Chromosome FISH, Prenatal

Patient: [redacted] DOB: [redacted] Age: 39 Gender: F **Patient Identifiers:** [redacted] **Visit Number (FIN):** [redacted]	Client: [redacted] Physician: [redacted]	ARUP Test Code: 2002297 Collection Date: 01/30/2019 Received in lab: 01/31/2019 Completion Date: 02/02/2019

Interpretation

Specimen Received
Specimen Type: Amniotic Fluid
Reason for Referral: Cystic Hygroma
Test Performed: FISHP

ABNORMAL FISH RESULT: Monosomy X (Turner Syndrome)

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 one hybridization signal for chromosome X and no other hybridization signals for sex chromosomes. Monosomy X is associated with abnormal ultrasound findings in the prenatal period that may include nuchal cystic hygroma and fetal hydrops, and postnatally with the clinical diagnosis of Turner syndrome.

Sex chromosome: X

ISCN:
nuc ish (DXZ1x1, DYZ3x0, D18Z1x2),
(RB1x2,D21S259/D21S341/D21S342x2)

Recommendations:
The American College of Medical Genetics recommends that (1) clinical decision-making should be based on information from two or 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 Xinjie Xu, PhD, FACMG

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

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