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Cytogenetics of Fragile X Chromosome: Autosomal Sites as Potential Markers for Fragile X Chromosome Analysis

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KEY WORDS Fragile X chromosome; autosomal mimics; autosomal indicators; differential diagnosis.

ABSTRACT Fragile X chromosome screening was undertaken among 100 non-specific mentally retarded group from the Indian population. 14 subjects from 10 families showed the presence of fragile X chromosomes in 4 to 40% of cells. Various 'C' group autosomes showed the fragile sites at the telomeric regions of long arm in low percentages, which is well enough to be confused with fragile X chromosome manifestation in unbanded chromosome preparations. The necessity of analysing good G-banded preparations and scoring more number of cells for the confirmation of fragile X syndrome cytogenetically has been stressed. The presence of constitutive fragile site at 3p14 and polymorphic 9qh+ in high percentages among fragile X positive subjects, made these markers a potential indicators in the diagnosis of fragile X syndrome. The implications of the autosomal mimics and indicators in the differential diagnosis of fragile X syndrome has been highlighted.

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