Donor 4715

Genetic Testing Summary

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

Last Updated: 09/24/20

Donor Reported Ancestry: Dutch, German, Italian 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
Hb Beta Chain-Related Hemoglobinopathy (including Sickle Cell and Beta Thalassemia)	Negative by genotyping for 28 mutations in the HBB gene	1/290
Cystic Fibrosis (CF) carrier screening (attached)	Negative by genotyping for 99 mutations in the CFTR gene	1/300
Spinal Muscular Atrophy (SMA) carrier screening (attached)	Negative for deletions of exon 7 in the SMN1 gene	1/600
Special Testing (attached)		
17-Alpha-Hydroxylase Deficiency (CYP17A1)	Negative by genotyping for 20 mutations in the CYP17A1 gene	Unknown
Non-Syndromic Hearing Loss (GJB2)	Negative by genotyping for 29 mutations in the GJB2 gene	1/62
Phenylalanine Hydroxylase Deficiency (PAH)	Negative by genotyping for 62 mutations in the PAH gene	1/189

*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	Male	Female
[redacted]	Name: DONOR 4715 DOB: [redacted] Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 09/06/2013 Date Received: 09/09/2013 Barcode: [redacted] Indication: Egg or Sperm Donor Test Type: The Counsyl Test	Not tested

Report Date: 09/12/2013

Counsyl Test Results Summary (Egg or Sperm Donor)

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses targeted genotyping and copy number analysis as described in the methods section on page 2 to determine carrier status associated with **3 diseases**. Please refer to page 3 for a complete list of diseases and genes included in this panel.

DONOR 4715

DONOR 4715's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Reproductive Risk Summary

No increased reproductive risks to highlight. Please refer to the following pages for detailed information about the results.

Clinical Notes

- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complimentary appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

ENTERED 9/13/13

Methods and Limitations

DONOR 4715: The Counsyl Test - targeted genotyping and copy number analysis.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 127 variants associated with 2 diseases. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

Copy number analysis: Targeted copy number analysis is used to determine the copy number of exon 7 of the SMN1 gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of SMN1 are carriers with two SMN1 genes on one chromosome and a SMN1 deletion on the other chromosome. In addition, a small percentage of SMA cases are caused by nondeletion mutations in the SMN1 gene. Thus, a test result of two SMN1 copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more SMN1 gene copies. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. The Counsyl test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet. Gynecol. 2007;109:229-37*).

This test was developed and its performance characteristics determined by Counsyl, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's workup. CLIA Number: **#05D1102604**.

Lab Directors:

Hyunseok Kang
H. Peter Kang, MD, MS, FCAP

Jelena Brezo
Jelena Brezo, PhD, FACMG

Counsyl

Male

Name: DONOR 4715
DOB: [redacted]

Female

Not tested

Diseases Tested

Cystic Fibrosis - Gene: CFTR. **Variants (99):** 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, 2143delT, 394delTT, 444delA, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), R117C, L206W, G330X, T338I, R352Q, S364P, G480C, C524X, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, R764X, Q890X, R1066C, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, 3199del6, 574delA, 663delT, 935delA, 936delTA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667del4, 3791delC, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, 3849+4A>G, Q359K/T360K. **Detection rate:** Northern European 91%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. **Variants (28):** Hb S, K17X, Q39X, 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), -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, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab. **Detection rate:** Northern European 83%.

Spinal Muscular Atrophy (copy number analysis only) - Gene: SMN1. **Variant (1):** SMN1 copy number. **Detection rate:** Northern European 95%.

Counsyl

Male

Name: DONOR 4715
DOB: [redacted]

Female

Not tested

Risk Calculations

Below are the risk calculations for all diseases tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Disease	DONOR 4715 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2017-01-19

Donor 4715

DOB: [redacted]
Gender: Male
Ethnicity:
Procedure ID: 77275
Kit Barcode: [redacted]
Specimen: Sperm, #78150
Specimen Collection: 2016-12-11
Specimen Received: 2016-12-13
Specimen Analyzed: 2017-01-19

TEST INFORMATION

Test: CarrierMap^{GEN} (Genotyping)
Panel: Custom Panel
Diseases Tested: 3
Genes Tested: 3
Mutations Tested: 111

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 4715 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

