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De novo interstitial deletion in the long arm of chromosome 11: a case report

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ABSTRACT. The 11q terminal deletion disorder is a rare genetic disorder associated with numerous clinical features. A few case reports have been made about *de novo* interstitial deletion of chromosome 11q. However, due to the heterogeneity in size and position of the deletions, a clear genotype-phenotype correlation is not easily made. Here we report a case interstitial 20.5-Mb deletion at chromosome 11q13.4q21, as confirmed by array comparative genomic hybridization. Dysmorphic features such as coarse facial features, congenital laryngomalacia, oblique inguinal hernia, high-arched palate, and camptodactyly were observed in the subject. The present case broadens the spectrum of clinical findings observed in individuals with 11q interstitial deletion.

Key words: Interstitial deletion; Array comparative genomic hybridization; Chromosome 11; Genotype-phenotype correlation

