CASE 045

CŌRE DIAGNOSTICS™

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CASE HISTORY

A 28 year old male clinically diagnosed as Klinefelter syndrome, was referred to our clinical cytogenetic laboratory for chromosomal analysis studies.

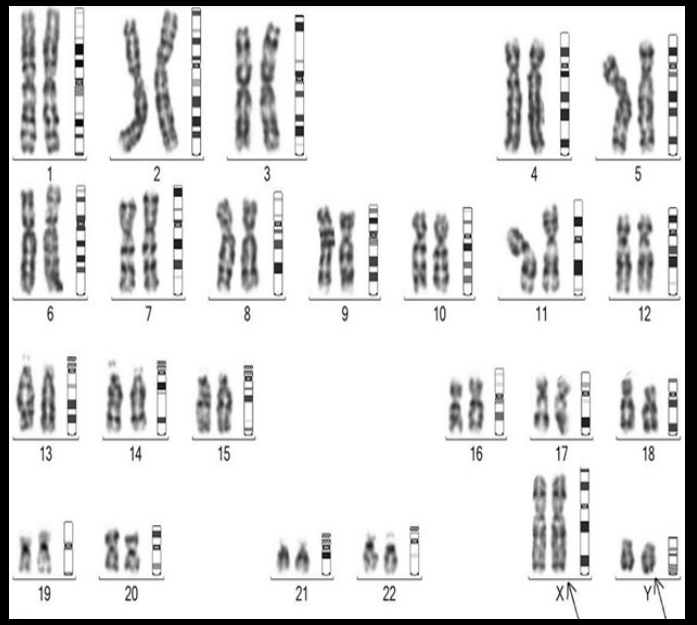
MATERIAL AND METHOD

Chromosomal analysis was performed on 2 ml heparinized peripheral blood sample using 72 hours stimulated cultures with appropriate serum, antibiotics and Phytohemagglutinin. A total of 30 metaphases were analyzed through GTG-banding (G bands by trypsin and Giemsa) according to International System for Human Cytogenetic nomenclature 2016. All Images were captured using fully automatic Olympus BX63 microscope and analyzed using Bio-view karyotyping software.

FINAL DIAGNOSIS

Chromosome analysis revealed an abnormal male chromosome complement in all metaphases examined with the presence of an extra X and Y chromosome. This result is consistent with the clinical diagnosis of Variant Klinefelter syndrome.

The karyotype results were 48, XXYY (48,XY,+X,+Y). The additional X and Y chromosomes showed parental origin.



Result: 48,XXYY (Variant Klinefelter Syndrome)