17-Alpha-Hydroxylase Deficiency (CYP17A1): Mutations (20): ♂ Genotyping | c.157_159delTTC (p.53delF), c.316T>C (p.S106P), c.715C>T (p.R239X), c.1024C>A (p.P342T), c.286C>T (p.R96W), c.1040G>A (p.R347H), c.1073G>A (p.R358Q), c.51G>A (p.W17X), c.340T>G (p.F114V), c.347A>T (p.D116V), c.1039C>T (p.R347C), c.1084C>T (p.R362C), c.1216T>C (p.W406R), c.985T>G (p.Y329D), c.601T>A (p.Y201N), c.81C>A (p.Y27X), c.287G>A (p.R96Q), c.1226C>G (p.P409R), c.1250T>G (p.F417C), c.278T>G (p.F93C)

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutations (29): ♂ Genotyping | c.167delT, c.235delC, c.312_325delGAAGTTCATCAAGG, c.358delGAG (p.120delE), c.35delG, c.370C>T (p.Q124X), c.427C>T (p.R143W), c.109G>A (p.V37I), c.231G>A (p.W77X), c.551G>C (p.R184P), c.71G>A (p.W24X), c.229T>C (p.W77R), c.269T>C (p.L90P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T, c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147K), c.-23+1G>A, c.550C>T (p.R184W), c.-259C>T

Phenylalanine Hydroxylase Deficiency (PAH): Mutations (62): ♂ Genotyping | c.1066-11G>A (IVS10-11G>A), c.1315+1G>A (IVS12+1G>A), c.1241A>G (p.Y414C), c.1222C>T (p.R408W), c.754C>T (p.R252W), c.1223G>A (p.R408Q), c.473G>A (p.R158Q), c.782G>A (p.R261Q), c.814G>T (p.G272X), c.143T>C (p.L48S), c.194T>C (p.I65T), c.896T>G (p.F299C), c.842C>T (p.P281L), c.838G>A (p.E280K), c.117C>G (p.F39L), c.3G>A (p.M1I), c.1A>G (p.M1V), c.611A>G (p.Y204C), c.721C>T (p.R241C), c.727C>T (p.R243X), c.1139C>T (p.T380M), c.926C>T (p.A309V), c.898G>T (p.A300S), c.734T>C (p.V245A), c.818C>T (p.S273F), c.997C>T (p.L333F), c.199T>C (p.S67P), c.1042C>G (p.L348V), c.136G>A (p.G46S), c.728G>A (p.R243Q), c.745C>T (p.L249F), c.581T>C (p.L194P), c.722G>T (p.R241L), c.829T>G (p.Y277D), c.899C>T (p.A300V), c.926C>A (p.A309D), c.1045T>C (p.S349P), c.1157A>G (p.Y386C), c.1169A>G (p.E390G), c.331C>T (p.R111X), c.241_256delACCCATTTGGATAAAC (p.T81fs), c.442-1G>A (IVS4-1G>A), c.463_464insTGTGTACC (p.R155fs), c.569T>G (p.V190G), c.682G>T (p.E228X), c.755G>A (p.R252Q), c.770G>T (p.G257V), c.781C>T (p.R261X), c.800A>G (p.Q267R), c.842+5G>A (IVS7+5G>A), c.856G>A (p.E286K), c.904delT (p.F302fs), c.913-7A>G (IVS8-7A>G), c.935G>T (p.G312V), c.1068C>G (p.Y356X), c.1238G>C (p.R413P), c.1301C>A (p.A434D), c.842+2T>A (IVS7+2T>A), c.764T>C (p.L255S), c.722G>A (p.R241H), c.533A>G (p.E178G), c.456_706+138del11653

