

Baylor Miraca
Genetics Laboratories

BAYLOR MIRACA GENETICS LABORATORIES

2450 Holcombe Blvd - Houston, TX 77021 - 1-800-411-4363

Fax: 713-798-2787 - www.bmgd.com - genetest@bcm.edu

Patient Name:	DONOR 4622	Lab Number:		Fairfax Cryobank Sendouts-Fairfax
Date of birth:		Family #:		3015 Williams Dr #110
Gender:	M	Date Collected:	08/10/2015	Tel No.: 703-876-3869
Hospital/MR #:		Date Received:	08/11/2015	Fax No.: 703-698-3933
Accession #:		Date Reported:	8/27/2015	CC: Suzanne Seitz Fax# 703-698-3933
Sample Type:	EXT DNA			CC: Harvey Stern Fax# 703-698-3933
Test Code:	3356			
Indication:	Asymptomatic/Positive Family History			

VLCAD Deficiency
ACADVL Sequence Analysis
Familial Mutation/Variant Analysis



62817-4390128

RESULTS:

A heterozygous c.553G>A (p.G185S) familial pathogenic variant was detected.

Pathogenic Variant(s)/ Mutation(s)

Nucleotide Change	Amino Acid Change	Location	Zygoty	Reference(s) / Comment(s)
c.553G>A	p.G185S	exon 7	heterozygous	PMID: 9973285

INTERPRETATION:

Test results should be interpreted in the context of the patient's clinical presentation and family history. Previous sequence analysis performed in this laboratory detected a heterozygous c.553G>A (p.G185S) pathogenic variant in this individual's child who had abnormal newborn screening results suggestive of VLCAD deficiency (Lab# 56744). This variant has been reported in patients with VLCAD deficiency (PMID: 9973285). We were requested to investigate this individual for the c.553G>A (p.G185S) pathogenic variant.

We received extracted DNA and the DNA sample was PCR amplified for the relevant region and then sequenced in the forward and reverse directions. Other regions of the ACADVL gene were not examined. This analysis indicates that this individual is heterozygous for the c.553G>A (p.G185S) pathogenic variant. Thus, this individual is a carrier of the familial c.553G>A (p.G185S) pathogenic variant.

Clinical correlation and genetic counseling are recommended. Targeted sequence analysis (test code 3356) is available for that this individual's relatives.

METHOD:

The region(s) containing the previously identified familial mutations/variants in the ACADVL (NM_000182) gene were PCR amplified and then sequenced in the forward and reverse directions using automated fluorescent dideoxy sequencing methods. Nucleotide 1 corresponds to the A of the start codon ATG.

W. Craig M.D.

William J. Craigen, M.D., Ph.D., FACMG
ABMG Certified Clinical Geneticist and Biochemical Geneticist
Medical Director

Leo-Jun C. Wong

Leo-Jun C. Wong, Ph.D., FACMG
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This test was developed and its performance characteristics determined by Baylor Miraca Genetics Laboratories (CAP# 2109314/CLIA# 480060090). 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.

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
3015 Williams Drive, #110, Fairfax, VA,
22031, US
Physician: Suzanne Seitz
Report Generated: 2015-09-08
Report Updated: 2015-09-08

Donor 4622

DOB: [REDACTED]
Gender: Male
Ethnicity: Other
Procedure ID: 29150
Kit Barcode: [REDACTED]
Method: Genotyping
Specimen: Sperm, #30392
Specimen Collection: 2015-08-25
Specimen Received: 2015-08-28
Specimen Analyzed: 2015-09-08

Partner Not Tested

SUMMARY OF RESULTS**NO MUTATIONS IDENTIFIED**

Donor 4622 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 , Diseases Tested: 21, Mutations Tested: 382, Genes Tested: 22, Null Calls: 0

Assay performed by 
Reprogenetics

CLIA ID: 31D1054821

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 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.

Diseases & Mutations Assayed

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

H	T	X	M	Disease	#	Mutations
●	○	○	○	Alpha Thalassemia	10	♂ Genotyping SEA deletion, 11.1 kb deletion, c.207C>A (p.N69K), c.223G>C (p.D75G), c.2T>C (p.M1T), 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	83	♂ Genotyping 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, c.-50A>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.L97fs), 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.N20S), 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
●	○	○	○	Bloom Syndrome	24	♂ 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)
●	○	○	○	Canavan Disease	8	♂ 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)

