# A rare 19p13.11-p13.12 deletion in a patient with microcephaly and developmental delay

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## Abstract/Resumo

Submicroscopic deletions in chromosome 19 have been rarely reported. The aim of this study was to describe the genotype-phenotype correlation of a 19p13.11-p13.12 deletion. A male patient was referred for genetic evaluation by presenting neurodevelopmental delay and facial dysmorphisms. He is the first child of non-consanguineous parents. Pregnancy and delivery were uneventful. Anthropometrical data at seven-year-old were weight 25,4 kg (p75), length 120 cm (p50) and head circumference 47,3 cm (< -2SD). It was also observed malar hypoplasia, low-set and dysmorphic ears, bilateral preauricular tags, synophrys, short nose with anteverted nares and flat nasal bridge, down-turned corners of the mouth, long philtrum, micrognathia, high palate and congenital clubfoot. Abdominal ultrasonography showed pyelocaliceal ectasia, transfontanellar ultrasound revealed mild enlargement of magna cisterna and a small cyst ate nucleous caudatus. GTG-banding was normal and chromosomal microarray analysis (CMA), using the CytoScan HD Array (Affymetrix®, Santa Clara, CA, USA) showed an interstitial deletion of approximately 1,4 Mb at the short arm of chromosome 19: arr[GRCh37] 19p13.11-p13.12 (14856624_16340130)x1. The parents were investigated with the Agilent SurePrint G3 Human CGH Microarray 8x60K (Agilent Technologies, Santa Clara, CA, USA) which revealed normal results. The chromosome 19 is one of the richest in number of genes and deletions in this chromosome are rarely reported. To date, there are eight cases with deletions overlapping the 19p13.11-p13.12 region. In the database of genomic variation and phenotype in humans using Ensemble Resources (DECIPHER) we found seven cases with deletions that overlap this region and five of them also present microcephaly and developmental delay. This overlapping region includes seven OMIM genes: ILVBL, SYDE1, NOTCH3, EPHX3, BRD4, AKAP8 and AKAP8L. Mutations in NOTCH3 lead to cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) but the effect of deletions of this gene is unknown. It was suggested that alterations of dosage in AKAP8 and AKAP8L genes might affect the head circumference due to regulation of cell proliferation in the brain. Considering the rarity of the genomic imbalances involving 19p13.11-p13.12 region, the case herein reported contributes to the phenotype delineation. Additional cases would improve the genotype-phenotype correlation.

Keyword/Palavras-chave: 19p13.11-p13.12 deletion; chromosomal microarray analysis (CMA); genotype-phenotype correlation

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