

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

Client: [REDACTED]  
Physician: [REDACTED]

ARUP Test Code: 2005763  
Collection Date: 12/02/2016  
Received in lab: 12/03/2016  
Completion Date: 12/07/2016

## Interpretation

Specimen received

Specimen type: Peripheral Blood  
Reason for referral: Trisomy 13 with Cleft Lip and Palate,  
Microcephaly  
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 20  
Number of cells analyzed: 5  
Number of cells karyotyped: 5  
ISCN Band level: 550  
Banding Method: G-Banding

## FINAL REPORT

Chromosome results: 47,XY,+13

### Diagnostic Impression:

Metaphase cells analyzed revealed a male chromosome complement with an additional copy of chromosome 13 seen in each metaphase.

This result is consistent with the diagnosis of trisomy 13 syndrome.

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.

Recommendation:  
Genetic counseling

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.

This result has been reviewed and approved by [REDACTED],



Patient: [REDACTED]  
ARUP Accession: 16-337-109131

# Chromosome Analysis, Peripheral Blood, with Reflex to Genomic Microarray

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

Ph.D., FACMG  
Electronic Signature

Notification of preliminary report was given to Mireille L. on  
12/05/2016 15:00 by 19231.

NOTE: Only the **Chromosome Analysis, Peripheral Blood** results are reported on this enhanced report. When the result of Chromosome Analysis is "normal," **Cytogenomic SNP Microarray** is performed. If the genomic microarray is performed, those results can be accessed via a patient report or electronic medical records system after the genomic microarray has been completed.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: [aruplab.com/CS](http://aruplab.com/CS)

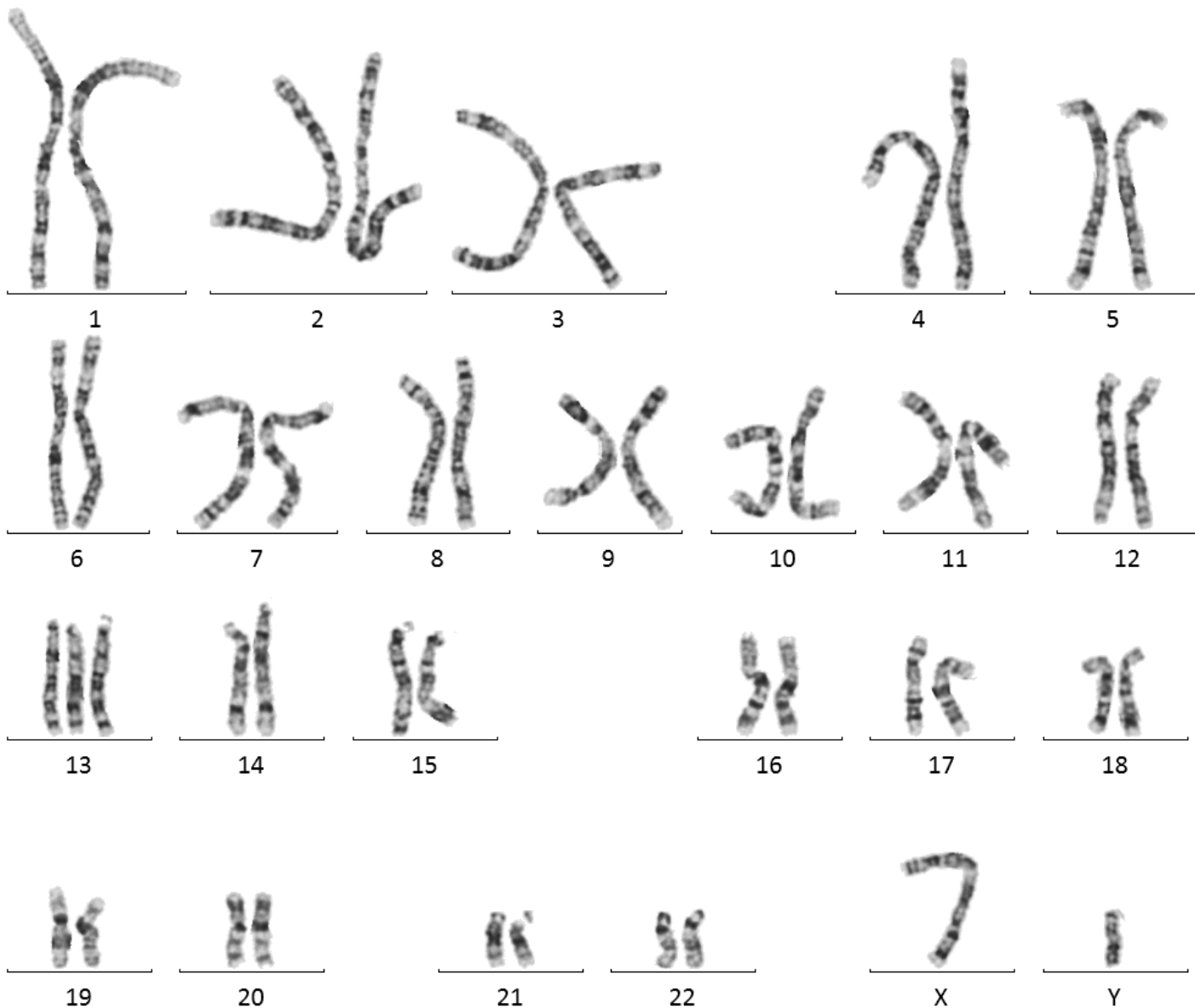


Patient: [REDACTED]  
ARUP Accession: 16-337-109131

# Chromosome Analysis, Peripheral Blood, with Reflex to Genomic Microarray

Patient: \_\_\_\_\_ | Date of Birth: \_\_\_\_\_ | Gender: \_\_\_\_\_ | Physician: \_\_\_\_\_  
Patient Identifiers: \_\_\_\_\_ | Visit Number (FIN): \_\_\_\_\_

Slide ID: \_\_\_\_\_



Patient: \_\_\_\_\_  
ARUP Accession: 16-337-109131