Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray

ARUP Accession number: 16-119-400123
Patient:
Date of birth:
Age:
Gender:

Physician:
Client:

ARUP Test Code: 2008367

Collection Date: 04/27/2016
Received in lab: 04/28/2016
Completion Date: 05/05/2016

Interpretation

Specimen received

Specimen type: Amniotic Fluid
Reason for referral: Abnormal ultrasound: Cardiac defect - VSD
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 15
Number of colonies counted: 15
Number of cells analyzed: 15
Number of cells karyotyped: 15
ISCN Band level: 450
Banding Method: G-Banding

Chromosome results: 47,XX,+18

Diagnostic Impression:
Metaphase cells analyzed showed a female chromosome complement with an additional chromosome 18 seen in each metaphase cell, consistent with the clinical diagnosis of trisomy 18 syndrome (Edwards syndrome).

Features associated with trisomy 18 may include: intellectual disability and major congenital abnormalities, including cardiac defects, feeble fetal activity, growth retardation, single umbilical artery, abdominal wall defects, renal abnormalities, and clenched hands. In liveborns, mortality during the first year of life is greater than 90.

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.

Since this analysis revealed an abnormal result, the cytogenomic microarray analysis, which was ordered as a reflex study, will not be performed unless we are notified otherwise. Please contact ARUP Client Services at 1-800-242-2787 if you want to pursue the microarray analysis.

Recommendation:
Genetic counseling.

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References:
1. Cereda A, Carey JC. 2012. The trisomy 18 syndrome. Orphanet J Rare Dis 7:81. http://www.ojrd.com/content/pdf/1750-1172-7-81.pdf
2. Jones KL, Jones MC, Del Campo M. 2013. Smith's Recognizable patterns of Human Malformations. 7th ed. Philadelphia, PA. Elsevier Saunders, pp 14-19.

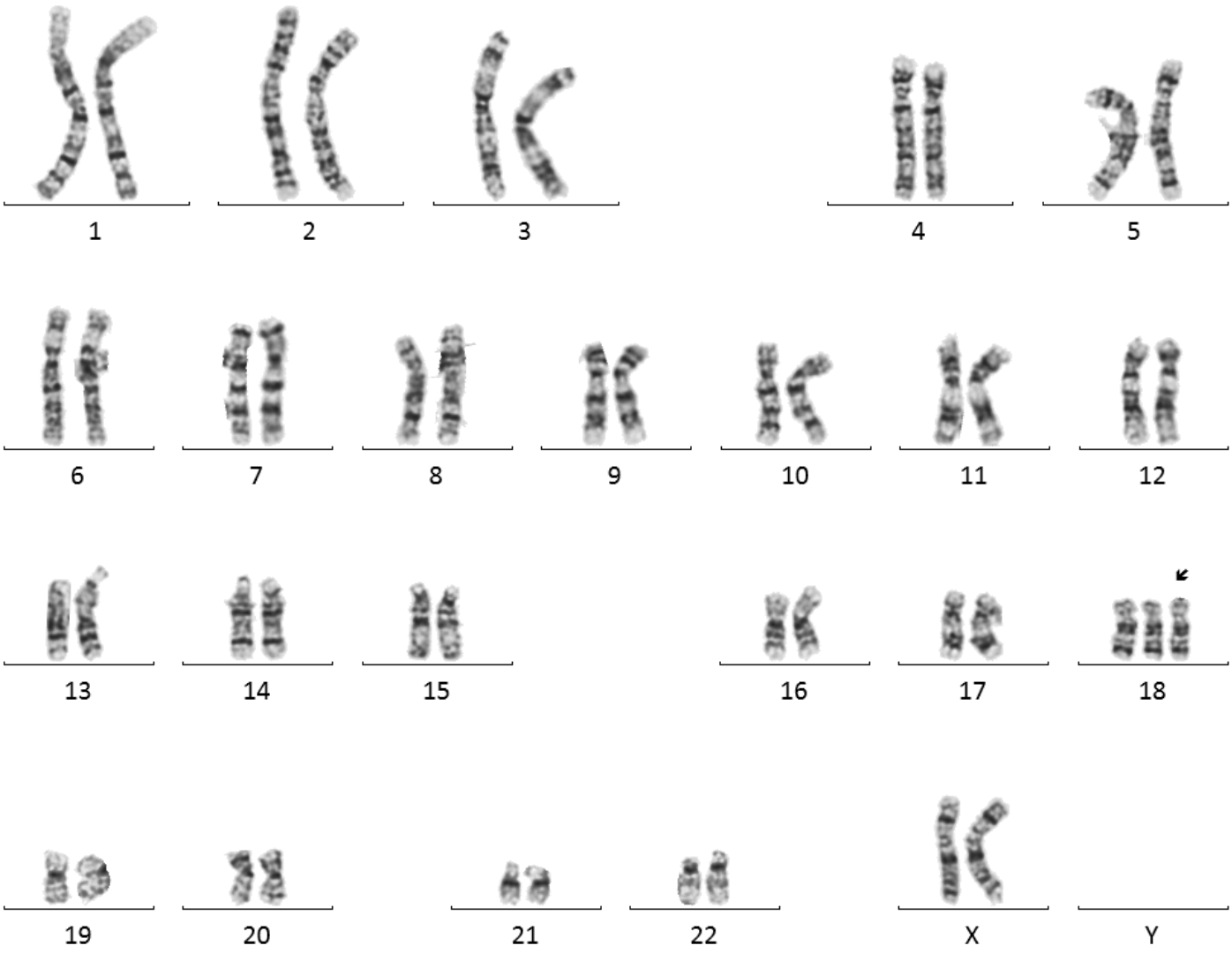
This result has been reviewed and approved by ,
Ph.D., FACMG
Electronic Signature

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

Chart continues on following page(s)

Patient: | **Date of Birth:** | **Gender:** | **Physician:**
Client: | **Client Address:**

Slide ID:



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