



Ordering Practice:

Practice Code: 926 Fairfax Cryobank

3015 Williams Drive, #110, Fairfax, VA,

SUMMARY OF RESULTS

22031, US

Physician:

Report Generated: 2015-12-09

Donor 5043

DOB:

Gender: Male

Ethnicity: African, European, and Native

American

Procedure ID: 35843

Kit Barcode:

Method: Genotyping Specimen: Blood, #37258 Specimen Collection: 2015-11-13 Specimen Received: 2015-11-16 Specimen Analyzed: 2015-12-09 Partner Not Tested

NO MUTATIONS IDENTIFIED

Donor 5043 was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

♂ Male

Panel: Fairfax Cryobank Panel V2, Diseases Tested: 22, Mutations Tested: 451, Genes Tested: 22, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31 D1054821
Lab Technician Bo Chu

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 >200 genes. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Spinal Muscular Atrophy: Spinal Muscular Atrophy is tested for via an Identity-by-State shared haplotype comparison algorithm. Detection is limited to haplotypes within our library of known carriers of the most common mutation (deletion of Exon 7).

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 mixup, 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.





● High Impact ● Treatment Benefits ● X-Linked ● Moderate Impact

Diseases & Mutations Assayed

	<u> </u>		
H T X M			Mutations
• 0 0 0	Alpha Thalassemia	11	of Genotyping SEA deletion, 11.1kb deletion, c.207C>A (p.N69K), c.223G>C (p.D75G), c.2T>C (p.M1T), c.95+2_95+6delTGAGG, c.207C>G (p.N69K), c.340_351delCTCCCCGCCGAG (p.L114_ E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G
	Beta Thalassemia	84	O' Genotyping c.124_127delTTCT (p.F42Lfs), c.17_18delCT, c.20delA (p.E7Gfs), c.217insA (p.S73Kfs), c.223+702_444+342del620insAAGTAGA, c.230delC, c.25_26delAA, c.315+1G>A, c.315+2T>C, c.316-197C>T, c.316-146T>G, c.315+745C>G, c.316-1G>A, c.316-1G>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, c.4delG (p.V2Cfs), c.51delC (p.K18Rfs), c.93-21G>A, c.92+1G>A, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>T, c50A>C, c.a-78g, c.a-79g, c.a-81g, c.A52T (p.K18X), c.c-137g, c.c-138t, c.c-151t, c.C118T (p.Q40X), c.G169C (p.G57R), c.G295A (p.V99M), c.G34A (p.V12I), c.G415C (p.A139P), c.G47A (p.W16X), c.G48A (p.W16X), c.t-80a, c.T2C (p.M1T), c.T75A (p.G25G), c.444+111A>G, c.g-29a, c.68_74delAAGTTGG, c.G92C (p.R31T), c.27_28insG, c.92+1G>T, c.92+1G>C, c.93-15T>G, c.93-1G>C, c.112delT, c.G113A (p.W38X), c.G114A (p.W38X), c.126delC, c.444+113A>G, c.250delG, c.225delC, c.383_385delAGG (p.Q128_A129delQAinsP), c.321_322insG (p.N109fs), c.316-1G>T, c.316-2A>C, c.316-106C>T, c.287_288insA (p.197fs), c.271G>T (p.E91X), c.203_204delTG (p.V68Afs), c.154delC (p.P52fs), c.135delC (p.F46fs), c.92+2T>A, c.92+2T>C, c.90C>T (p.G30G), c.59A>G (p.N10S), c.46delT (p.W16Gfs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.2T>G (p.M1R), c.1A>G (p.M1V), c.c-137t, c.c-136g, c.c-142t, c.c-140t
• 0 0 0	Bloom Syndrome	24	of Genotyping c.2207_2212delATCTGAinsTAGATTC (p.Y736Lfs), c.2407insT, c.557_559delCAA (p.S186X), c.1284G>A (p.W428X), c.1701G>A (p.W567X), c.1933C>T (p.Q645X), c.C2528T (p.T843I), c.C2695T (p.R899X), c.G3107T (p.C1036F), c.2923delC (p.Q975K), c.3558+1G>T, c.3875-2A>G, c.2074+2T>A, c.2343_2344dupGA (p.781EfsX), c.380delC (p.127Tfs), c.3564delC (p.1188Dfs), c.4008delG (p.1336Rfs), c.C947G (p.S316X), c.2193+1_2193+9del9, c.C1642T (p.Q548X), c.3143delA (p.1048NfsX), c.356_357delTA (p.Cys120Hisfs), c.4076+1delG, c.C3281A (p.S1094X)
•000	Canavan Disease	8	of Genotyping c.433-2A>G, c.A854C (p.E285A), c.C693A (p.Y231X), c.C914A (p.A305E), c.A71G (p.E24G), c.C654A (p.C218X), c.T2C (p.M1T), c.G79A (p.G27R)



