

Patient: [REDACTED]
DOB: [REDACTED] Age: [REDACTED] Gender: [REDACTED]
Patient Identifiers: [REDACTED]
Visit Number (FIN): [REDACTED]

Client: [REDACTED]
Physician: [REDACTED]

ARUP Test Code: 2002293
Collection Date: 11/30/2016
Received in lab: 12/06/2016
Completion Date: 12/13/2016

Interpretation

Specimen received

Specimen type: Amniotic Fluid
Reason for referral: Abnormal Ultrasound: Truncus Arteriosus
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 20
Number of colonies counted: N/A
Number of cells analyzed: 20
Number of cells karyotyped: 20
ISCN Band level: 400
Banding Method: G-Banding

Chromosome results: 45,XY,der(9)t(9;22)(q34;q11.2),-22

Diagnostic Impression:

Evaluation of cultured amniocytes revealed a male chromosome complement with a derivative chromosome 9 generated by an unbalanced translocation between the long arms of chromosomes 9 and 22, and monosomy 22. These findings result in loss of genetic material from the terminal end of the long arm of chromosome 9 and from 22q11.2 to the centromere of chromosome 22.

Please see the report of ARRAY FE (ARUP accession #16335117680) for the phenotypic consequence associated with these cytogenetic aberrations.

Recommendations:

1. Genetic counseling
2. Parental chromosome analysis may be considered to determine a potential origin of this rearrangement.

This result has been reviewed and approved by [REDACTED], MD, FACMG
Electronic Signature

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



Patient: [REDACTED]
ARUP Accession: 16-335-117680

Chromosome Analysis, Amniotic Fluid

Patient: [redacted] | Date of Birth: [redacted] | Gender: [redacted] | Physician: [redacted]
Patient Identifiers: [redacted] | Visit Number (FIN): [redacted]

Slide ID: [redacted]