H	T	X	M	Disease	#	Mutations
<input checked="" type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	Cystic Fibrosis	130	♂ Genotyping c.1029delC, 1153_1154insAT, c.1519_1521delATC (p.507delI), 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>T, c.1766+5G>T, c.1818del84, c.1911delG, c.1923delCTCAAACTinsA, c.1973delGAAATCAATCTinsAGAAA, c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2051_2052delAAinsG (p.K684SfsX38), c.2174insA, c.261delTT, c.2657+5G>A, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2988+1G>A, c.3039delC, c.3140-26A>G, c.325delTAinsG, c.3527delC, c.3535delACCA, c.3691delT, c.3717+12191C>T, c.3744delA, c.3773_3774insT (p.L1258fs), 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.I269fs), 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.P574H), c.C2125T (p.R709X), c.C223T (p.R75X), c.C2668T (p.Q890X), c.C3196T (p.R1066C), c.C3276G (p.Y1092X), c.C3472T (p.R1158X), c.C3484T (p.R1162X), c.C349T (p.R117C), c.C3587G (p.S1196X), 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.S549I), 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.G274T (p.E92X), c.G3209A (p.R1070Q), c.G3266A (p.W1089X), c.G3454C (p.D1152H), c.G350A (p.R117H), c.G3611A (p.W1204X), c.G3752A (p.S1251N), c.G3846A (p.W1282X), c.G3848T (p.R1283M), c.G532A (p.G178R), c.G988T (p.G330X), c.T1090C (p.S364P), c.T3302A (p.M1101K), c.T617G (p.L206W), c.C14T (p.P5L), c.G19T (p.E7X), c.G171A (p.W57X), c.313delA (p.I105fs), c.G328C (p.D110H), c.580-1G>T, c.G1055A (p.R352Q), c.C1075A (p.Q359X), c.C1079A (p.T360K), c.T1647G (p.S549R), c.1976delA (p.N659fs), c.C2290T (p.R764X), c.2737_2738insG (p.Y913X), c.3067_3072delATAGTG (p.I1023_V1024delT), c.3536_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.G3808A (p.D1270N), c.G4056C (p.Q1352H), c.C4364G (p.S1455X), c.C4003T (p.L1335F), c.G2538A (p.W846X), c.C200T (p.P67L), c.C4426T (p.Q1476X), c.1116+1G>A, c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.T263G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.C658T (p.Q220X), c.C868T (p.Q290X), c.1526delG (p.G509fs), c.2908+1085-3367+260del7201, c.C11A (p.S4X), c.A3700G (p.I1234V), c.A416T (p.H139L), c.T366A (p.Y122X)
<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	Familial Dysautonomia	4	♂ Genotyping c.2204+6T>C, c.C2741T (p.P914L), c.G2087C (p.R696P), c.C2128T (p.Q710X)
<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	Familial Hyperinsulinism: Type 1: ABCC8 Related	10	♂ Genotyping c.3989-9G>A, c.4159_4161delITC (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
<input checked="" type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	Fanconi Anemia: Type C	8	♂ 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)
<input checked="" type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	Gaucher Disease	6	♂ Genotyping c.84_85insG, c.A1226G (p.N409S), c.A1343T (p.D448V), c.C1504T (p.R502C), c.G1297T (p.V433L), c.G1604A (p.R535H)
<input checked="" type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	Glycogen Storage Disease: Type IA	13	♂ Genotyping c.376_377insTA, c.79delC, c.979_981delITC (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
<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	Joubert Syndrome	1	♂ Genotyping c.G35T (p.R12L)
<input checked="" type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	Maple Syrup Urine Disease: Type 1B	6	♂ 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)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Maple Syrup Urine Disease: Type 3	8	♂ Genotyping c.104_105insA, c.G685T (p.G229C), c.A214G (p.K72E), c.A1081G (p.M361V), c.G1123A (p.E375K), c.T1178C (p.I393T), c.C1463T (p.P488L), c.A1483G (p.R495G)
●	○	○	○	Mucopolidosis: Type IV	4	♂ Genotyping c.406-2A>G, c.G1084T (p.D362Y), c.C304T (p.R102X), c.244delC (p.L82fsX)
●	○	○	○	Nemaline Myopathy: NEB Related	1	♂ Genotyping c.7434_7536del2502bp
●	○	○	○	Niemann-Pick Disease: Type A	6	♂ Genotyping c.996delC, c.G1493T (p.R498L), c.T911C (p.L304P), c.C1267T (p.H423Y), c.G1734C (p.K578N), c.1493G>A (p.R498H)
●	○	○	○	Spinal Muscular Atrophy: SMN1 Linked	19	♂ 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_835-12delCCTTAT, c.835G>T, c.836G>T
●	○	○	○	Tay-Sachs Disease	30	♂ 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.I335F), 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)
●	○	○	○	Usher Syndrome: Type 1F	6	♂ Genotyping c.C733T (p.R245X), c.2067C>A (p.Y684X), c.C7T (p.R3X), c.C1942T (p.R648X), c.2800C>T (p.R934X), c.4272delA (p.L1425fs)
●	○	○	○	Usher Syndrome: Type 3	4	♂ Genotyping c.T144G (p.N48K), c.T359A (p.M120K), c.300T>G (p.Y176X), c.C634T (p.Q212X)
●	○	○	○	Walker-Warburg Syndrome	1	♂ Genotyping c.1167insA (p.F390fs)

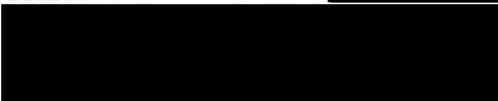
LABORATORY REPORT

IKONMAIL



12738

FAIRFAX CRYOBANK-



Collected Date 10/01/2012 Received Date 10/01/2012 1847 Collection Time 1330
Reported Date 10/05/2012 Other I.D. Fasting U

REQUISITION NO. 127380000000086 PHYSICIAN

PATIENT ID,4622 DONOR DOB AGE SEX M PATIENT ID ACCESSION NO.

REQUESTS RESULTS OUT OF RANGE RESULTS REFERENCE RANGE UNITS FN

* Previously Reported on: 10/04/2012 @ 8:13PM *

CBC W/DIFF, W/PLT

Table with 4 columns: Test Name, Value, Reference Range, Units. Includes WBC, RBC, Hemoglobin, Hematocrit, MCV, MCH, MCHC, RDW, Platelet Count, etc.

ENTERED (10-2-13)

Table with 4 columns: Test Name, Value, Reference Range, Units. Includes AST, Cholesterol, ALT, Blood Group AND RH TYPE.

Sonora Quest Laboratories no longer performs "weak D" or "Du" testing routinely. It is no longer a regulatory requirement, nor is it recommended by the American Association of Blood Banks (AABB), except in testing of donor units.

HEMOGLOBINOPATHY EVALUATION

Table with 5 columns: Test Name, Value, Reference Range, Units, Flag. Includes RED BLOOD CELL COUNT, Hemoglobin, MCV, RDW.

ID,4622 DONOR - E90278930 - REPRINT REPORT

CONTINUED ON PAGE 2

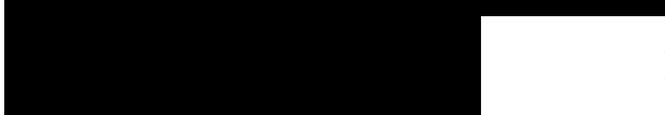
LABORATORY REPORT

IKONMAIL



12738

FAIRFAX CRYOBANK



Collected Date 10/01/2012	Received Date 10/01/2012 1847	Collection Time 1330
Reported Date 10/05/2012	Other I.D.	Fasting U

REQUISITION NO. 127380000000	PHYSICIAN 086
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PATIENT ID, 4622 DONOR	DOB [REDACTED]	AGE [REDACTED]	SEX M	PATIENT ID	ACCESSION NO. [REDACTED]
REQUESTS	RESULTS	OUT OF RANGE RESULTS	REFERENCE RANGE	UNITS	FN

HEMOGLOBIN A	97.3		94.5-98.5	%	G
HEMOGLOBIN A2	2.7		1.8-3.5	%	G
PATHOLOGIST INTERPRETATION					G

Normal phenotype. Reviewed by Dr. Louis Novoa-Takara

Evaluation performed by High Performance Liquid Chromatography (HPLC). Gel electrophoresis performed only when indicated.

Hemoglobinopathy Evaluation examines specimens for common variant hemoglobins such as S, C, and E as well as most other less common variant hemoglobins. Many, but not all, thalassemic disorders may be detected.

If, in spite of normal findings, a clinical suspicion of a hemoglobin abnormality persists please contact the laboratory.

RDWSD	43.4		36.0-55.0	%	G
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TESTS ORDERED: AST;CHOLESTEROL;ALT;BLOOD GROUP AND RH TYPE
CBC W/DIFF,W/PLT;HEMOGLOBINOPATHY EVALUATION

Unless otherwise noted,
all testing performed at: Sonora Quest Laboratories
1255 West Washington Street
Tempe, AZ 85281
602.685.5000 or 800.766.6721

= Test performed at: Banner Good Samaritan Medical Center
1111 E. McDowell Road
Phoenix, AZ 85006
602.239.2000

ENTERED
10-7-12

END OF REPORT. PRINTED 10/05/2012 @ 08:31:24 AM
ID,4622 DONOR - E90278930 - REPRINT REPORT



GENETICS & IVF
Institute

ENTERED
10.1.12

Cytogenetic Report

Client Fairfax Cryobank [REDACTED]

Address [REDACTED]

Reporting Phone # [REDACTED]

Fax # [REDACTED]