нтх м			Mutations
	Cystic Fibrosis	147	Genotyping c.1029delC, 1153_1154insAT, c.1519_1521delATC (p.507dell), c.1521_1523delCTT (p.508delF), c.1545_1546delTA (p.Y515Xfs), c.1585-1G>A, c.164+12T>C, c.1680-886A>G, c.1680-1G>A, c.1766+1G>A, c.1766+1G>A, c.1766+1G>T, c.1766+5G>T, c.1818del84, c.1911delG, c.1923delCTCAAAACTinsA, c.1973delGAAATTCAATCCTinsAGAAA, c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2051_2052delAAinsG (p.K6845fsX38), c.2174insA, c.261delTT, c.2657+5G>A, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2738+1G>A, c.2051delT, c.3572delC, c.3140-26A>G, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2988+1G>A, c.3091delT, c.3717+12191C>T, c.3744delA, c.3373_3774insT (p.11258fs), c.442delA, c.489+1G>T, c.531delT, c.579+1G>T, c.579+5G>A (IVS4+5G>A), c.803delA (p.N268fs), c.805_806delAT (p.1269fs), c.933_935delCTT (p.311delF), c.A1645C (p.S549R), c.A2128T (p.K710X), c.C1000T (p.R334W), c.C1013T (p.T338I), c.C1364A (p.A455E), c.C1477T (p.Q493X), c.C1572A (p.C524X), c.C1654T (p.Q552X), c.C1657T (p.R553X), c.C1721A (p.F574H), c.C2125T (p.R709X), c.C223T (p.R75X), c.C2668T (p.Q890X), c.C3196T (p.R106C), c.C3276G (p.Y1092X), c.C3712T (p.Q1238X), c.C3764A (p.S1255X), c.C3909G (p.N1303K), c.G1040A (p.R347H), c.G1040C (p.R347P), c.G1438T (p.G480C), c.G1624T (p.G542X), c.G1646A (p.S549N), c.G1646T (p.S549), c.G1652A (p.G551D), c.G1675A (p.A559T), c.G1679C (p.R560T), c.G178T (p.E60X), c.G1865A (p.G622D), c.G254A (p.G85E), c.G271A (p.G91R), c.G346A (p.W108X), c.G3388T (p.R178M), c.G352A (p.G532A (p.G85E), c.G271A (p.G91R), c.G374T (p.E92X), c.G3209A (p.R1070Q), c.G3266A (p.W1089X), c.G3464C (p.D1152H), c.G350A (p.R117H), c.G350A (p.R117
•000	Familial Dysautonomia	4	of Genotyping c.2204+6T>C, c.C2741T (p.P914L), c.G2087C (p.R696P), c.C2128T (p.Q710X)
• 0 0 0	Familial Hyperinsulinism: Type 1: ABCC8 Related	10	of Genotyping c.3989-9G>A, c.4159_4161delTTC (p.1387delF), c.C4258T (p.R1420C), c.C4477T (p.R1493W), c.G2147T (p.G716V), c.G4055C (p.R1352P), c.T560A (p.V187D), c.4516G>A (p.E1506K), c.C2506T (p.Q836X), c.579+2T>A
•••	Fanconi Anemia: Type C	8	of Genotyping c.456+4A>T, c.67delG, c.C37T (p.Q13X), c.C553T (p.R185X), c.T1661C (p.L554P), c.C1642T (p.R548X), c.G66A (p.W22X), c.G65A (p.W22X)
• • 0 0	Gaucher Disease	6	of Genotyping c.84_85insG, c.A1226G (p.N409S), c.A1343T (p.D448V), c.C1504T (p.R502C), c.G1297T (p.V433L), c.G1604A (p.R535H)
	Glycogen Storage Disease: Type IA	13	σ' Genotyping c.376_377insTA, c.79delC, c.979_981delTTC (p.327delF), c.C1039T (p.Q347X), c.C247T (p.R83C), c.C724T (p.Q242X), c.G248A (p.R83H), c.G562C (p.G188R), c.G648T, c.G809T (p.G270V), c.A113T (p.D38V), c.975delG (p.L326fs), c.724delC





нтх м			Mutations
•000	Joubert Syndrome	2	o [®] Genotyping c.G35T (p.R12L), c.218G>A (p.R73H)
••00	Maple Syrup Urine Disease: Type 1B	6	of Genotyping c.G1114T (p.E372X), c.G548C (p.R183P), c.G832A (p.G278S), c.C970T (p.R324X), c.G487T (p.E163X), c.C853T (p.R285X)
	Maple Syrup Urine Disease: Type 3	8	of Genotyping c.104_105insA, c.G685T (p.G229C), c.A214G (p.K72E), c.A1081G (p.M361V), c.G1123A (p.E375K), c.T1178C (p.1393T), c.C1463T (p.P488L), c.A1483G (p.R495G)
• 0 0 0	Mucolipidosis: Type IV	4	o [®] Genotyping c.406-2A>G, c.G1084T (p.D362Y), c.C304T (p.R102X), c.244delC (p.L82fsX)
•000	Nemaline Myopathy: NEB Related	1	of Genotyping c.7434_7536del2502bp
•000	Niemann-Pick Disease: Type A	6	o ^a Genotyping c.996delC, c.G1493T (p.R498L), c.T911C (p.L304P), c.C1267T (p.H423Y), c.G1734C (p.K578N), c.1493G>A (p.R498H)
	Sickle-Cell Anemia	1	o Genotyping c.A20T (p.E7V)
• 0 0 0	Spinal Muscular Atrophy: SMN1 Linked	19	of Genotyping DEL EXON 7, c.22_23insA, c.43C>T (p.Q15X), c.91_92insT, c.305G>A (p.W102X), c.400G>A (p.E134K), c.439_443delGAAGT, c.558delA, c.585_586insT, c.683T>A (p.L228X), c.734C>T (p.P245L), c.768_778dupTGCTGATGCTT, c.815A>G (p.Y272C), c.821C>T (p.T274I), c.823G>A (p.G275S), c.834+2T>G, c.835-18_835-12delCCTTTAT, c.835G>T, c.836G>T
	Tay-Sachs Disease	76	Ø Genotyping c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.C532T (p.R178C), c.G533A (p.R178H), c.G805A (p.G269S), c.C1510T (p.R504C), c.G1496A (p.R499H), c.G509A (p.R170Q), c.A1003T (p.1335F), c.910_912delTTC (p.305delF), c.G749A (p.G250D), c.T632C (p.F211S), c.C629T (p.S210F), c.613delC, c.A611G (p.H204R), c.G598A (p.V200M), c.A590C (p.K197T), c.571-1G>T, c.C540G (p.Y180X), c.T538C (p.Y180H), c.G533T (p.R178L), c.C508T (p.R170W), c.C409T (p.R137X), c.T380G (p.L127R), c.346+1G>C, c.T116G (p.L39R), c.G78A (p.W26X), c.A1G (p.M1V), c.1495C>T (p.R499C), c.459+5G>A, c.1422-2A>G, c.535C>T (p.H179Y), c.1141delG (p.V381fs), c.796T>G (p.W266G), c.155C>A (p.S52X), c.426delT (p.F142fs), c.413-2A>G, c.570+3A>G, c.536A>G (p.H179R), c.1146+1G>A, c.736G>A (p.A246T), c.1302C>G (p.F434L), c.778C>T (p.P260S), c.1008G>T (p.Q336H), c.1385A>T (p.E462V), c.964G>A (p.D322N), c.340G>A (p.E114K), c.1432G>A (p.G478R), c.1178G>C (p.R393P), c.805+1G>C, c.1426A>T (p.R476X), c.623A>T (p.D208V), c.1537C>T (p.Q513X), c.1511G>T (p.R504L), c.1307_1308delTA (p.1436fs), c.571-8A>G, c.624_627delTCCT (p.D208fs), c.1211_1212delTG (p.L404fs), c.621T>G (p.D207E), c.1511G>A (p.R504H), c.1177C>T (p.R393X), c.2T>C (p.M1T), c.1292G>A (p.W431X), c.947_948insA (p.Y316fs), c.607T>G (p.W203G), c.1061_1063delTCT (p.F354_Y355delinsX), c.615delG (p.L205fs), c.805+2T>C, c.1123delG (p.E375fs), c.1121A>G (p.Q374R), c.1043_1046delTCAA (p.F348fs), c.1510delC (p.R504fs), c.1451T>C (p.L484P), c.964G>T (p.D322Y)
•000	Usher Syndrome: Type 1F	7	of Genotyping c.C733T (p.R245X), c.2067C>A (p.Y684X), c.C7T (p.R3X), c.C1942T (p.R648X), c.1101delT (p.A367fsX), c.2800C>T (p.R934X), c.4272delA (p.L1425fs)
• 0 0 0	Usher Syndrome: Type 3	5	σ' Genotyping c.T144G (p.N48K), c.T359A (p.M120K), c.300T>G (p.Y176X), c.C634T (p.Q212X), c.221T>C (p.L74P)
•000	Walker-Warburg Syndrome	1	♂ Genotyping c.1167insA (p.F390fs)

