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

Physician: Doctor, Example

Patient: Patient, Example

DOB	1/4/1985
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

Specimen received

Specimen type: Amniotic Fluid
Reason for referral: AMA, abnormal NIPT (trisomy 21)
Test performed: Chromosome Analysis

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)

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.

NOTE: FISHP was performed on this sample and reported under ARUP accession #(21-034-121231). FISH results were ABNORMAL.

Recommendation:
Genetic counseling

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.

This result has been reviewed and approved by [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

Access ARUP Enhanced Report using the link below:

-Direct access: [redacted]

Chromosome FISH, Amniotic Fluid with Reflex to Chromosome Analysis or Genomic Microarray

ARUP test code 2011130

Chromosome FISH, Prenatal

See Note (Ref Interval: Normal)

Specimen Received
Specimen Type: Amniotic Fluid
Reason for Referral: Advanced Maternal Age; Abnormal NIPT-T21
Test Performed: FISH, Prenatal

ABNORMAL FISH RESULT: Trisomy 21

DIAGNOSTIC IMPRESSION:
Prenatal interphase fluorescence in situ hybridization (FISH) analysis was performed with chromosome enumeration probes for 13, 18, 21, X and Y using the FDA-approved AneuVysion probe kit (Abbott Molecular). 50 interphase cells were scored for each probe.

This analysis showed three hybridization signals for chromosome 21, consistent with trisomy 21.

Sex chromosomes: XY (male)

This specimen is being reflexed to chromosome analysis.

ISCN:
nuc ish(DXZ1x1,DYZ3x1,D18Z1x2),
(RB1x2,D21S259/D21S341/D21S342x3)

Recommendations:
The American College of Medical Genetics recommends that (1) clinical decision-making should be based on information from two of three of the following: positive FISH results, confirmatory chromosome analysis, or consistent clinical information, (2) for genetic counseling of families that have a fetus identified as positive by FISH, chromosome analysis to determine the mutational mechanism accounting for the FISH result is performed, and (3) genetic counseling for all abnormal results.

This result has been reviewed and approved by [redacted]

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

INTERPRETIVE INFORMATION: Chromosome Analysis,
Prenatal FISH

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Amniotic Fluid	21-034-121231	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis Amniotic Fluid	21-034-121231	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Chromosome FISH, Prenatal	21-034-121231	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