

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 9/27/1976
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Chromosome Analysis, Chorionic Villus

ARUP test code 2002291

Chromosome Analysis, Chorionic Villus

See Note (Ref Interval: Normal)

Specimen received

Specimen type: Chorionic villi
Reason for referral: Advanced Maternal Age, Abnormal Ultrasound finding, thickened nuchal fold
Test performed: Chromosome Analysis

Laboratory analysis

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

Chromosome results: 47,XY,+21

Diagnostic Impression:
Metaphase cells analyzed revealed a male chromosome complement with an additional chromosome 21 seen in each metaphase cell, consistent with the clinical diagnosis of Down syndrome (trisomy 21).

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 microarray analysis.

Cytogenetic analysis performed on CVS presumes that the fetal chromosome complement is accurately reflected in the extra-embryonic tissue. There are rare examples in which the karyotype of the CVS is not consistent with that of the fetus.

NOTE: FISH was performed on this sample and reported under patient ID#18139400355. FISH results were ABNORMAL and consistent with this analysis.

Recommendation:
Genetic counseling.

This result has been reviewed and approved by Bo Hong, MD, FACMG
Electronic Signature

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

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

EER Chromosome Analysis Chorionic Villus

See Note
Access ARUP Enhanced Report using either link below:



Chromosome FISH, Chorionic Villus with Reflex to Chromosome Analysis or Genomic Microarray

ARUP test code 2011131

Chorionic Villus, FISH

See Note (Ref Interval: Normal)

Specimen Received
Specimen Type: CVS
Reason for Referral: AMA; abnormal USG: thickened nuchal fold
Test Performed: CVS F RFLX

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 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)

There was no evidence of a numerical abnormality for chromosomes 13, 18, X, and Y.

FISH analysis performed on CVS presumes that the fetal chromosome complement is accurately reflected in the extra-embryonic tissue. There are rare examples in which the karyotype of the CVS is not consistent with that of the fetus.

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

Recommendations:
The American College of Medical Genetics recommends (1) that 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) chromosome analysis to determine the mutational mechanism accounting for the abnormal FISH result, and (3) genetic counseling.

This result has been reviewed and approved by Reha Toydemir, MD, PhD, FACMG
Electronic Signature

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

INTERPRETIVE INFORMATION: Flourescence in Situ
Hybridization, CVS
Test developed and characteristics determined by ARUP
Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Chorionic Villus	18-139-400355	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis Chorionic Villus	18-139-400355	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Chorionic Villus, FISH	18-139-400355	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