

Trisomy - T(21;21) with Mosaicism in a Down Syndrome Girl Child Case Report

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KEYWORDS Chromosomal Aberration. Cytogenetics. Mental Retardation. Nondisjunction. Translocation

ABSTRACT Mosaicism with a derivative of 21 translocation and normal cell line is one of rare cases of Down syndrome patients. In the present case dysmorphic features and developmental delay were compatible with clinical diagnosis of Down syndrome. Cytogenetic analysis demonstrated a mosaic pattern of normal cell line and a cell line with translocation 21;21[(46,xx/46,xx,T(21;21),+21]. Presence of normal cell line as well as translocated 21 indicates the mitotic nondisjunction (NDJ) of an euploid zygote and mosaicism may be due to loss of the supernumerary chromosome of a trisomic zygote or some complex mechanism may be involved in chromosomal aberration. Present case shows the mosaicism in a girl child with features of Down syndrome and atypical karyotype.