

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

Client: [REDACTED]
Physician: [REDACTED]

ARUP Test Code: 2002287
Collection Date: 12/01/2016
Received in lab: 12/03/2016
Completion Date: 12/07/2016

Interpretation

Specimen received

Specimen type: Peripheral Blood (Mosaic Study)
Reason for referral: Prenatal Diagnosis of Mosaic Trisomy
21 by Amniocentesis; PDA, DiGeorge/VCF Syndrome
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 50
Number of cells analyzed: 6
Number of cells karyotyped: 6
ISCN Band level: 550
Banding Method: G-Banding

FINAL REPORT

Chromosome results: 47,XX,+21[6]/46,XX[44]

Diagnostic Impression:
Two cell lines were detected in multiple cultures from this patient. One cell line showed an extra copy of chromosome 21 in 6/50 (12%) cells. The remaining 44/50 (88%) cells showed a normal female chromosome complement. These findings are consistent with the diagnosis of trisomy 21 mosaicism.

Clinical findings of patients with trisomy 21 mosaicism range from normal or nearly normal to full expression of Down syndrome features, including developmental delays and intellectual disability, congenital heart defects, short stature, and characteristic facial features. There is no clear correlation between the percentage of trisomy 21 cells and the severity of clinical presentation.

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

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

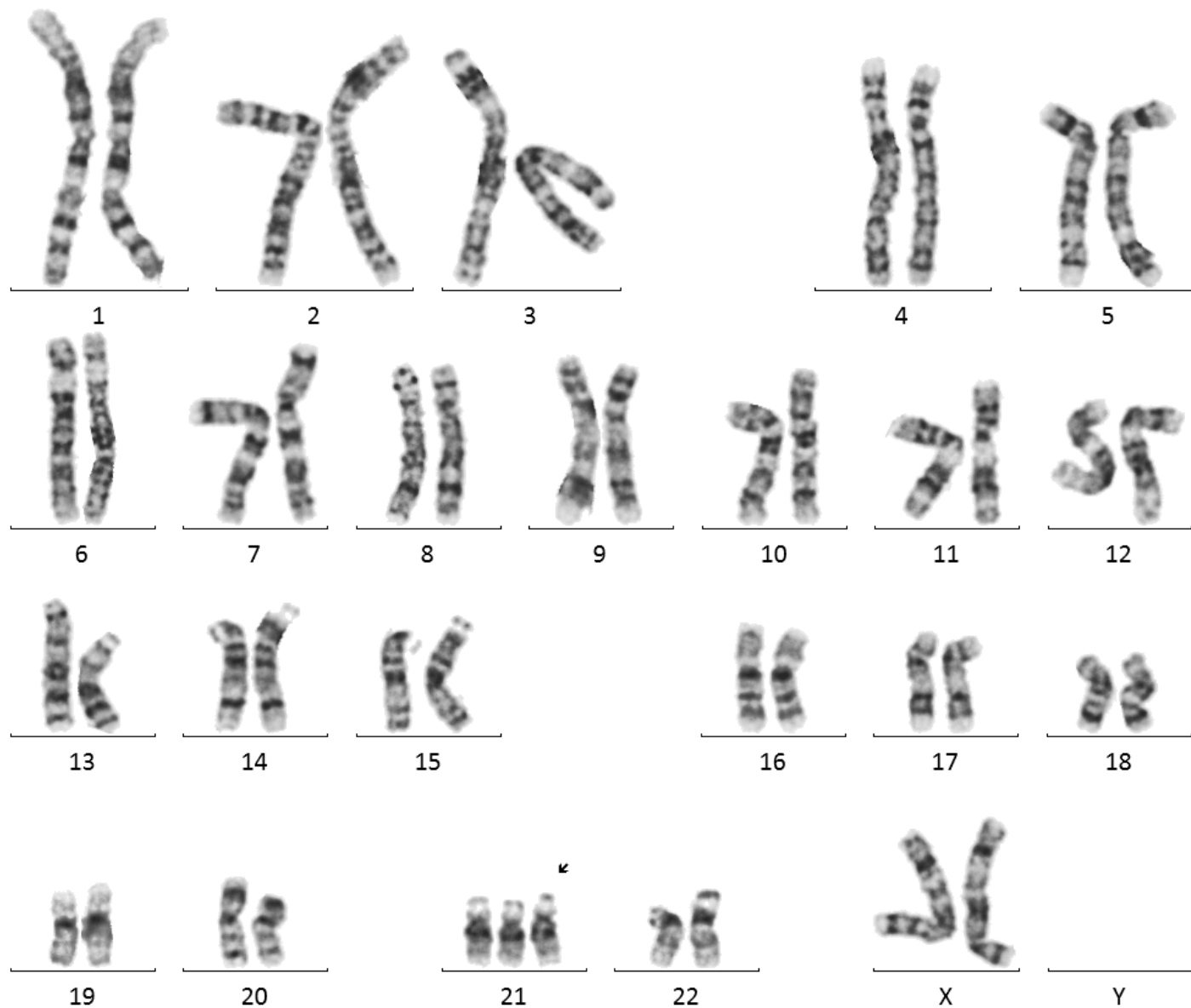


Patient: [REDACTED]
ARUP Accession: 16-336-135810

Chromosome Analysis, Rule Out Mosaicism

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

Slide ID: _____



Chromosome Analysis, Rule Out Mosaicism

Patient: [REDACTED] | Date of Birth: [REDACTED] | Gender: [REDACTED] | Physician: [REDACTED]
Client: [REDACTED] | Client Address: [REDACTED]

Slide ID: 0086

