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

Physician: Doctor, Example

Patient: Patient, Example

DOB	11/27/1976
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray

ARUP test code 2008367

Chromosome Analysis, Amniotic Fluid

See Note (Ref Interval: Normal)

Specimen received

Specimen type: Amniotic Fluid
Reason for referral: AMA, echogenic intracardiac focus, positive NIPT for trisomy 21
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 20
Number of colonies counted: 9
Number of cells analyzed: 20
Number of cells karyotyped: 20
ISCN Band level: 400
Banding Method: G-Banding

RESULT
Abnormal Karyotype (Male)

Trisomy 21 (Down syndrome)

47,XY,+21

INTERPRETATION
This analysis showed an additional copy (trisomy) of chromosome 21 in each metaphase.

This result is consistent with a clinical diagnosis of trisomy 21 (Down syndrome). Features associated with Down syndrome may include hypotonia, a characteristic facial appearance, developmental delays/intellectual disability, and short stature. Other findings may include congenital heart defects, hypothyroidism, single transverse palmar crease, hearing and vision difficulties, digestive abnormalities, as well as an increased risk for developing leukemia and Alzheimer's disease later in life.

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.

Recommendation:
Genetic counseling

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

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

References:
1) Sheets et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. J Genet Couns. 2011 Oct;20(5):432-41. PMID: 21618060.
2) Bull. Committee on Genetics. Health supervision for children with Down syndrome. Pediatrics. 2011 Aug;128(2):393-406. PMID: 21788214.
3) Jones et al. Smith's Recognizable Patterns of Human Malformations. 7th edition. Philadelphia, PA: Elsevier Saunders; 2013:7-13.

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.

This result has been reviewed and approved by [redacted]

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

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

EER Chrom Analysis AF w/Rflx to Array

See Note

Access ARUP Enhanced Report using either link below:

-Direct access: [redacted]

-Enter Username, Password: [redacted]
Username: [redacted]
Password: [redacted]

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Amniotic Fluid	20-261-155726	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chrom Analysis AF w/Rflx to Array	20-261-155726	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