Chromosome FISH, Metaphase

ARUP LABORATORIES

Patient: [redacted] DOB: [redacted] Age: 14 Sex: F **Patient Identifiers:** [redacted] **Visit Number (FIN):** [redacted]	Client: [redacted] Physician: [redacted]	ARUP Test Code: 2002299 Collection Date: 02/18/2023 Received in lab: 02/20/2023 Completion Date: 03/01/2023

Interpretation

Test Performed: Chromosome FISH, Metaphase (CHR FISHM)
Specimen Type: Peripheral blood
Indication for Testing: Premature ovarian failure, clitoromegaly, deep voice; Rule out X;Y translocation involving SRY

RESULT
Normal FISH Result

XX Chromosome Complement

X (DXZ1): two copies detected
Yp11.3 (SRY): not detected

INTERPRETATION
There was no evidence of Yp11.3 (SRY). This analysis showed the expected signal patterns for a normal XX chromosome complement.

Structural abnormalities involving other loci, aneuploidy, and mosaicism have not been ruled out by this analysis. If this patient is symptomatic, additional testing by chromosome analysis or genomic microarray analysis and/or a genetics consult is recommended. If additional testing is being performed, results will be reported separately.

This assay only analyzes the DNA locus complimentary to the FISH probe for enumeration and localization of that sequence. This result does not rule out low-level mosaicism or copy number variants outside of, or smaller than, the probe target.

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.

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

Cytogenomic Nomenclature (ISCN):
ish X(DXZ1x2),Yp11.3(SRYx0).
nuc ish(DXZ1x2,SRYx0)

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following location(s):
[redacted]

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

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This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. 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 at www.aruplab.com.

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