

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

Admitting Provider: Doctor, Example

Patient: Patient, Example

DOB: 12/31/1752
Sex: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

FISH Metaphase, SRY

ARUP test code 3021316
Ordering Provider: Doctor, Example

FISH Metaphase, SRY

See Note

Test Performed: Chromosome FISH, Metaphase (CHR FISHM)
Specimen Type: Peripheral blood
Indication for Testing: Ambiguous genitalia

RESULT

Abnormal FISH Result

XY Chromosome Complement *See Note

X (DXZ1): 1 copy detected
Yp11.3 (SRY): 1 copy detected

INTERPRETATION

This analysis showed a signal pattern consistent with one copy of the X chromosome and the presence of Yp11.3 (SRY) on the Y chromosome.

NOTE: The genotypic sex differs from the information provided for this patient.

Non-syndromic differences of sexual development have multiple etiologies. Referral to endocrinology is recommended to guide medical management.

This assay only analyzes the DNA locus complementary to the FISH probe for enumeration and localization of that sequence. This result does not rule out copy number variants or structural abnormalities smaller than the probe target or those involving other loci, aneuploidy, or low-level mosaicism.

This analysis was performed with the CEP X (DXZ1)/Yp11.3 (SRY) probe (Abbott). A total of 10 metaphase and 100 interphase cells were scored. A normal male control was used to establish the performance of this probe.

Recommendations:

- 1) Genetic counseling
- 2) Endocrine consultation

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

1) Mohnach. Nonsyndromic Disorders of Testicular Development. GeneReviews. 2022. (www.ncbi.nlm.nih.gov/books/NBK1547/). PMID: 20301714.

2) Delot et al. Genetics of Disorders of Sex Development: The DSD-TRN Experience. Endocrinol Metab Clin North Am. 2017

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 26-258-111138
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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Jun;46(2):519-537. PMID: 28476235.

Cytogenomic Nomenclature (ISCN):
ish X(DXZ1x1),Yp11.3(SRYx1).
nuc ish(DXZ1,SRY)x1

This result has been reviewed and approved by [REDACTED]

INTERPRETIVE INFORMATION: FISH Metaphase SRY

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.

EER FISH Metaphase-SRY

See Note
Authorized individuals can access the ARUP Enhanced Report with an ARUP Connect account using the following link.
Your local lab can assist you in obtaining the patient report if you don't have a Connect account.

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
FISH Metaphase, SRY	26-258-111138	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER FISH Metaphase-SRY	26-258-111138	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

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Patient: Patient, Example
ARUP Accession: 26-258-111138
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