CASE 073

CŌRE DIAGNOSTICS™

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CASE HISTORY:

A 3-days-old female child suspected to have some genetic disorder based on the phenotype and was referred to the cytogenetic laboratory for chromosomal analysis.

MATERIALS AND METHODS:

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 analysed through GTG-banding (G bands by trypsin and Giemsa) according to the International System for Human Cytogenetic nomenclature, 2018. All Images were captured using fully automatic Olympus BX63 microscope and were analysed using Bio-view karyotyping software.

FINAL DIAGNOSIS:

Chromosome analysis revealed an abnormal female karyotype in all metaphases examined with trisomy of chromosome 13. The trisomy 13 has resulted from a robertsonian tranlocation between chromosome 13 and 14. The karyotype result was 46,XX,rob(13;14)(q10;q10),+13. The karyotype result is consistent with the clinical diagnosis of Patau's syndrome (Trisomy 13).