Trabalhos Científicos

Título: Ring Chromosome 13 Syndrome In A Patient With Developmental Delay And Anomalies Characterized By Array Comparative Genomic Hybridization

Autores: CESAR ALFONSO GOMEZ FONTALVO (UNIVERSIDAD INDUSTRIAL DE SANTANDER); GUSTAVO ADOLFO CONTRERAS GARCIA (UNIVERSIDAD INDUSTRIAL DE SANTANDER); VIVIANA LUCIA PEREZ FORERO (UNIVERSIDAD INDUSTRIAL DE SANTANDER)

Resumo: INTRODUCTION: The ring chromosome 13 syndrome is caused by structural and functional regions monosomy of chromosome 13. It has a low incidence, estimated at 1/ 58,000 in live births. According to cutoff allowing deletion ring formation occurs one variant phenotype dividing into 3 groups syndrome. Group 1: deletion from the proximal region to 13q32, cognitive disorder characterized by mild to moderate and facial abnormalities. Group 2: deletion of 13q32, is associated with a more severe phenotype that includes multiple malformations, short stature and cognitive impairment. Group 3: distal deletions of 13q33-34 bands, generates cognitive disorder, microcephaly and genital malformations in men; usually it is not associated with other abnormalities. CASE REPORT: A 5-year-old female patient attending consultation for developmental delay from eight months. No family pathological history or consanguinity. First pregnancy product, present oligoamnios and fetal growth retardation. Birth of 37 weeks, weight 1,9 Kg, length 42 cm. Uncomplicated delivery. She has short stature proportionate, brachycephaly, strabismus, epicanthic folds, unilateral ptosis, low-set ears, smooth philtrum, midface hypoplasia, abnormality of external genitalia: hypoplastic labia minora, clinodactyly of hallux and generalized hypotonia. Showed spinal defect closure in L5, patent ductus arteriosus and tricuspid insufficiency. G-banding karyotype high resolution was performed: 46,XX,r(13)(p13q33). It was decided to perform research array comparative genomic hybridization (aCGH): arr[hg18] 13q33.2q34(104,524.989-114,101,444)x1. Parental karyotype normal. DISCUSSION: Partial deletions of chromosome 13 are rare. The clinical presentation have expression variability with different afected systems. Agree on clinical features, these deletions have been categorized into three groups. We compared the clinical features of our patient with previously published patients with deletions of chromosome region 13q33–34. CONCLUSION: Patients with developmental delay should be performed cytogenetic study including array analysis to identify whether the cause of neurodevelopmental disorder is chromosomal origin. This result is important for prognosis and genetic counseling to the family.