

CASE 047

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PATIENT HISTORY

A 15 year old girl who was clinically diagnosed as Turner syndrome with short stature and pubertal delay was referred to our cytogenetic laboratory for chromosomal analysis.

MATERIAL AND METHOD

Chromosomal analysis was performed on 2ml heparinized peripheral blood sample. The cultures were incubated for 72 hours and stimulated by Phytohemagglutinin (PHA). A total of 30 metaphases were captured and 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.

RESULTS

Chromosome analysis revealed an abnormal female chromosome complement in all metaphases examined with the presence of a normal X chromosome and an aberrant X chromosome consisting of twice the long arm and short arm of the X chromosome [nonmosaic 46,X,psu idic(X)(p22.3)].

