

Test Performed: Chromosome FISH, Prenatal (CHR FISHP)
Specimen Type: Direct (uncultured) amniocytes
Indication for Testing: Abnormal NIPT T21

RESULT
Abnormal FISH Result (Female)

Trisomy 21

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

Aneuploidy of other chromosomes, structural abnormalities, and mosaicism have not been ruled out by this analysis. According to ACMG guidelines, clinical decision-making should not be based on the result of this test alone. Additional testing is recommended for the final interpretation of this result; pending results will be reported separately.

NOTE: Interphase FISH analysis cannot provide structural information accounting for this gain. It is uncertain whether this finding represents three independent copies of chromosome 21 or an unbalanced Robertsonian translocation. Therefore, chromosome analysis is recommended.

This analysis was performed with chromosome enumeration probes for 13, 18, 21, X and Y using the FDA-approved AneuVysion probe kit (Abbott Molecular). A total of 50 interphase cells were scored for each probe.

Recommendations:
1) Genetic counseling
2) Chromosome analysis (if feasible and not already performed). This test is available, at a charge, through ARUP Laboratories. Please order test code 2002293, Chromosome Analysis, Amniotic Fluid. If adding testing to this prenatal sample, testing must be added within 7 days to ensure sample availability. Alternatively, chromosome analysis may be performed after delivery on peripheral blood (2002289, Chromosome Analysis, Constitutional Peripheral Blood) or on a products of conception specimen (2002288, Chromosome Analysis, Products of Conception).

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

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

This result has been reviewed and approved by [REDACTED]

INTERPRETIVE INFORMATION: Chromosome Analysis,
Prenatal FISH

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 FISH, Prenatal

See Note

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

Authorized individuals can access the ARUP
Enhanced Report using the following link:

[REDACTED LINK]

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome FISH, Prenatal	23-052-132351	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome FISH, Prenatal	23-052-132351	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

Unless otherwise indicated, testing performed at: