Interesting case report ECARUCA (2016-03)

A novel de novo 1.8 Mb microdeletion of 17q21.33 associated with intellectual disability and dysmorphic features

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Abstract:
We report on a de novo 17q21.33 microdeletion, 1.8 Mb in size, detected in a patient with mild intellectual disability, growth retardation, poor weight gain, microcephaly, long face, large beaked nose, thick lower lip, micrognathia and other dysmorphic features. The deletion was detected by whole-genome genotyping BeadChip assay and involves the genomic region between 45,682,246 and 47,544,816 bp on chromosome 17. Among the 24 RefSeq genes included in this deletion are the CA10 and CACNA1G genes that are involved in brain development and neurological processes. A possible candidate gene for the prenatal and postnatal growth retardation is the CHAD gene, which product chondroadherin is a cartilage protein with cell binding properties. These three genes may be responsible for the patient's phenotype.

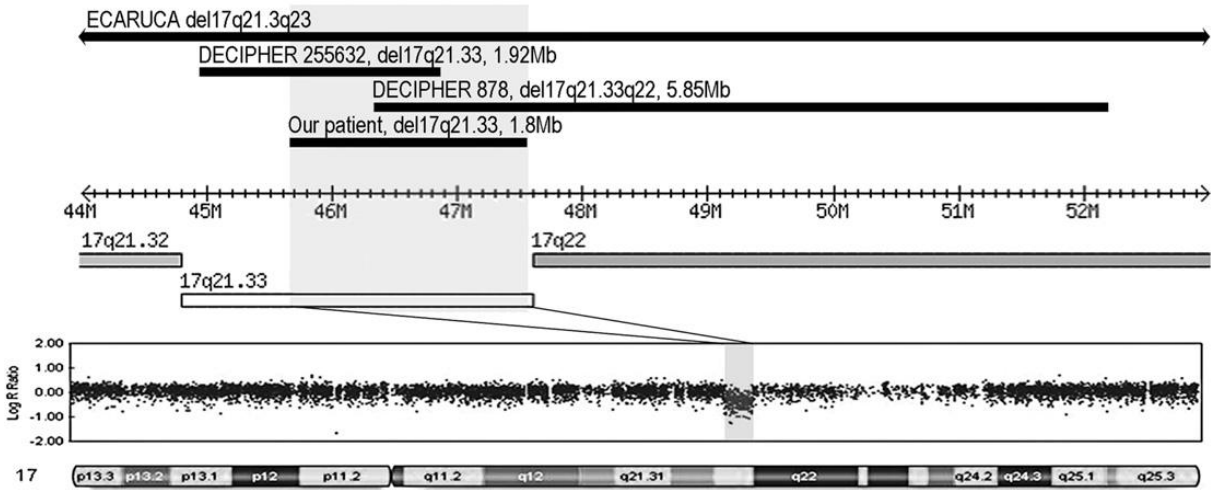

Fig. 1. 1.8 Mb deletion in chromosome band 17q21.33, between 45,682,246 bp and 47,544,816 bp (Build 36:Mar. 2006) detected using Infinium HD whole-genome genotyping assay with the HumanCytoSNP-12 BeadChip is presented in the bottom. Above the map of deleted (in black lines) and duplicated (in white lines) regions according DECIPHER, ECARUCA and previously reported cases in literature is presented.

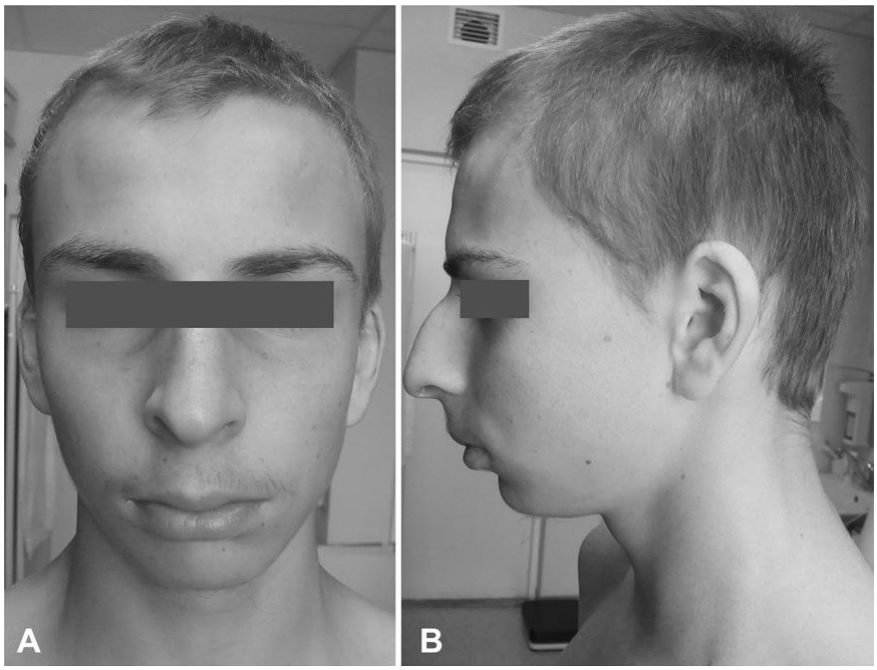

Fig. 2. Front and side lateral views of the patient. Note long face, thick lower lip (A), large beaked nose and micrognathia (B).