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

Physician: Doctor, Example

Patient: Patient, Example

DOB	11/12/1990
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Amniotic Fluid

ARUP test code 2002293

Chromosome Analysis, Amniotic Fluid	See Note	(Ref Interval: Normal)

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

Test performed: Chromosome Analysis
Specimen type: Amniotic Fluid
Reason for referral: Abnormal NIPT XXY

Laboratory analysis

Number of cells counted: 15
Number of colonies counted: 15
Number of cells analyzed: 15
Number of cells karyotyped: 15
ISCN Band level: 400
Banding Method: G-Banding

RESULT
Abnormal Karyotype (Male)

Klinefelter syndrome

47,XXY

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

This result is consistent with a clinical diagnosis of Klinefelter syndrome (47,XXY). Features associated with Klinefelter syndrome may include delayed or incomplete puberty, infertility, hypogenitalism, gynecomastia, and taller than average stature. Characteristic features show wide variability and may also include decreased muscle tone during infancy, delayed speech and language skills, and difficulty in reading. Affected boys also tend to be shy and social integration may be difficult. Reduced testosterone levels (hypogonadism) may warrant hormonal supplementation. Adults with Klinefelter syndrome may have an increased risk for developing breast cancer.

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.

Recommendations:
Genetic counseling

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

References:
1) Aksglaede et al. 47,XXY Klinefelter syndrome: clinical characteristics and age-specific recommendations for medical management. Am J Med Genet C Semin Med Genet. 2013 Feb 15; 163C(1):55-63. PMID: 23345262.
2) Groth et al. Clinical review: Klinefelter syndrome--a clinical update. J Clin Endocrinol Metab. 2013 Jan;98(1):20-30. PMID: 23118429.

This result has been reviewed and approved by [redacted]

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

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

INTERPRETIVE INFORMATION: Chromosome Analysis, Amniotic Fluid

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 Amniotic Fluid

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, Amniotic Fluid	23-039-107323	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis Amniotic Fluid	23-039-107323	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