Email [REDACTED]

Patient name/Donor Alias Donor # 4622

Patient DOB N/A

Donor # 4622-121001

Specimen type Peripheral Blood

Collection Date 10/01/2012

Accession # [REDACTED]

Date Received 10/02/2012

RESULTS

CYTOGENETIC ANALYSIS

FISH

Cells counted 20

Type of banding GTG

Probe(s) N/A

Cells analyzed 5

Band resolution 550

Nuclei scored N/A

Cells karyotyped 2

Modal chromosome # 46

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

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10/16/12
Date

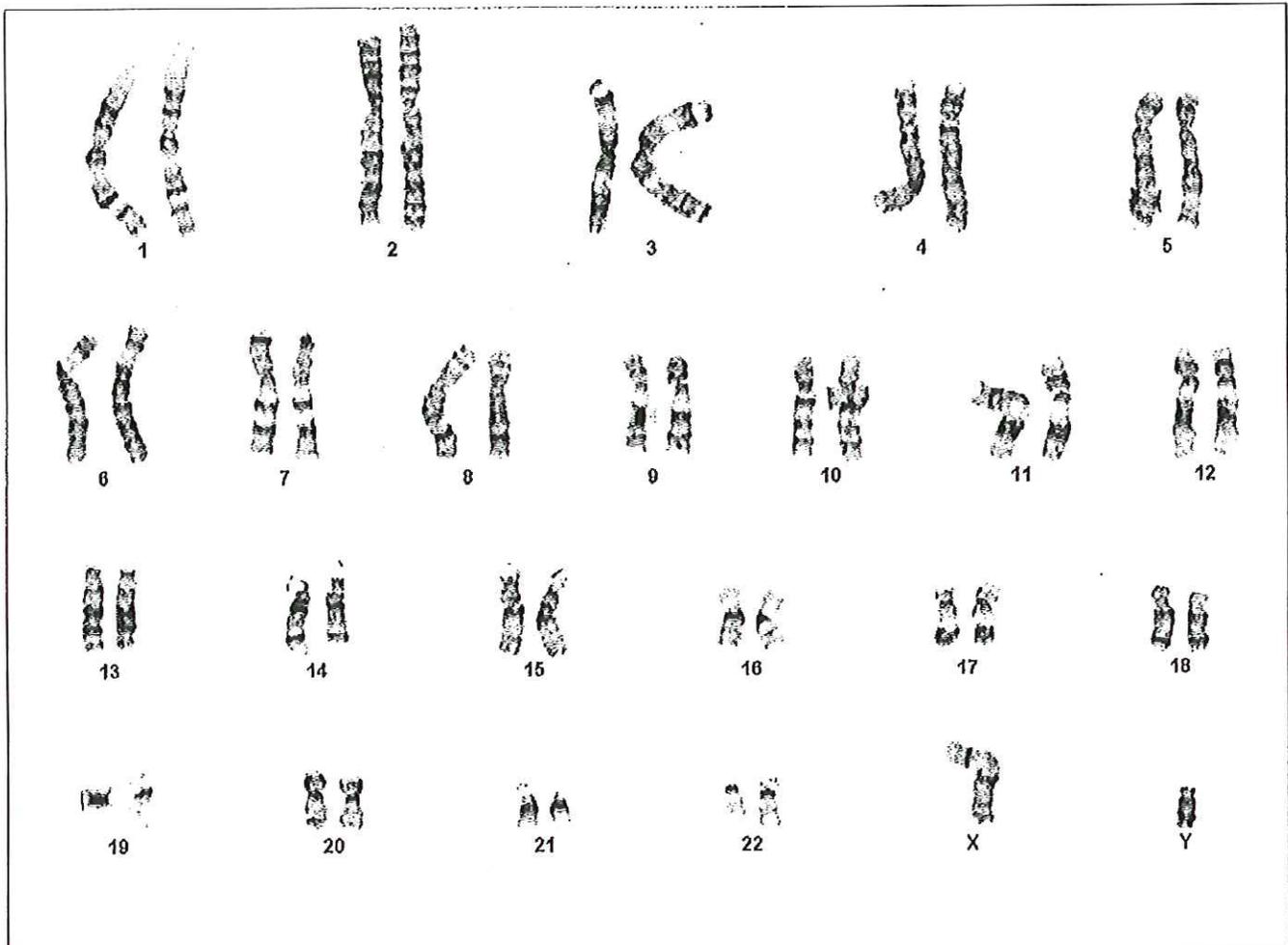
Genetics and IVF Preimplantation Genetics Laboratory

Patient name: DONOR # 4622

Case name: [REDACTED]

ENTERED
12-7-14

46,XY



Case: 12-143CG Slide: A1 Cell: 7

