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Genotype/Phenotype correlation with partial duplications of 6p and 6q

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Abstract/Resumo

Partial trisomy 6p and 6q are rare and clinical features include craniofacial changes, mental retardation and developmental delay. The study aims to correlate the genotype/ phenotype of a 6p and 6q duplication carrier. MVSN, female, single daughter of nonconsanguineous and healthy young couple. From an earlier relationship, the father had a 13-year-old son. Delivered by C-section, gestation of 37 weeks. The clinical analysis revealed: microcephaly, brachycephaly, syndromic facies, prominent forehead, hyperarousalism, strabismus, short nose, anteverted nares, smooth philtrum, thin lip, upper lip tented, micrognathism, dental anomalies, low-set ears, slender fingers, nails malformation, digitiform thumb, bilateral flat foot, hypotonia and neuropsychomotor developmental delay (NPMD). In the physical examination performed at 2 years of age, weight was 7,570 Kg (<3,58%), length was 74 cm (<3,95%) and cephalic perimeter was 43 cm (<2%). Classic cytogenetic techniques were performed (GTG banding and high resolution GTG), molecular cytogenetic (FISH) and cytogenetic (aCGH). The karyotype showed a mosaicism 47, XX, + mar [9]/46, XX [11] (ISCN, 2016). The parental karyotype was normal. The chromosomal changes found by aCGH indicate a genetic gain at chromosomal regions 6p11.2→q12 and 6q14.1→q14.3 with a size of 10.335 Mb and 10. 765 Mb, respectively. The patient presented mosaicism of a marker chromosome (sSMC) identified by the technique of FISH as coming from chromosome 6. In region 6p11.2→q12 the genes present are: PRIM2, GUSBP4, MTRNR2L9, KHDRBS2, LGSN, PTP4A1, PHF3, EYS and MCART3P. Heart defects and kidney problems were cited in the literature as being characteristic of trisomy 6p. However, patient did not present any of these phenotypic changes. The region 6q14.1→q14.3 contains more than 20 genes. The clinical signs found in patient such as: developmental delay, mental retardation, forehead prominent, short nose and thin lips has been described in other studies in the literature as characteristic of 6q trisomy. The presence of mosaicism excludes the possibility of sSMC as the sole responsible for the patient's phenotype. Our study correlates the phenotypic characteristics presented by the patient with partial trisomy 6q collaborating for family genetic counseling.

Keyword/Palavras-chave: Trisomy 6p and 6q; Marker chromosome (sSMC); Developmental delay

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