Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Products of Conception, with Reflex to Genomic Microarray

ARUP test code 2005762

Chromosome Analysis, Prod Concp

Normal (Ref Interval: Normal)

POC REFLEXChromosome Analysis, Products of Conception, with Reflex to Genomic Microarray Normal

Test Performed: Chromosome Analysis
Specimen Type: Placental Tissue (Villi)
Indication for Testing: Miscarriage

Number of cells counted: 20
Number of cells analyzed: 11
Number of cells karyotyped: 11
ISCN band level: 400
Banding method: G-Banding

RESULT
Normal Karyotype (Female)

46,XX

This specimen is being reflexed to genomic microarray.

INTERPRETATION
This analysis showed a normal result.

Analysis was performed on chorionic villus dissected from the specimen submitted. These results most likely reflect the fetal rather than the maternal karyotype, however due to the nature of the tissue submitted, the possibility of maternal cell contamination cannot be excluded.

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 genomic microarray analysis.

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

This result has been reviewed and approved by [redacted]

Test Performed: Genomic SNP Microarray, Products of Conception (ARRAY POC)
Specimen Type: Placental Tissue (Villi)
Indication for Testing: Miscarriage

RESULT SUMMARY
Region of Homozygosity (Uniparental Disomy 14 Suspected) (Female)

H=High, L=Low, *=Abnormal, C=Critical

Size: 5.0 Mb

RESULT DESCRIPTION
This analysis showed a terminal region of homozygosity (ROH) involving chromosome 14 within 14q32.31q32.33. No clinically significant copy number changes were identified.

INTERPRETATION
The clinical significance of this finding is uncertain. The presence of a large ROH increases suspicion for uniparental disomy (UPD). Due to imprinting, UPD of chromosome 14 (UPD14) is associated with either Kagami-Ogata syndrome or Temple syndrome, depending on the parent of origin. Paternal UPD14 is associated with Kagami-Ogata syndrome and maternal UPD14 is associated with Temple syndrome. It is possible this finding is unrelated to the indication for testing (pregnancy loss). Additionally, this finding raises the suspicion for a recessive disorder mapping to this region.

As this finding is not diagnostic for UPD14, clinical correlation is recommended if possible, and additional testing may be warranted.

Detection of homozygosity also increases the suspicion for, but is not diagnostic of, a recessive condition. Online tools available to assist in the identification of candidate recessive genes within the ROH identified in this study: www.sivotecbioinformatics.com/ and https://cma-search.broadinstitute.org/

Recommendations:
1) Genetic counseling
2) Parental chromosome analysis may be considered to rule out a Robertsonian translocation involving chromosome 14 for recurrence risk counseling. This test is available, at a charge, through ARUP Laboratories. Please order test code 2002289, Chromosome Analysis, Peripheral Blood and include the accession number for this case.

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

References:
1) Ioannides et al. Temple syndrome: improving the recognition of an underdiagnosed chromosome 14 imprinting disorder: an analysis of 51 published cases. J Med Genet. 2014 Aug;51(8):495-501. PMID: 24891339.
2) Kagami et al. Comprehensive clinical studies in 34 patients with molecularly defined UPD(14)pat and related conditions (Kagami-Ogata syndrome). Eur J Hum Genet. 2015 Nov;23(11):14881498. PMID: 25689926.
3) Wierenga et al. A clinical evaluation tool for SNP arrays, especially for autosomal recessive conditions in offspring of consanguineous parents. Genet Med. 2013 May;15(5):354-60. PMID: 23100014.
4) Unique: Understanding Rare Chromosome and Gene Disorders. (www.rarechromo.org)

Cytogenomic Nomenclature (ISCN):
arr[GRCh37] 14q32.31q32.33(102318221_107285437)x2 hmz

INTERPRETIVE INFORMATION: Chromosome Analysis,
Products of Conception

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Prod Concp	24-102-102991	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical