Chorionic Villus, FISH

Patient:	Client:	ARUP Test Code: 0040203
DOB: Age: Gender:		Collection Date: 12/06/2016
Patient Identifiers:	Physician:	Received in lab: 12/07/2016
Visit Number (FIN):		Completion Date: 12/09/2016

Interpretation

Specimen Received
Specimen Type: Chorionic Villi
Reason for Referral: Positive ctDNA Test Result for Trisomy 18, Previous Child Born w/ Trisomy 18 in 1999 (No Records)
Test Performed: FISHCVS

ABNORMAL FISH RESULT: Trisomy 18

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 18, consistent with trisomy 18.

Sex chromosomes: XX (female)

There was no evidence of a numerical abnormality for chromosomes 13, 21, 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(DXZ1x2,DYZ3x0,D18Z1x3),
(RB1x2,D21S259/D21S341/D21S342x2)

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 , MD, FACMG
Electronic Signature

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

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