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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Constitutional Blood, with Reflex to Genomic Microarray

ARUP test code 2005763

Chromosome Analysis Constitutional Blood

See Note (Ref Interval: Normal)

Test Performed: Chromosome Analysis
Specimen Type: Peripheral Blood
Indication for Testing: Evaluate for trisomy X

Number of cells counted: 20
Number of cells analyzed: 5
Number of cells karyotyped: 5
ISCN band level: 550
Banding method: G-Banding

RESULT
Abnormal Karyotype (Female)

Trisomy X (XXX or Triple X syndrome)

47,XXX

INTERPRETATION
This analysis showed an additional X chromosome in each metaphase.

This result is consistent with a clinical diagnosis of triple X syndrome (trisomy X or 47,XXX). The 47,XXX karyotype is most often observed in physically normal females with tall stature. Pubertal development and fertility are typically normal. Cognition is typically within normal range; however, there is a susceptibility to delays in speech and neuromotor skills, as well as learning disabilities and behavioral difficulties, for which interventions and support may be helpful. Other features, typically minor, have been reported in some individuals with triple X syndrome (see references below). Considerable variability in the expression of these traits has been observed; therefore, a precise prognosis is not possible.

Historically, most individuals with triple X syndrome went undiagnosed; however, this is changing due to the increased availability of prenatal screening and testing. Individuals who are diagnosed prenatally tend to have milder features than those who are diagnosed postnatally, but this is likely due to ascertainment bias.

No other abnormalities were detected. 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 genomic microarray analysis.

Since this analysis revealed an abnormal result, genomic microarray analysis, which was ordered as a reflex study, will

not be performed unless we are notified otherwise. To proceed with this testing, please contact ARUP Genetics Processing at (800) 242-2787 ext. 3301.

Recommendation:
Genetic counseling

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

References:
1) Otter et al. Triple X syndrome: a review of the literature. Eur J Hum Genet. 2010 Mar;18(3):265-71. PMID: 19568271.
2) Tartaglia et al. A review of trisomy X (47,XXX). Orphanet J Rare Dis. 2010 May; 11(5):8. PMID: 20459843.
3) Wigby et al. Expanding the phenotype of triple X syndrome: a comparison of prenatal versus postnatal diagnosis. Am J Med Genet A. 2016 Nov;170(11):2870-2881. PMID: 27644018.
4) Unique: Understanding Rare Chromosome and Gene Disorders. (www.rarechromo.org)

This result has been reviewed and approved by [redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis
Constitutional Blood
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 Chrom Analysis PB w/Rflx to Array

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 Constitutional Blood	23-159-122939	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chrom Analysis PB w/Rflx to Array	23-159-122939	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