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

Physician: Doctor, Example

Patient: Patient, Example

DOB	1/1/1982
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Chorionic Villus

ARUP test code 2002291

Chromosome Analysis, Chorionic Villus

See Note (Ref Interval: Normal)

Test Performed: Chromosome Analysis
Specimen Type: Chorionic Villi
Indication for Testing: Advanced Maternal Age; Abnormal NIPT-T13; Ultrasound Abnormalities: Micrognathia, Polydactyly, Omphalocele, Suspected Cardiac Defect

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

RESULT
Abnormal Karyotype (Male)

Trisomy 13

47,XY,+13

INTERPRETATION
This analysis showed an additional copy (trisomy) of chromosome 13 in each metaphase.

This result is consistent with a clinical diagnosis of trisomy 13. Features associated with trisomy 13 may include heart defects, malformations of the central nervous system, midline defects (holoprosencephaly, microphthalmia, cleft lip with or without cleft palate), renal malformations, polydactyly, clenched fists, rocker-bottom feet, hypotonia, and severe to profound neurocognitive deficits. Trisomy 13 is also associated with high neonatal, and infant mortality.

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 genomic microarray analysis.

NOTE: FISH was performed on this sample and reported under ARUP accession #20337105775. FISH results were ABNORMAL.

Cytogenetic analysis performed on CVS presumes that the fetal chromosome complement is accurately reflected in the extra-embryonic tissue. There are rare examples in which the karyotype of the CVS is not consistent with that of the fetus.

Recommendation:
Genetic counseling

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

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

References and Resources:
1) Pont et al. Congenital malformations among liveborn infants with trisomies 18 and 13. Am J Med Genet A. 2006 Aug 15;140(16):1749-56. PMID: 16835915.
2) Carey and Kosho. Perspectives on the care and advances in the management of children with trisomy 13 and 18. Am J Med Genet C Semin Med Genet. 2016 Sep;172(3):249-50. PMID: 27643592.
3) Andrews et al. Shared decision making and the pathways approach in the prenatal and postnatal management of the trisomy 13 and trisomy 18 syndromes. Am J Med Genet C Semin Med Genet. 2016 Sep;172(3):257-63. PMID: 27557275.
4) Jones et al. Smith's Recognizable Patterns of Human Malformations. 7th edition. Philadelphia, PA: Elsevier Saunders; 2013: 20-23.
5) Support Organization for Trisomy 18, 13, and Related Disorders (SOFT). (www.trisomy.org)

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following location(s):
[redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis, Chorionic Villus
Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

EER Chromosome Analysis Chorionic Villus

See Note

Access ARUP Enhanced Report using the link below:

-Direct access: [redacted]

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Chorionic Villus	20-337-105776	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis Chorionic Villus	20-337-105776	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