



Chromosome Analysis

Patient Name: Sample Patient

Referring Physician: John Doe, M.D.

Patient ID: 20000000-1

DOB: 00/00/1995 Date Collected: 09/19/2012 SSN: ***-*** Date Received: 09/20/2012

Lab ID: Hospital ID:

Specimen Type: Peripheral Blood

Indication: Amenorrhea

City Hospital 1 Main Street Anywhere, USA

Metaphases Counted:20Banding Technique:GTWMetaphases Analyzed:5Number of Cultures:2Banding Resolution:550Metaphases Karyotyped:2Dept. Section:B1

RESULTS: 45,XX,der(13;14)(q10;q10)

Translocation karyotype, female

INTERPRETATION:

Cytogenetic analysis shows an apparently balanced Robertsonian translocation between the long arms of one chromosome 13 and one chromosome 14.

A balanced rearrangement carried by a parent can result in unbalanced gametes, and can lead to decreased fertility, miscarriage, or chromosomally abnormal offspring.

RECOMMENDATION:

Prenatal diagnosis (including UPD analysis, when appropriate) should be offered in future pregnancies, and blood chromosome analysis should be offered to family members who could also be carriers.

Genetic counseling is recommended for this family.

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.

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Signed: Date: 09/27/2012