

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 5/9/2007
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)

Specimen received

Specimen type: Peripheral Blood (Mosaic Study)
Reason for referral: Short stature
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 50
Number of cells analyzed: 6
Number of cells karyotyped: 6
ISCN Band level: 550
Banding Method: G-Banding

RESULT
Abnormal Karyotype

Monosomy X with X Chromosome Mosaicism

45,X[4]/46,XX[46]

INTERPRETATION
This analysis detected two cell lines. One cell line showed a single X chromosome (monosomy) without a second sex chromosome in 4/50 cells (8%). The remaining cells showed a normal female karyotype. 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). However, individuals mosaic for monosomy X and a 46,XX cell line can have a phenotype that ranges from normal female to classic TS. Based on prospective studies, the majority of 45,X/46,XX mosaic individuals diagnosed prenatally have a normal phenotype at birth, and if features of TS exist, they tend to be mild. 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.

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,

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

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:

- 1) Pinsker. Clinical review: Turner syndrome: updating the paradigm of clinical care. J Clin Endocrinol Metab. 2012 Jun;97(6):E994-1003. PMID: 22472565.
- 2) Sybert and McCauley. Turners syndrome. N Engl J Med. 2004 Sept 16;351(12):1227-38. PMID: 15371580.
- 3) Gravholt et al. Clinical practice guidelines for the care of girls and women with Turner syndrome: proceedings from the 2016 Cincinnati International Turner Syndrome Meeting. Eur J Endocrinol. 2017 Sept; 177(3): G1-G70. PMID: 28705803.
- 4) Wolff et al. Working Group of the ACMG Laboratory Quality Assurance Committee. Laboratory guideline for Turner syndrome. Genet Med. 2010 Jan;12 (1):52-5. PMID: 20081420.

This result has been reviewed and approved by [REDACTED]

[REDACTED]

INTERPRETIVE INFORMATION: Chromosome Analysis,
Rule Out Mosaicism
Test developed and characteristics determined by ARUP
Laboratories. See Compliance Statement C: aruplab.com/CS

EER Chromosome Analysis, R/O Mosaicism

See Note

Access ARUP Enhanced Report using either link below:

-Direct access: [REDACTED]

-Enter Username, Password: [REDACTED]

Username: [REDACTED]
Password: [REDACTED]

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Rule Out Mosaicism	20-228-402015	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis, R/O Mosaicism	20-228-402015	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:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 20-228-402015
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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