

Patient: [REDACTED]
DOB: [REDACTED] Age: 33 Gender: F
Patient Identifiers: [REDACTED]
Visit Number (FIN): [REDACTED]

Client: [REDACTED]
Physician: [REDACTED]

ARUP Test Code: 2002297
Collection Date: 03/06/2018
Received in lab: 03/07/2018
Completion Date: 03/08/2018

Interpretation

Specimen Received
Specimen Type: Amniotic fluid
Reason for Referral: NIPT positive for trisomy 21
Test Performed: FISHP

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: XX (female)

ISCN:
nuc ish(DXZ1x2,DYZ3x0,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 Roger Schultz,
Ph.D., FACMG
Electronic Signature

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



Patient: [REDACTED]
ARUP Accession: 18-065-117648