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CLEFT LIP/PALATE, SHORT STATURE AND DEVELOPMENTAL DELAY IN A BOY WITH 5,6 MB INTERSTITIAL DELETION INVOLVING 10P15.3-P14

FREITAS LT, Gamba BF***, Rosenberg C, Richieri-Costa A, Ribeiro-Bicudo LA

Laboratório de Genética Molecular, Hospital de Reabilitação de Anomalias Craniofaciais - HRAC-USP, Bauru/SP

OBJECTIVE: The chromosome interval involving 10p15.3-p14 harbors some dozen of genes. To date this region has been implicated in few well known human phenotypes, namely the HDR syndrome (hypoparathyroidism, sensorineural deafness, and renal dysplasia) and the DGS2 (DiGeorge syndrome 2), but several variable phenotypes has been also reported. Deletions of different sizes restricted to 10p15 seem to present clinical findings with no significant phenotypic differences and haploinsufficiency of two major genes – ZMYND11 and/or DIP2C harbored in this region may be responsible of isolated, or minimally syndromic intellectual disability. **CLINICAL REPORT:** In the present work we report a boy referred to our Hospital at the age of 3 months for assessment and management of cleft lip/palate. He was the first child of a normal and unrelated couple. Pregnancy was unremarkable with no exposure to known teratogens. There was no relevant family history. Neuropsychological development was delayed. At present age he has normal cervical control, is able to roll over, but he cannot sit without support. Behavioral hearing tests were inconclusive since he presented impacted cerumen and an evaluation through auditory evoked potential was proposed. **CONCLUSION:** Array Comparative Genomic Hybridization (arrayCGH) analysis showed a large 5 Mb deletion on 10p15.3-p14. The haploinsufficiency of some genes in the deleted region may be responsible for the mild phenotype presented by the patient.

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