

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 8/31/2004  
**Gender:** Female  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Chromosome Analysis, Rule Out Mosaicism**

ARUP test code 2002287

Chromosome Analysis, Rule Out Mosaicism      See Note      (Ref Interval: Normal)

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at [www.aruplab.com](http://www.aruplab.com). Incidental findings are not reported unless clinically significant but are available upon request.

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Test Performed: Chromosome Analysis  
Specimen Type: Peripheral Blood  
Indication for Testing: Secondary amenorrhea, Endocrine disorder, unspecified

Number of cells counted: 50  
Number of cells analyzed: 50  
Number of cells karyotyped: 12  
ISCN band level: 550  
Banding method: G-Banding

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**RESULT**

Abnormal Karyotype

Monosomy X with XXX Chromosome Mosaicism

45,X[39]/47,XXX[11]

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**INTERPRETATION**

This analysis detected two cell lines. One cell line showed a single X chromosome (monosomy) without a second sex chromosome in 39/50 cells (78%). The remaining cells showed three X chromosomes (47,XXX). This result is consistent with a clinical diagnosis of Turner syndrome (monosomy X) with mosaicism.

Complete monosomy X (45,X) is consistent with a clinical diagnosis of Turner syndrome (TS). Features associated with TS may include kidney and cardiac defects, lymphedema, a short and webbed neck, broad chest with widely spaced nipples, short stature, and gonadal dysgenesis. Although fertility is often spared in women with monosomy X mosaicism, premature ovarian failure is common. Intelligence is generally within the normal range, but delayed motor skills/poor coordination as well as difficulties with math and social skills may be present.

Most individuals with 45,X/47,XXX mosaicism present with variable features of Turner syndrome. The presence of the 47,XXX cell line may modify the phenotypic features of the syndrome. At least one study suggests that some patients with Turner syndrome due to 45,X/47,XXX mosaicism may be more mildly affected for phenotypic features and ovarian function.

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

No other abnormalities were detected. The standard cytogenetic methodology used in this analysis may not detect small rearrangements, cannot detect submicroscopic deletions or duplications that are detectable by genomic microarray analysis, and does not rule out mosaicism for another cell line(s).

**Recommendations:**

- 1) Genetic counseling
- 2) Endocrine consultation

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

**References:**

Powell CM. Sex chromosomes, sex chromosome disorders, and disorders of sex development. in Gersen SL and Keagle MB. The Principle of Clinical Cytogenetics (3rd Ed), 2013, Springer.

This result has been reviewed and approved by [REDACTED]

**INTERPRETIVE INFORMATION:** Chromosome Analysis,  
Rule Out Mosaicism

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 Chromosome Analysis, R/O Mosaicism**

See Note

Authorized individuals can access the ARUP Enhanced Report using the following link:

[REDACTED]

**VERIFIED/REPORTED DATES**

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Rule Out Mosaicism	22-173-129791	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis, R/O Mosaicism	22-173-129791	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**

Unless otherwise indicated, testing performed at: