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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/29/1976
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray

ARUP test code 2008367

Chromosome Analysis, Amniotic Fluid

See Note (Ref Interval: Normal)

```
-------------------------------------------------------------
Test performed:      Chromosome Analysis
Specimen type:       Amniotic Fluid
Reason for referral: Abnormal ultrasound, cardiac defects,
clenched hands, SUA

Laboratory analysis

Number of cells counted:    15
Number of colonies counted: 15
Number of cells analyzed:   15
Number of cells karyotyped: 15
ISCN Band level:            400
Banding Method:             G-Banding
-------------------------------------------------------------
RESULT
Abnormal Karyotype (Female)

Trisomy 18 (Edwards syndrome)

47,XX,+18
-------------------------------------------------------------
INTERPRETATION
This analysis showed an additional copy (trisomy) of chromosome
18 in each metaphase.

This result is consistent with a clinical diagnosis of trisomy
18 (Edwards syndrome). Features associated with trisomy 18 may
include intrauterine growth restriction with low birth weight,
multiple congenital anomalies involving the brain, spinal cord,
heart, abdominal wall and kidneys, hypotonia at birth
progressing to hypertonia in later infancy, feeding
difficulties, and severe to profound intellectual disability and
developmental delay. Other findings may include abnormal head
shape, a small jaw (micrognathia) and mouth, low-set, malformed
ears, clenched fists with overlapping fingers, rocker-bottom or
clubfeet, cystic hygroma and increased nuchal translucency.
Trisomy 18 is also associated with high prenatal, neonatal, and
infant mortality.

NOTE: Since this analysis revealed an abnormal result, genomic
microarray analysis, which was ordered as a reflex study, will
not be performed unless we are notified otherwise. To proceed
with this testing, please contact ARUP Genetics Processing at
(800) 242-2787 ext. 3301.

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
```

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

genomic microarray analysis.

Recommendation:
Genetic counseling

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

References:
1) Cereda and Carey. The trisomy 18 syndrome. Orphanet J Rare Dis. 2012 Oct 23;7:81. PMID: 23088440.
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; 2013:14-19.
5) The Trisomy 18 Foundation Support Group. (www.trisomy18.org)
6) The 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, Amniotic Fluid

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 Chrom Analysis AF w/Rflx to Array

See Note

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

[redacted]

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Amniotic Fluid	22-179-154583	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chrom Analysis AF w/Rflx to Array	22-179-154583	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