Interesting case report ECARUCA (2015-11)

Molecular cytogenetic characterization of 2p23.2p23.3 deletion in a child with developmental delay, hypotonia and cryptorchism

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Abstract:
Deletions of the short arm of chromosome 2 are exceedingly rare and only nine cases involving regions from 2p23 to 2pter have been reported to date. Most of these deletions had only been analysed by GTG banding. Here, we report an interstitial de novo deletion resulting in a microdeletion of 3.9 Mb involving 2p23.2-p23.3 segment, detected by SNP-array analysis, in a 5 year-old boy showing hypotonia, overweight, dysmorphic facial features and cryptorchidism. We compared the clinical features of the present case to previously described patients with deletions within this chromosomal region. Our case adds new information to the deletion of the distal part of chromosome 2p improving the knowledge on this rearrangement.

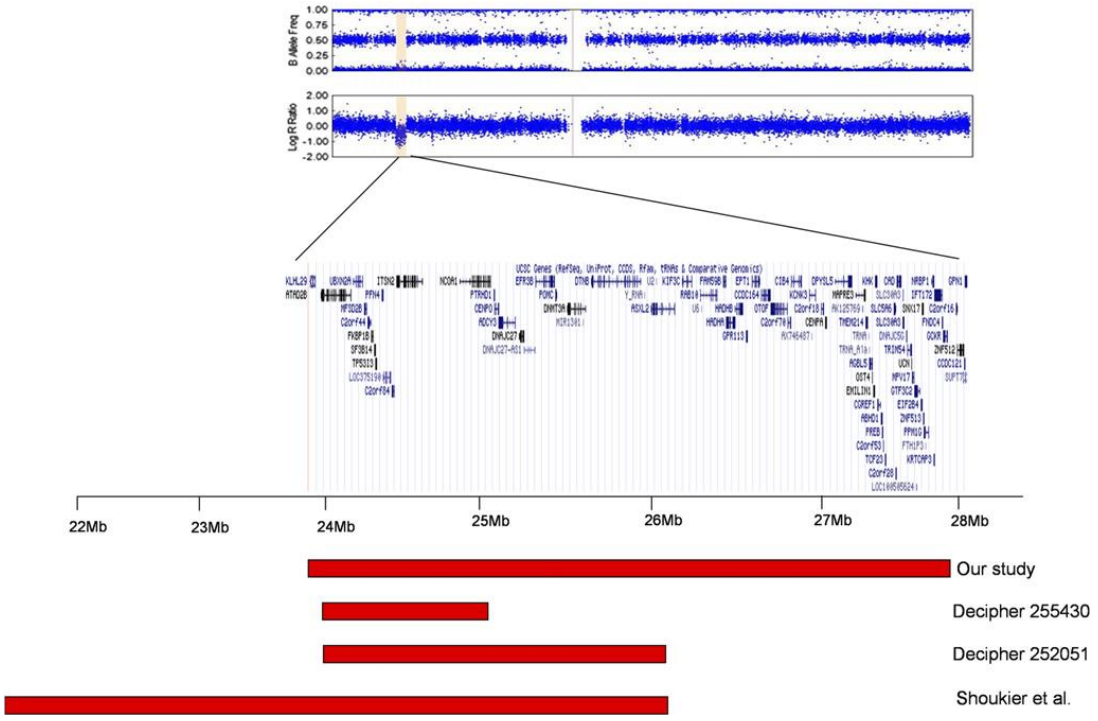

Fig. 1. Data from Human-370 BeadChip and schematic representation of deleted region on 2p23.2p23.3 in our patient in comparison with the patients with overlapping deletions characterized by molecular methods. Screenshot obtained from UCSC Genome Browser on human May 2009 Assembly.

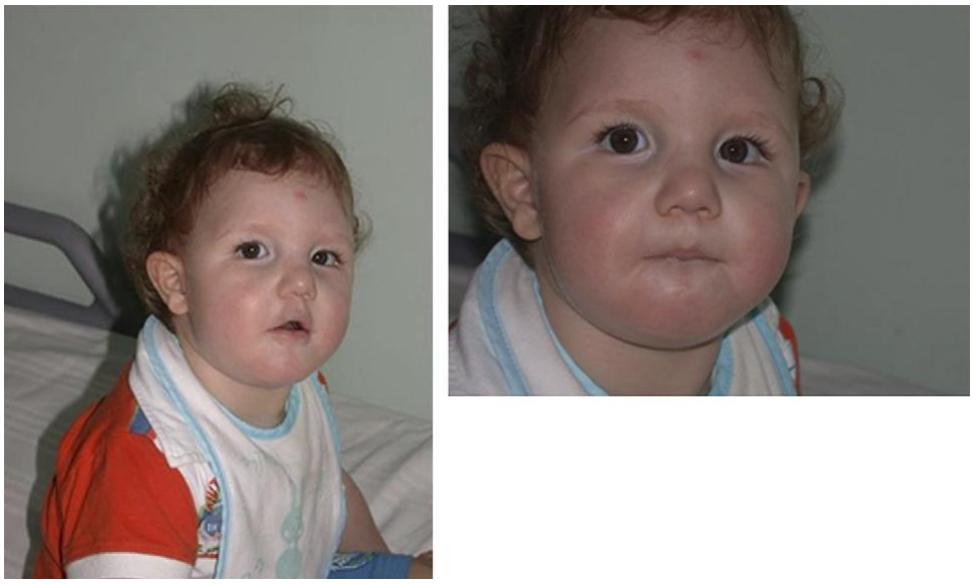

Fig. 2. Facial image of the proband.