

Example Client, XYZ123 1234 Warde Road Ann Arbor MI 48108 **EXAMPLE, REPORT W**

WX0000003826 F 12/05/1988 34 Y

Referral Testing

Collected: 09/01/2023 08:56 Received: 09/01/2023 08:56

<u>Test Name</u> <u>Result</u> <u>Flag Ref-Ranges</u> <u>Units</u> <u>Site</u>

Cytogenetics

Chromosome Analysis, Amniotic Fluid SEE BELOW QARL

SPECIMEN TYPE: Amniotic Fluid

CLINICAL HISTORY: Monosomy X on NIPT, Cystic Hygroma, Hydrops, Short

Femurs and Humeri

CYTOGENETICS LAB NUMBER: PREVIOUS CASE: None Known

TESTS ORDERED

Chromosome Analysis, AFP, Prenatal Ploidy Assessment

INTERPRETATION

OVERALL INTERPRETATION Chromosome Analysis

Abnormal Karyotype - Monosomy X Karyotype

All metaphases examined contain 45 chromosomes with a single X chromosome. No other consistent numerical or structural chromosome abnormalities were observed.

This is predictive of a clinical diagnosis of Turner syndrome. Genetic counseling is recommended.

COMMENT

Chromosome analysis will not detect subtle translocations, deletions, inversions or other cytogenetic abnormalities that are beyond the resolution limits of the technology used.

SIGNATURE

RESULTS

ISCN RESULTS

45,X

CULTURES

CULTURES: 4

COLONIES

COLONIES: 15

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, $\,$. - NOT TESTED

F301000001 WX0000003826 Printed D&T: 09/28/23 11:14 Ordered By: KAJAL SITWALA, MD, PhD WX00000000002353

Kajal V. Sitwala, MD, PhD - Medical Director Form: MM RL1 PAGE 1 OF 5



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CELLS EXAMINED: 15

BANDING/STAINING TECHNIQUE(S): G-BANDING

CELLS ANALYZED: 15

BAND RESOLUTION: 450

CELLS KARYOTYPED: 2

KARYOTYPES

45,X

CYTOGENETICS LAB NUMBER:

CPT CODES

88274x2, 88280, 88271x5, 88235, 82106, 88267

The CPT codes provided are for information purposes only, and are based on AMA guidelines without regard to specific payor requirements.
END OF REPORT

FINAL REPORT-ACC FINAL

AmeriPath Northeast, 1 Greenwich Place, Shelton, CT 06484. P(866) 436-9632.

F(203)929-2344.

Medical Director: Kamraan Z. Gill, M.D.

CLIA 07D1035411, CT CL-0645

CLINICAL INFORMATION

SPECIMEN SUBMITTED: Amniotic Fluid

INDICATION: Monosomy X on NIPT, Cystic Hygroma, Hydrops, Short Femurs

and Humeri

CYTOGENETICS LAB NUMBER:

IMAGES

Microscopic image is a symbolic representation of the key findings of your specific report. The image is not intended to replace a complete review and reading of the final diagnostic report.

RESULTS

MODAL SIGNAL

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MODAL SIGNAL # PER NUCLEUS:

Normal Chromosome 13

Normal Chromosome 18

2

Normal Chromosome 21

2

Abnormal X Chromosome

1

Abnormal Y Chromosome

0

ISCN

nuc ish

Xcen(DXZ1x1), Ycen(DYZ3x0), 13q14.2(RB1x2), 18q21.3(MALT1x2), 21q22.2(DSCR4x

INTERPRETATION

INTERPRETATION

Abnormal Result.

Fluorescence in situ hybridization (FISH) was performed on uncultured amniocytes using MetaSystems probes specific for chromosomes 13, 18, 21, X, and Y. The pattern observed is consistent with a monosomy X fetus with two copies each of chromosomes 13, 18, and 21.

These results should be considered to be an adjunct to complete chromosome analysis, the results of which are pending. This FISH analysis will not detect structural chromosome abnormalities, mosaicism, or numerical abnormalities involving other chromosomes.

The American College of Medical Genetics and Genomics recommends that irreversible therapeutic action should not be taken based on the results of FISH analysis alone (Am. J. Hum. Genet. 53:526-527, 1993).

SIGNATURE

Electronic Signature:

COMMENT

This test was developed and its analytical performance characteristics have been determined by AmeriPath Northeast. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

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END OF REPORT

FINAL REPORT-ACC FINAL

AmeriPath Northeast,1 Greenwich Place, Shelton, CT 06484. P(866) 436-9632.

F(203) 929-2344. Medical Director: Kamraan Z. Gill, M.D. CLIA
07D1035411, CT CL-0645

Specimen: Amniotic Fluid

Alpha-Fetoprotein: 9.6 mcg/mL

Multiple of the Median: 1.36 (Reference Range less than/=1.99)

This amniotic fluid AFP result is ***** NORMAL***** based on the weeks of gestation provided. A small percentage (less than 3%) of open neural tube defects have a MoM less than or equal to 2.0. 5-10 % of all neural tube defects are closed; most of these will also have a MoM less than or equal to 2.0.

If you have questions concerning this report, please feel free to call our Prenatal Screening Program Center.

Weeks of Gestation: 20 3/7 weeks

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute, San Juan Capistrano. It has not been cleared or approved by the FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Acetylcholinesterase: Test Not Performed. Reflex testing not required since established criteria was not met.

Fetal Hemoglobin: Test Not Performed. Reflex testing not required since established criteria was not met.

Technical Services Performed At: Quest Diagnostics Nichols Institute - SJC, 33608 Ortega Highway, San Juan Capistrano, CA, 92675. P(866)894-6920. F(833)434-1031.

Reading Location: Quest Diagnostics Nichols Institute - SJC, 33608

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F(203)929-2344. Medical Director: Kamraan Z. Gill, M.D. CLIA

07D1035411, CT CL-0645

Reported Date: 2023.09.28 11:14 CHRAF

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