



LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 09/01/2023 09:46 Received: 09/01/2023 09:46

Table with 6 columns: Test Name, Result, Flag, Ref-Ranges, Units, Site. Row 1: Y Chromosome Microdeletion, DNA Analysis. Row 2: Referring Physician Phone, na, QCRl. Row 3: Y Chromosome Microdeletion, See Below, QCRl.

RESULT: POSITIVE FOR A DELETION IN THE AZFc REGION OF THE Y CHROMOSOME

Interpretation: This individual is positive for a deletion in the AZFc region of the Y chromosome. The markers deleted within this region are DYS236, DAZ (SY242, SY208, SY254, SY255), and DYS240. The deleted markers span the AZFc region. Complete deletion of the AZFc interval is the most common known genetic cause of male infertility. This abnormal result is significant and supports a diagnosis of male infertility due to Y chromosome abnormalities. Males with an AZFc deletion may be candidates for intracytoplasmic sperm injection (ICSI) or testicular sperm extraction (TESE). Y chromosome deletions can be passed from father to son during ICSI/TESE. Genetic counseling is recommended.

Laboratory results and submitted clinical information monitored by Arlene Buller-Burckle, Ph.D., FACMG, HCLD, CGMB.

DETAILED ASSAY INFORMATION: Approximately 10% - 20% of male infertility is caused by deletions in one or more regions on the long arm of the Y chromosome (Yq11.2). Deletions of the Y chromosome have been observed rarely in fertile men (NEJM 336(8): 534-539, 1997). Greater than 95% of the Y chromosome deletions that have been reported in the literature are detectable by the methodology used in this assay.

METHODOLOGY: Multiplex polymerase chain reaction and agarose gel electrophoresis were used to detect 20 regions on the long arm of the Y chromosome. Lack of amplification of two or more adjacent markers indicates a Y chromosome deletion.

Markers tested: SY14 (SRY), SY81 (DYS271), SY86 (DYS148), SY84 (DYS273), SY182 (KALY), SY121 (DYS212), SYPR3 (SMCY), SY124 (DYS215), SY127 (DYS218), SY128 (DYS219), SY130 (DYS221), SY133 (DYS223), SY134 (DYS224), SY145 (DYF51S1), SY152 (DYS236), SY242 (DAZ), SY208 (DAZ), SY254 (DAZ), SY255 (DAZ), SY157 (DYS240).

LIMITATIONS: This assay is limited to the detection of deletions that affect the markers listed above. We are unable to determine if the absence of a single marker is caused by a deletion or a nucleotide sequence variation in the binding site for one of the PCR

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED



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<u>Test Name</u>	<u>Result</u>	<u>Flag</u>	<u>Ref-Ranges</u>	<u>Units</u>	<u>Site</u>
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primers used to amplify that marker. Although rare, false positive or false negative results may occur. All results should be interpreted in the context of clinical findings, relevant history, and other laboratory data.

Health care providers, please contact your local Quest Diagnostics' genetic counselor or call 1-866-GENEINFO (1-866-436-3463) for assistance with the interpretation of these results.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Reviewed and signed by Laboratory results and submitted clinical information monitored by Arlene Buller-Burckle, Ph.D., FACMG, HCLD, CGMB, Signed on 11/02/2022 at 18:14

Test Performed at:

Quest Diagnostics Nichols Institute  
33608 Ortega Highway

San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

Performing Site:

QCRL: QUEST DIAGNOSTICS REFERENCE LAB CAPISTRANO 33608 Ortega Highway San Juan Capistrano CA 92675

**Reported Date:** 2023.09.01 9:46 YCMIC

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F301000003  
WX0000003827

Ordered By: KAJAL SITWALA, MD, PhD  
WX00000000002365

Kajal V. Sitwala, MD, PhD - Medical Director

Form: MM RL1

Printed D&T: 09/01/23 09:47

PAGE 2 OF 2