

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

Client: [REDACTED]
Physician: [REDACTED]

ARUP Test Code: 0040208
Collection Date: 11/21/2016
Received in lab: 11/23/2016
Completion Date: 11/24/2016

Interpretation

Specimen Received
Specimen Type: Peripheral Blood
Reason for Referral: Characteristics of Down syndrome
Test Performed: FISHANEU

ABNORMAL FISH RESULTS: Trisomy 21

DIAGNOSTIC IMPRESSION:

Interphase fluorescence in situ hybridization (FISH) analysis was performed with chromosome enumeration probes for 13, 18, 21, X and Y using the AneuVysion probe kit (Abbott Molecular). 200 interphase cells were scored for each probe.

This analysis showed three hybridization signals for chromosome 21, consistent with the clinical diagnosis of Down syndrome (trisomy 21).

Sex chromosomes: XX (female)

ISCN:
nuc ish(DXZ1x2,DYZ3x0,D18Z1x2),
(RB1x2,D21S259/D21S341/D21S342x3)

Recommendations:

1. Genetic counseling
2. For recurrence risk identification, chromosome analysis is recommended to determine the mutational mechanism accounting for this FISH result.

This result has been reviewed and approved by [REDACTED],
Ph.D., FACMG
Electronic Signature

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



Patient: [REDACTED]
ARUP Accession: 16-326-140118