

Patient: [REDACTED]
DOB: [REDACTED] Age: 15 Gender: F
Patient Identifiers: [REDACTED]
Visit Number (FIN): [REDACTED]

Client: [REDACTED]
Physician: [REDACTED]

ARUP Test Code: 2002287
Collection Date: 09/03/2020
Received in lab: 09/04/2020
Completion Date: 09/16/2020

Interpretation

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

Chromosome Results: 46,XX

Diagnostic Impression:

Metaphase cells analyzed revealed a normal female chromosome complement. The standard cytogenetic methodology used in this analysis may not detect small rearrangements or low level mosaicism, and cannot detect submicroscopic deletions or duplications that are detectable by microarray analysis.

A total of 50 metaphase cells were evaluated. This should exclude mosaicism of greater than 9 percent with 99 percent confidence in this tissue (Hook et al., Am J Hum Genet 1977, 29:94-97).

If mosaicism is still of concern, consider testing an alternative tissue sample as levels of mosaicism may vary across different tissues.

NOTE: Further analysis of this patient by genomic microarray may prove informative. Genomic microarray analysis is recommended as a first-tier test for the detection of genomic imbalances causing autism spectrum disorders, intellectual disability / developmental delay, and multiple congenital anomalies. Information regarding this test may be found at <http://www.aruplab.com/guides/ug/tests/2003414.jsp>. For additional information or questions, please contact an ARUP genetic counselor at (800)242-2787 x2141.

This result has been reviewed and approved by [REDACTED],
PhD, FACMG

A portion of this analysis was performed at the following



Patient: [REDACTED]
ARUP Accession: 20-247-124136
[REDACTED]

Chromosome Analysis, Rule Out Mosaicism

Patient: [REDACTED] | Date of Birth: [REDACTED] | Gender: F | Physician: [REDACTED]
Client: [REDACTED] | Client Address: [REDACTED]

Location(s):

[REDACTED]
[REDACTED]
[REDACTED]

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS



Chromosome Analysis, Rule Out Mosaicism

Patient: [REDACTED] | Date of Birth: [REDACTED] | Gender: F | Physician: [REDACTED]
Client: [REDACTED] | Client Address: [REDACTED]

Slide ID: 0066

