## SAMPLE REPORT



## **Chromosome Analysis**

Patient Name: Sample Patient

Referring Physician: John Doe, M.D.

Specimen #: 10000000

Client #: 12345

Patient ID: 20000000-1

DOB: 00/00/1971 SSN: \*\*\*-\*\*-

Date Collected: 09/19/2012 Date Received: 09/20/2012

Lab ID: Hospital ID:

Specimen Type: AMNIO

Indication: Abnormality seen on ultrasound - multiple congenital anomalies (MCA)

**Metaphases Counted:** 

15

Colonies Counted:

13

**Banding Technique:** 

**GTW** 

Metaphases Analyzed:

5

Number of Cultures: 3

**Banding Resolution:** 

400

Metaphases Karyotyped: 2

Subculture:

N

Dept. Section:

City Hospital 1 Main Street

Anywhere, USA

A1

RESULTS: 69,XXY

Abnormal karyotype, male

## **INTERPRETATION:**

Cytogenetic analysis shows three copies of each chromosome, resulting in triploidy.

Triploidy is associated with multiple malformations, neurologic dysfunction and severe failure to thrive. It is a common cause of first trimester spontaneous abortions. Triploid conceptions which survive longer in pregnancy are often delivered prematurely or are stillborn. If liveborn, they rarely survive longer than a few days to a few months. Approximately 69% of triploidies are diandric partial hydatidiform moles, in which two of the three sets of chromosomes are paternally derived (Jones, K.L., Smith's Recognizable Patterns of Human Malformation, 5th edition. Philadelphia: Elsevier, Inc., 2006. Pp. 28-31).

## COMMENT:

No other chromosome abnormalities are observed. The standard cytogenetic methodology utilized in this analysis does not routinely detect subtle rearrangements or low-level mosaicism and cannot detect microdeletions. Also, it cannot detect molecular cytogenetic abnormalities (such as microdeletions and microduplications) that may be detectable by microarray analysis.

Genetic counseling is recommended for this family.

Follow-up confirmatory analysis is available.

Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

Signed:

Date: 09/28/2012