Tay-Sachs Enzyme Analysis

Patient Name: Donor 5043, .

Referring Physician: Harvey Stern, MD

Specimen #: Patient ID:

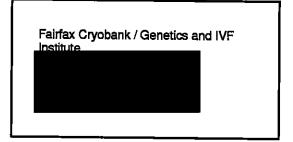
Client #:

DOB SSN: ***-**-

Date Collected: 07/22/2016 Date Received: 07/23/2016

Lab ID: Hospital ID:

Specimen Type: White Blood Cells



RESULTS:

Hexosaminidase Activity: 1251 nmol/mg protein

Hexosaminidase Percent A: 65.4

Plasma/Serum

WBC

Expected Non-Carrier Range:

Hex A <u>></u>54%

<u>></u>54%

Expected Carrier Range:

Hex A 20 - 49%

INTERPRETATION: NON CARRIER

This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

Under the direction of:

Philip R. Wyatt M.D., Ph.D

Date: 07/29/2016

Page 1 of 1

Testing Parformed At Eacterix Genetic Laboratories, LLC 2000 Vivigen Way Santa Fe, NM 87505 Phillip R. Wyatt M.D., Ph.D, Laboratory Director 1-800-848-4436

Nichols Institute, Chantilly

SPECIMEN INFORMATION

SPECIMEN:
REQUISITION:
LAB REF NO:

COLLECTED: 11/13/2015 00:00 RECEIVED: 11/16/2015 10:36 REPORTED: 11/25/2015 11:44 patient information 5043, DONOR

DOB: Age: SEX: M

REPORT STATUS Final

ORDERING PHYSICIAN

CLIENT INFORMATION 9595

Test Name	In Range	Out of Range	Reference Range	Lab
Hemoglobinopathy Evaluation				AMD
ERYTHROCYTE COUNT	5.06		4.20-5.80 Mill/uL	
HEMOGLOBIN	15.3		13.2-17.1 g/dL	
HEMATOCRIT	47.5		38.5-50.0 %	
MCV	93.7		80.0-100.0 FL	
MCH	30.2		27.0-33.0 pg	
RDW	14.5		11.0-15.0 %	
Hemoglobin A	97.9		>96.0 %	
Hemoglobin F	0.0		<2.0 %	
Hemoglobin A2	2.1		1.8-3.5 %	
Interpretation				

NORMAL PATTERN

By high-performance liquid chromatography (HPLC), there is a normal pattern of hemoglobins and normal levels of HbA2 and HbF are present.

No variant hemoglobins are observed. This is consistent with A/A phenotype.

If iron deficiency coexists with beta thalassemia trait HbA2 may be in the normal range.

Rare variant hemoglobins have been known to co-elute with hemoglobin A by high-performance liquid chromatography. If clinically indicated, Thalassemia and Hemoglobinopathy comprehensive is available (Test code 17365X[12658]).



1.

PATIENT INFORMATION 5043, DONOR

REPORT STATUS Final

Nichols Institute, Chantilly

ORDERING PHYSICIAN

DOB:

Age:

COLLECTED: 11/13/2015 00:00 REPORTED: 11/25/2015 11:44

ID:

Test Name

In Range

Out of Range

Reference Range

Lab AMD

Chromosome Analysis, Blood Chromosome Analysis, Blood

CYTOGENETIC RESULTS

Cytogenetic Reference #: CB-15-020288

Test Setup Date: 11/16/2015 Test Completion Date: 11/25/2015 Specimen Source: Peripheral Blood

Clinical History:Screening

Metaphases Counted:20 Analyzed:5

Karyotyped:2

Banding Level (G-bands):>=550

KARYOTYPE:

46, XY

INTERPRETATION and COMMENTS:

NORMAL MALE karyotype

Within the limits of standard cytogenetic methodologies, the chromosomes had normal G-banding patterns without apparent structural abnormality or rearrangement.

