

Cryptic Rearrangements in Idiopathic Intellectual Disability Diagnosed by Molecular Cytogenetic Analysis

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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 was 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 supported 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.