



LabCorp RTP
1912 TW Alexander Drive
RTP, NC 27709-0150

Phone: 800-735-4087

Specimen Number 257-225-9007-0		Patient ID		Control Number	Account Number 90000999	Account Phone Number 336-436-8645	Route 00
SAMPLE REPORT				Account Address LabCorp Test Master Test Account 3060 South Church Street Burlington NC 27215			
Patient First Name 511075		Patient Middle Name					
Patient SS#		Patient Phone		Total Volume			
Age (Y/M/D) 31/03/01	Date of Birth 06/12/85	Sex M	Fasting				
Patient Address				Additional Information NORMAL REPORT			
Date and Time Collected 09/13/16 00:00	Date Entered 09/13/16	Date and Time Reported		Physician Name	NPI	Physician ID	

Tests Ordered
Chromosome Routine Rfx DAZ; Count 15-20 cells, 2 Karyotype; Chromosome Blood Routine 88230

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
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Chromosome Routine Rfx DAZ

Specimen Type	Comment:	01
BLOOD		
Cells Counted	20	01
Cells Analyzed	20	01
Cells Karyotyped	2	01
GTG Band Resolution Achieved	500	01
Cytogenetic Result	Comment:	01
46,XY		
Interpretation	Comment:	01
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.

Director Review:	Comment:	01
Inder K. Gadi, PhD, FACMG		

Reflex		01
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Y Deletion analysis is being performed. Final report will follow under separate cover.

Infertility-Male, Y Chrom Anal		02
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Molecular analysis report has been mailed.
Results of this test are for Investigational Purposes Only. The performance characteristics of this assay have been determined by LabCorp. The result should not be used as a diagnostic procedure without confirmation of the diagnosis by another medically established diagnostic product or procedure.

SAMPLE REPORT, 511075		257-225-9007-0	Seq # 0000
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10/04/16 12:20 ET

DUPLICATE FINAL REPORT

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LabCorp RTP
1912 TW Alexander Drive
RTP, NC 27709-0150

Phone: 800-735-4087

Patient Name					Specimen Number		
SAMPLE REPORT, 511075					257-225-9007-0		
Account Number	Patient ID	Control Number	Date and Time Collected	Date Reported	Sex	Age(Y/M/D)	Date of Birth
90000999			09/13/16 00:00		M	31/03/01	06/12/85

01	YU	LabCorp RTP 1904 TW Alexander Drive Suite C, RTP, NC 27709-0153	Dir: Arundhati Chatterjee, MD
02	TG	LabCorp RTP 1912 TW Alexander Drive, RTP, NC 27709-0150	Dir: Arundhati Chatterjee, MD
For inquiries, the physician may contact Branch: 800-222-7566 Lab: 800-735-4087			

SAMPLE REPORT, 511075		257-225-9007-0	Seq # 0000
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10/04/16 12:20 ET

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DOC1 Ver: 1.49

LCLS Specimen Number: 257-225-9007-0

Patient Name: **SAMPLE REPORT, 511075**

Date of Birth: 06/12/1985

Gender: M

Patient ID:

Lab Number: YU16-73627 L

Indications:

Account Number: 90000999

Ordering Physician:

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 09/13/2016

Date Received: 09/16/2016

Date Reported: **09/16/2016**

Test: **Chromosome Routine Rfx DAZ**

Cells Counted: 20

Cells Analyzed: 20

Cells Karyotyped: 2

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.



Inder K. Gadi, PhD, FACMG
Board Certified Cytogeneticist

Arundhati Chatterjee, MD
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive, RTP, NC, 27709-0153 (800) 345-4363

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Arundhati Chatterjee, MD.
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September 21, 2016



LabCorp Test Master
Test Account
3060 South Church Street
Burlington, NC 27215

Test Results of: SAMPLE REPORT, 511075
DOB: 06/12/1985 Age: 31.2 Y Sex: M
Collected on: 09/13/2016
Received on: 09/13/2016
Reported on: 09/21/2016

Branch Number: POE00
Account Number: 90000999
Specimen Number: 257-225-9007-0
Specimen Type: Blood

Patient ID#:

Physician:

Test: DAZ-INFERTILITY, DNA ANALYSIS

Result:

POSITIVE FOR Y CHROMOSOME DELETION
Deletion encompasses (DAZ & SPGY)

Interpretation:

A deletion of the proximal long arm of the Y chromosome associated with male infertility was detected. The deleted Y chromosome markers are indicated above. All other azoospermia associated loci that were analyzed are present. Genetic counselors are available for health care providers at 1-800-345-GENE to answer any questions about this test or other tests that may be indicated for this patient.

Background:

Genes associated with normal spermatogenesis are located on the proximal long arm of the Y chromosome. The loss of the proximal Y long arm has been reported in approximately 7% of infertile men. These *de novo* deletions occur during paternal meiosis. The deletions of the distal AZFc region are associated with oligospermia whereas more proximal deletions of the AZFa and AZFb regions are associated with azoospermia (Sertoli cell-only syndrome). Klinefelter (47,XXY) syndrome and cystic fibrosis mutations (particularly the 5T allele) are also associated with azoospermia. Balanced chromosomal rearrangements are also associated with oligospermia.

Methodology:

The analysis detects deletions in the AZFa (DYS273 and DYS275), AZFb (DYS209 and DYS224) and AZFc (DAZ and SPGY) regions of the Y chromosome by multiplex polymerase chain reaction (PCR) and gel electrophoresis. Some small, rare deletions may not be detected by this assay. Molecular-based testing is highly accurate, but as in any laboratory test, rare diagnostic errors may occur.

Comment:

Results of this test are for investigational purposes only. The performance characteristics of this assay have been determined by LabCorp. The result should not be used as a diagnostic procedure without confirmation of the diagnosis by another medically established diagnostic product or procedure.

References:

Pryor JL, et al. Microdeletions in the Y chromosome of infertile men. *N Engl J Med.* 1997;336:534-9.

Mulhall JP, Reijo R, Alagappan R, Brown L, Page D, Carson R, Oates RD. Azoospermic men with deletion of the DAZ gene cluster are capable of completing spermatogenesis: fertilization, normal embryonic development and pregnancy occur when retrieved testicular spermatozoa are used for intracytoplasmic sperm injection. *Hum Reprod.* 1997;12:503-8.

Simoni M. Molecular diagnosis of Y chromosome microdeletions in Europe: state-of-the-art and quality control. *Hum Reprod.* 2001;16:402-9.

Simoni M, Bakker E, and Krausz C. EAA/EMQN best practice guidelines for molecular diagnosis of Y-chromosome microdeletions. State of the art 2004. *Intl J Androl.* 2004;27:240-9.

Results Released By: Hongli Zhan, Ph.D., Director
Report Released By: Amy C. Dexter, MS, CGC, Genetic Counselor

Arundhati Chatterjee, M.D.
Medical Director

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

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LabCorp RTP
1912 TW Alexander Drive
RTP, NC 27709-0150

Phone: 800-735-4087

Specimen Number 257-225-9008-0		Patient ID		Control Number	Account Number 90000999	Account Phone Number 336-436-8645	Route 00
SAMPLE REPORT				Account Address LabCorp Test Master Test Account 3060 South Church Street Burlington NC 27215			
Patient First Name 511075		Patient Middle Name					
Patient SS#		Patient Phone		Total Volume			
Age (Y/M/D) 31/03/01	Date of Birth 06/12/85	Sex M	Fasting				
Patient Address				Additional Information ABNORMAL REPORT			
Date and Time Collected 09/13/16 00:00	Date Entered 09/13/16	Date and Time Reported		Physician Name	NPI	Physician ID	

Tests Ordered
Chromosome Routine Rfx DAZ; Count 15-20 cells, 2 Karyotype; Chromosome Blood Routine 88230

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Chromosome Routine Rfx DAZ					
Specimen Type	Comment:				01
BLOOD					
Cells Counted	20				01
Cells Analyzed	20				01
Cells Karyotyped	2				01
GTG Band Resolution Achieved	500				01
Cytogenetic Result	Comment:				01
47,XXY					
Interpretation	Comment:				01
KLINFELTER SYNDROME					

Cytogenetic analysis of PHA stimulated cultures revealed a male karyotype with an extra X chromosome in all GTG banded metaphases. This is consistent with a diagnosis of Klinefelter syndrome (tall stature, infertility, gynecomastia).

Genetic counseling is recommended.

Director Review: Comment: 01
Inder K. Gadi, PhD, FACMG
Reflex
Deletion analysis not indicated. 01

01 YU LabCorp RTP Dir: Arundhati Chatterjee, MD
1904 TW Alexander Drive Suite C, RTP, NC 27709-0153
For inquiries, the physician may contact Branch: 800-222-7566 Lab: 800-735-4087

SAMPLE REPORT, 511075		257-225-9008-0	Seq # 0000
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LCLS Specimen Number: 257-225-9008-0

Patient Name: **SAMPLE REPORT, 511075**

Date of Birth: 06/12/1985

Gender: M

Patient ID:

Lab Number: YU16-73625 L

Indications:

Account Number: 90000999

Ordering Physician:

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 09/13/2016

Date Received: 09/16/2016

Date Reported: **09/16/2016**

Test: **Chromosome Routine Rfx DAZ**

Cells Counted: 20

Cells Analyzed: 20

Cells Karyotyped: 2

Band Resolution: 500

CYTOGENETIC RESULT: 47,XXY

INTERPRETATION: KLINEFELTER SYNDROME

Cytogenetic analysis of PHA stimulated cultures revealed a male karyotype with an extra X chromosome in all GTG banded metaphases. This is consistent with a diagnosis of Klinefelter syndrome (tall stature, infertility, gynecomastia).

Genetic counseling is recommended.



Inder K. Gadi, PhD, FACMG
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