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods, or rare events such as low level mosaicism or very subtle rearrangements.

Electronic Signature on File

Nicole C. Christacos, Ph.D., FACMG Technical Director, Cytogenetics, 703-802-7156

Results Received

11/25/15

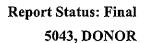
Reference lab accession: CB15020288EC

For more information on this test, go to http://education.questdiagnostics.com/faq/chromsblood

Performing Laboratory Information:

AMD QUEST DIAGNOSTICS INCORPORATED NICHOLS INSTITUTE 14225 NEWBROOK DR CHANTILLY VA 20151
Laboratory Director: PATRICK W. MASON, MD, PHD

ī





Patient Information	Specimen Information	Client Information	
5043, DONOR DOB: Gender: M Phone: NG Patient ID: NG	Specimen: Requisition: Lab Ref #: Collected: 11/13/2015 / 00:00 EST Received: 11/16/2015 / 10:36 EST Reported: 11/25/2015 / 11:51 EST	Client #: 9595 Not Provided C 3	

Cytogenetics Report

Chromosome Analysis, Blood - 14596

Lab:AMD

Case Number

CB-15-020288

Specimen Source

Peripheral Blood

Clinical History

Screening

Metaphases Counted

20

Metaphases Analyzed

20

wetaphases Analyzeu

5

Metaphases Karyotyped

2

Banding Level

>=550

Karyotype

46,XY

Interpretation and Comments

NORMAL MALE karyotype

Within the limits of standard cytogenetic methodologies, the chromosomes had normal G-banding patterns without apparent structural abnormality or rearrangement.

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods, or rare events such as low level mosaicism or very subtle rearrangements.

Signature

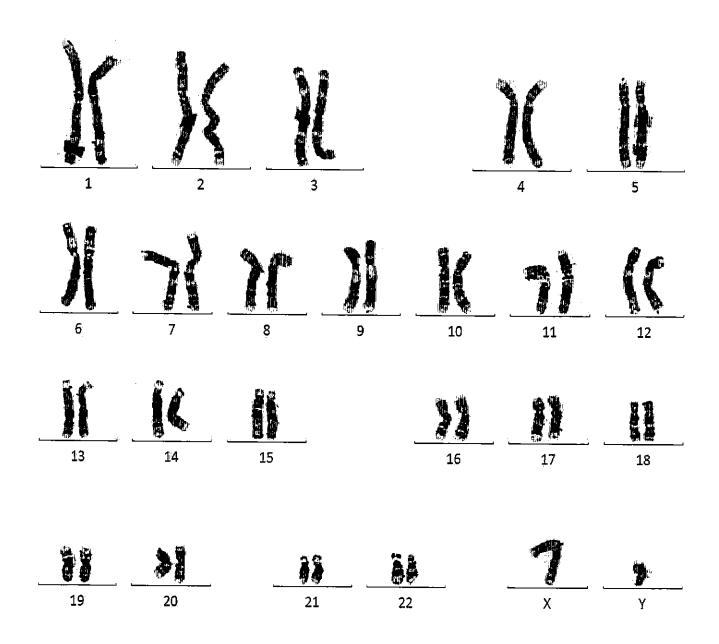
Electronic Signature on File

Nicole C. Christacos, Ph.D., FACMG Technical Director, Cytogenetics, 703-802-7156





Patient Information	Specimen Information	Client Information
5043, DONOR	Specimen: CH313884G Collected: 11/13/2015 / 00:00 E	Client #: 9595 Not Provided
DOB: Gender: M Patient ID: NG	Received: 11/16/2015 / 10:36 ES Reported: 11/25/2015 / 11:51 ES	ST



PERFORMING SITE:

AMD QUEST DIAGNOSTICS/NICHOLS CHANTILLY, 14225 NEWBROOK DRIVE, CHANTILLY, VA 20151-0841 Laboratory Director: Patrick W Mason, MD., Ph.D., CLIA: 49D0221801 This is supplemental to your standard report.

CLIENT SERVICES: 866-677-0742 (Opt#1)

SPECIMEN: CH313884G

PAGE 2 OF 2