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

Physician: Doctor, Example

Patient: Patient, Example

DOB	8/29/1989
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: Abnormal ultrasound, hydrops

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

RRESULT
Abnormal Karyotype (Female)

Trisomy 21 (Down syndrome)

47,XX,+21

INTERPRETATION
This analysis showed an additional copy (trisomy) of chromosome 21 in each metaphase.

This result is consistent with a clinical diagnosis of trisomy 21 (Down syndrome). Features associated with Down syndrome may include hypotonia, a characteristic facial appearance, developmental delays / intellectual disability, and short stature. Other findings may include congenital abnormalities of the heart, diaphragm, and digestive tract, hypothyroidism, and hearing and vision difficulties. Minor physical anomalies commonly seen in individuals with Down syndrome include single transverse palmar crease, low-set ears, and sandal-gap toe. Not all fetuses with Down syndrome will have ultrasound anomalies. Life span may be shortened as compared to the general population, and there is an increased risk of developing leukemia and Alzheimer's disease later in life.

NOTE: FISH was performed on this sample and reported under ARUP accession #(22-336-127395). FISH results were ABNORMAL.

NOTE: Genomic microarray is being performed on this sample and will be reported under ARUP accession #(22-336-127396).

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.

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:
1) Sheets et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. J Genet Couns. 2011 Oct;20(5):432-41. PMID: 21618060.
2) Bull. Committee on Genetics. Health supervision for children with Down syndrome. Pediatrics. 2011 Aug;128(2):393-406. PMID: 21788214.
3) Jones et al. Smith's Recognizable Patterns of Human Malformations. 7th edition. Philadelphia, PA: Elsevier Saunders; 2013:7-13.

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

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.

EER Chromosome Analysis Chorionic Villus

See Note

Authorized individuals can access the ARUP Enhanced Report using the following link:
[redacted]

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Chorionic Villus	22-347-402143	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis Chorionic Villus	22-347-402143	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