

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 9/5/1984
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Y Chromosome Microdeletion

ARUP test code 2001778

Y CHROM Specimen whole Blood

Y Chromosome Result

AZF C *

Indication for testing: Male infertility.

ONE AZFC Y CHROMOSOME MICRODELETION DETECTED: A Y chromosome microdeletion of the azoospermia factor region c(AZFC) was detected. The AZFC microdeletion has a variable expression and may result in azoospermia oligospermia, or abnormal sperm morphology. Assisted reproductive techniques involving testicular sperm extraction and intracytoplasmic sperm injection have been effective for men carrying the AZFC microdeletion. All male offspring will inherit the microdeletion and thus are at high risk for infertility. Female offspring will not inherit the microdeletion. Genetic counseling is recommended.

This result has been reviewed and approved by [REDACTED]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Y Chromosome Microdeletion

CHARACTERISTICS: Y chromosome microdeletions are typically characterized by azoospermia, severe to moderate oligospermia, or abnormal sperm morphology/motility in men with a normal physical evaluation. Assisted reproductive techniques are contraindicated for men carrying AZFa, AZFb, AZFbc or AZFabc microdeletions, which are classically associated with spermatogenic failure.

PREVALENCE: 1 in 2,000 to 3,000 males carry Y chromosome deletions/microdeletions.

PENETRANCE: Approaches 100 percent in males; variable expression may result in intra-familial variation of fertility in men with an identical microdeletion.

INHERITANCE: Y-linked; microdeletions are typically de novo.

CAUSE: Microdeletions of the Y chromosome azoospermia factor regions a, b or c (AZFa, AZFb or AZFc).

MUTATIONS TESTED: Five common Y chromosome microdeletions: AZFa, AZFb, AZFc, AZFbc, and AZFabc.

CLINICAL SENSITIVITY: Estimated at 5 to 10 percent for men with non-obstructive azoospermia or severe oligospermia.

METHODOLOGY: Multiplex polymerase chain reaction (PCR) followed by electrophoresis.

ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Mutations within individual genes included in the AZF regions will not be detected. Breakpoints of identified microdeletions will not be determined. Male infertility due to causes other than Y chromosome microdeletions tested, has not been excluded.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Y CHROM Specimen	23-082-113935	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Y Chromosome Result	23-082-113935	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical