor, 5972
Image ID: AKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 03/26/2009
Date Reviewed: 04/03/2009
Reviewed By: JLMC

genzyme
GENERAL
genetics

LabCorp
Laboratory Corporation of America

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

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
[redacted]		[redacted]	[redacted]	626-432-1681	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 5972	X					[redacted]	[redacted]	M	No

Account Address
Pacific Reproductive Svcs Pasa
65 N Madison Ave Ste 610
Pasadena CA 91101

Patient Address

Additional Information

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
02/04/09 14:00	02/05/09	02/10/09 17:06ET			

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

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
CBC With Differential/Platelet					
WBC	7.0		x10E3/uL	4.0 - 10.5	01
RBC	5.34		x10E6/uL	4.10 - 5.60	01
Hemoglobin	15.2		g/dL	12.5 - 17.0	01
Hematocrit	44.7		%	36.0 - 50.0	01
MCV	84		fL	80 - 98	01
MCH	28.4		pg	27.0 - 34.0	01
MCHC	34.0		g/dL	32.0 - 36.0	01
RDW	13.6		%	11.7 - 15.0	01
Platelets	288	✓	x10E3/uL	140 - 415	01
Neutrophils	63		%	40 - 74	01
Lymphs	29		%	14 - 46	01
Monocytes	6		%	4 - 13	01
Eos	1		%	0 - 7	01
Basos	1		%	0 - 3	01
Neutrophils (Absolute)	4.4		x10E3/uL	1.8 - 7.8	01
Lymphs (Absolute)	2.0		x10E3/uL	0.7 - 4.5	01
Monocytes(Absolute)	0.4		x10E3/uL	0.1 - 1.0	01
Eos (Absolute)	0.1		x10E3/uL	0.0 - 0.4	01
Baso (Absolute)	0.1		x10E3/uL	0.0 - 0.2	01
Hgb Frac. w/o Solubility					
Hgb A	**98.2**	**High** *	%	94.0 - 98.0	02
Hgb S	0.0		%	0.0	02
Hgb C	0.0		%	0.0	02
Hgb A2	1.8		%	0.7 - 3.1	02
Hgb F	0.0		%	0.0 - 2.0	02
Interpretation					
Normal adult hemoglobin present. ✓					02

2/12/09 Renewed OK to fill 8.

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

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

DONOR 5972, X		035-229-4909-0	Seq # 0964

FINAL REPORT

* Confirmed ≥ Lab Corp - statistically normal variation. Results are considered to