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
17-Alpha-Hydroxylase Deficiency	♂ Brazilian: Unknown	54.55%	Unknown
	♂ Japanese: Unknown	45.45%	Unknown
Nonsyndromic Hearing Loss and Deafness: GJB2 Related	♂ Ashkenazi Jewish: 1/20	95.83%	1/480
	♂ Chinese: 1/100	82.26%	1/564
	♂ European: 1/53	82.47%	1/302
	♂ Ghanaian: Unknown	90.91%	Unknown
	♂ Indian: Unknown	66.98%	Unknown
	♂ Israeli: 1/16	93.10%	1/232
	♂ Japanese: 1/75	75.00%	1/300
	♂ Roma: Unknown	>99%	Unknown
	♂ United States: 1/34	45.22%	1/62
Phenylalanine Hydroxylase Deficiency	♂ Arab: Unknown	46.08%	Unknown
	♂ Ashkenazi Jewish: 1/224	44.44%	1/403
	♂ Brazilian: 1/71	56.41%	1/163
	♂ Chinese: 1/51	76.57%	1/218
	♂ Cuban: 1/71	69.64%	1/234
	♂ European: 1/51	73.00%	1/189
	♂ French Canadian: 1/80	76.27%	1/337
	♂ Iranian: 1/31	66.94%	1/94
	♂ Korean: 1/51	57.58%	1/120
	♂ Non-Ashkenazi Jewish: Unknown	63.64%	Unknown
	♂ Slovakian Gypsy: 1/39	>99%	<1/3,900
	♂ Spanish Gypsy: 1/4	93.75%	1/64
	♂ Taiwanese: Unknown	83.10%	Unknown
	♂ US Amish: 1/16	86.84%	1/122

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted]

Patient name/Donor Alias Donor # 4715

Donor # 4715-130906

Collection Date 09/06/2013

Date Received 09/07/2013

Patient DOB N/A

Specimen type Peripheral Blood

Accession # [redacted]

RESULTS

CYTOGENETIC ANALYSIS

Cells counted 61

Cells analyzed 5

Cells karyotyped 2

Modal chromosome # 46

Type of banding GTG

Band resolution 550

FISH

Probe(s) N/A

Nuclei scored N/A

KARYOTYPE 46,XY

INTERPRETATION

Normal male karyotype
No clonal numerical or structural abnormalities were identified. This normal cytogenetic result does not exclude the possibility of the presence of subtle rearrangements beyond the technical limits of detection with this test.

Comments

Wayne S. Stanley, Ph.D., FACMG
Clinical Cytogeneticist

4/20/13
Date

REVIEWED 9/24/13

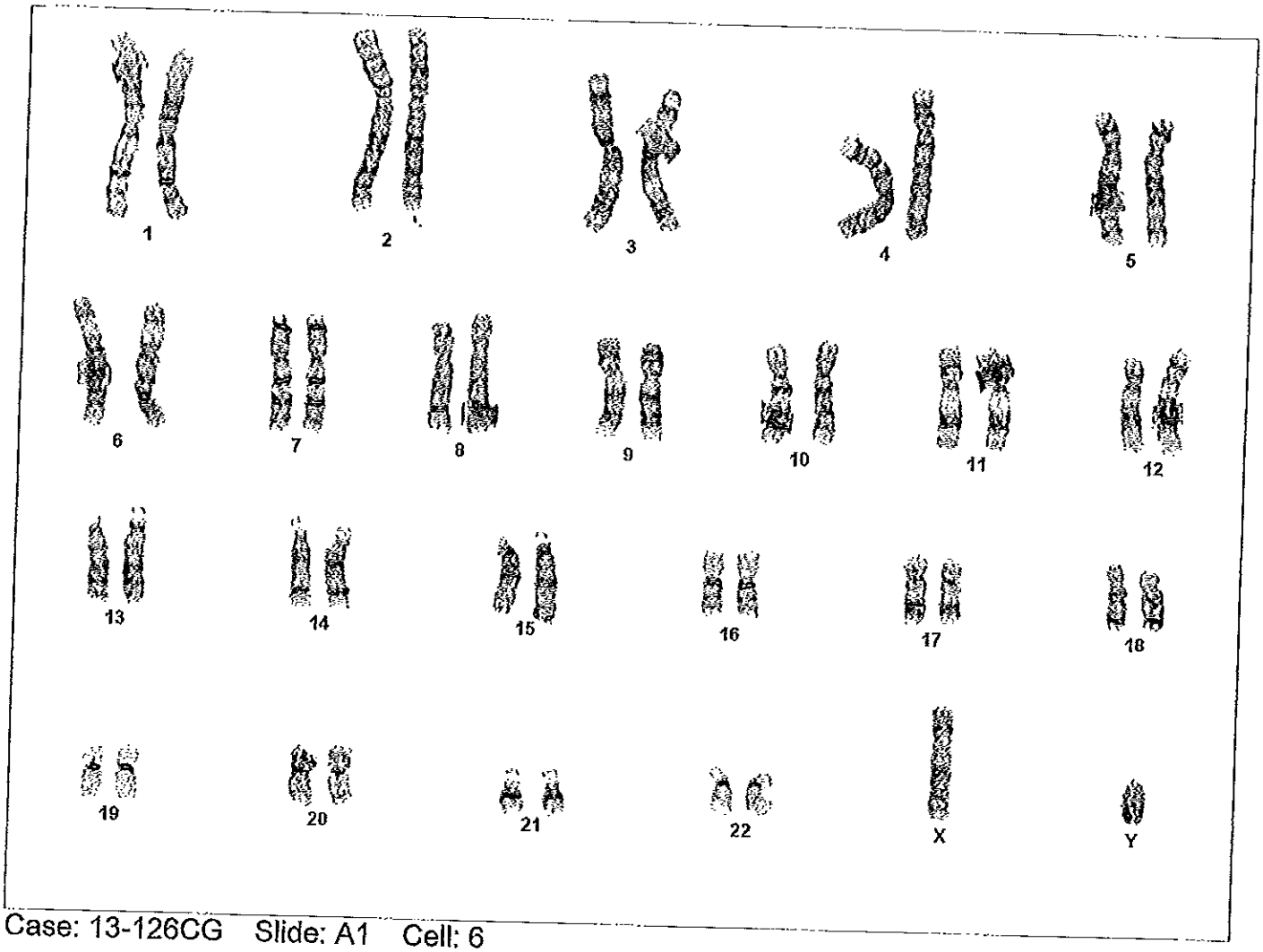
3015 Williams Drive | Fairfax, VA 22031 | 703.698.7355 | 800.552.4363

