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Molecular Cytogenetic Evaluation of Xq Deletion Mosaicism in a Case of Primary Amenorrhea

Babu Rao Vundinti , Lily Kerketta , Seema Korgaonkar, Kanjaksha Ghosh and Dipika Mohanty

Institute of Immunohaematology (ICMR) 13th floor, New Multistoryed Building, K.E.M Hospital Campus, Parel, Mumbai 400 012, Maharashtra, India

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ABSTRACT The X chromosome contains determinants necessary to assure normal ovarian function and normal statural growth. Deletions of proportions of the X chromosome have been reported in a large number of patients, most which are isolated. We report a mosaic long arm deletion of X chromosome (Xq-) in a women with primary amenorrhea. Chromosomal analysis using GTG-banding showed two cell lines(45,X/46,X,del(X)). The Fluorescence *in situ* hybridization study helped to detect the breakpoint at q13.3 region of the X chromosome and also enable to detect low grade third cell line with 47,XX,del(X)(q13.3). Hence, molecular cytogenetic methods are essential to detect low level mosaicism which is important in better counselling.

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