ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 12(3): 157-171 (2012)
DOI: 10.31901/24566330.2012/12.03.05

Cryptic Rearrangements in Idiopathic Intellectual Disability Diagnosed by Molecular Cytogenetic Analysis

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KEYWORDS Dysmorphisms. FISH. Karyotype. Subtelomeric Anomalies. Telomeres.

ABSTRACT With the development of molecular cytogenetic techniques, it is possible to identify cryptic rearrangements involving the end of chromosomes. Subtelomeric chromosomal rearrangements represent a significant cause of idiopathic intellectual disability accounting for 6-10% of moderate to severe cases and 0.5% in individuals with mild intellectual disability. We investigated 50 patients with severe intellectual disability combined with a dysmorphic features and normal 400-550 band karyotype for unbalanced subtelomeric rearrangements by using fluorescence in situ hybridization with probes mapping to forty one telomeric-specific regions. Nine positive cases (18%) were found. Six were de novo deletions (1p, 2q, 6p, 9q, 10q, 22q) and one wasis de novo duplication (10q) .Two unbalanced translocation (a der(3)t(3p; 2q) and a der(3)t(3p; Xq)) were inherited from the balanced mothers. Our study supportsed the hypothesis that subtelomeric rearrangements are a significant cause of idiopathic intellectual disability. The clinical features of patients with subtelomeric abnormalities and the candidate genes proposed inside each region will help to better delineate the phenotype- genotype correlation.