Genetics and IVF Preimplantation Genetics Laboratory

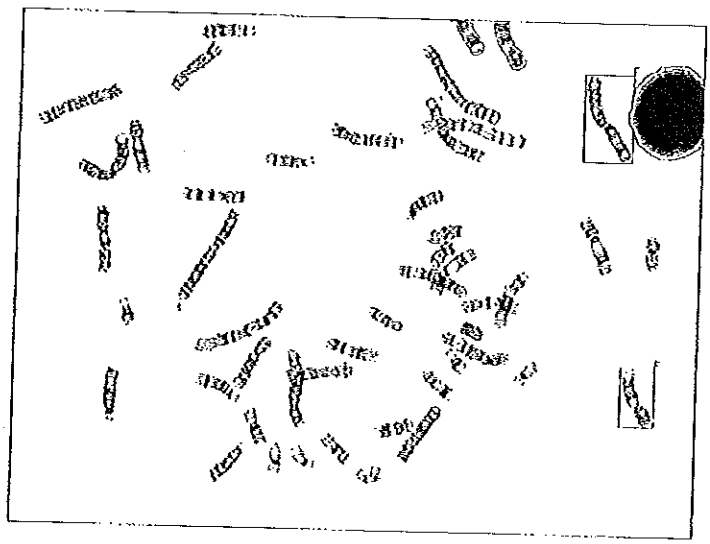
Patient name: DONOR # 4715

Case name: [redacted]

46,XY



Case: 13-126CG Slide: A1 Cell: 6



Quest Diagnostics

Report Status: Final

DONOR, ID4715

Patient Information	Specimen Information	Client Information
DONOR, ID4715 **DOB:** [redacted] **AGE:** [redacted] Gender: M Fasting: U Phone: NG Patient ID: [redacted]	Specimen: Requisition: [redacted] Collected: 09/06/2013 / 10:00 CDT Received: 09/07/2013 / 05:52 CDT Reported: 09/10/2013 / 09:04 CDT	Client #: 41550 AUS0000 STER FAIRFAX CRYOBANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT		4.19 L	4.20-5.80 Million/uL	IG
HEMOGLOBIN	13.6		13.2-17.1 g/dL	
HEMATOCRIT	40.7		38.5-50.0 %	
MCV	97.0		80.0-100.0 fL	
MCH	32.5		27.0-33.0 pg	
RDW	13.5		11.0-15.0 %	
HEMOGLOBIN A	97.3		>96.0 %	IG
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.7		1.8-3.5 %	
INTERPRETATION				
Normal phenotype.				

PERFORMING SITE:

IG QUEST DIAGNOSTICS-IRVING, 4770 REGENT BLVD., IRVING, TX 75063 Laboratory Director: ELISABETH S BROCKIE, DO, CLIA: 45D0697943

DATE ENTERED 9/13/13

CLIENT SERVICES: 866.697.8378

SPECIMEN: [redacted]