عنوان مقاله:

A Rare Case of Duplication of Chromosome 2 (q31.3q36.3) in a 4.5-year-old Boy and Review of the Literature

محل انتشار:

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نویسندگان:

Maryam Abolhasani - *Molecular Pathology and Cytogenetic Section, Pathology Department, Medical Faculty, Tehran University of Medical Sciences, Tehran, Iran.*

Mohammad Vasei - *Oncopathology Research Center, Iran University of Medical Sciences, Tehran, Iran.*

Moeinadin Safavi - *Department of Pathology, Children s Medical Center, Tehran University of Medical Sciences, Tehran, Iran.*

خلاصه مقاله:

De novo duplication of 2q is very rare. Most cases of 2q duplications result from familial translocations, and are associated with simultaneous monosomy of another chromosome segment. To our knowledge and search in English literature there are less than 20 reported cases of isolated 2q duplication. Hereby we introduce a 4.5-year-old Iranian boy of a non-consanguineous marriage who was referred for cytogenetic study due to developmental delay and intellectual disability. He also had short stature and dysmorphic facial features. He had depressed broad nasal bridge and broad nasal tip, long philtrum and thin upper lip. His hands were edematous and the first phalanxes were broad and the thumbs were larger than normal. The chromosomal analysis revealed isolated 2q31.3q36.3 duplication, and array comparative genomic hybridisation (CGH) confirmed the diagnosis. After six months follow- up, could not walk or speak despite occupational therapy. We also, describe the common morphological characteristics of isolated 2q duplication.

کلمات کلیدی:

Array CGH, Karyotype, Developmental delay, 2q duplication