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, Skin Biopsy

ARUP test code 2002286

Chromosome Analysis, Skin Biopsy	Abnormal	*	(Ref Interval: Normal)

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

Test Performed: Chromosome Analysis
Specimen Type: Skin
Indication for Testing: Trisomy 18

Number of cells counted: 20
Number of cells analyzed: 5
Number of cells karyotyped: 5
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 neurocognitive deficits. Other findings may include abnormal head shape, a small jaw (micrognathia) and mouth, low-set, malformed ears, clenched fists with overlapping fingers, and rocker-bottom or clubfeet. Trisomy 18 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.

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]

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

INTERPRETIVE INFORMATION: Chromosome Analysis, Skin Biopsy

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, Skin	EERUnavailable

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Skin Biopsy	24-135-103516	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis, Skin	24-135-103516	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