

## Interesting case report ECARUCA (2014-03)

### A 1 Mb de novo deletion within 11q13.1q13.2 in a boy with mild intellectual disability and minor dysmorphic features

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#### Abstract:

We report a 11 year old male patient ascertained for mild intellectual disability and minor dysmorphic

features, carrying a 1Mb de novo deletion on chromosome 11q13.1q13.2 detected by aCGH. This is the first report of a deletion in this region in a patient presenting with intellectual impairment and mild dysmorphic traits. The 1 Mb deleted area encompasses 47 RefSeq genes, including Cornichon homologue 2 (CNIH2), Cofilin-1 (CFL1) and neuronal PAS domain-containing protein 4 (NPAS4), which are highly expressed in the central nervous system. Knockout of the CNIH2 and CFL1 orthologues in animals results in migration disturbances, while low or no expression of Npas4 in mice results in impairment of memory and learning. These three genes have previously been suggested as candidate genes for neurological disorders.

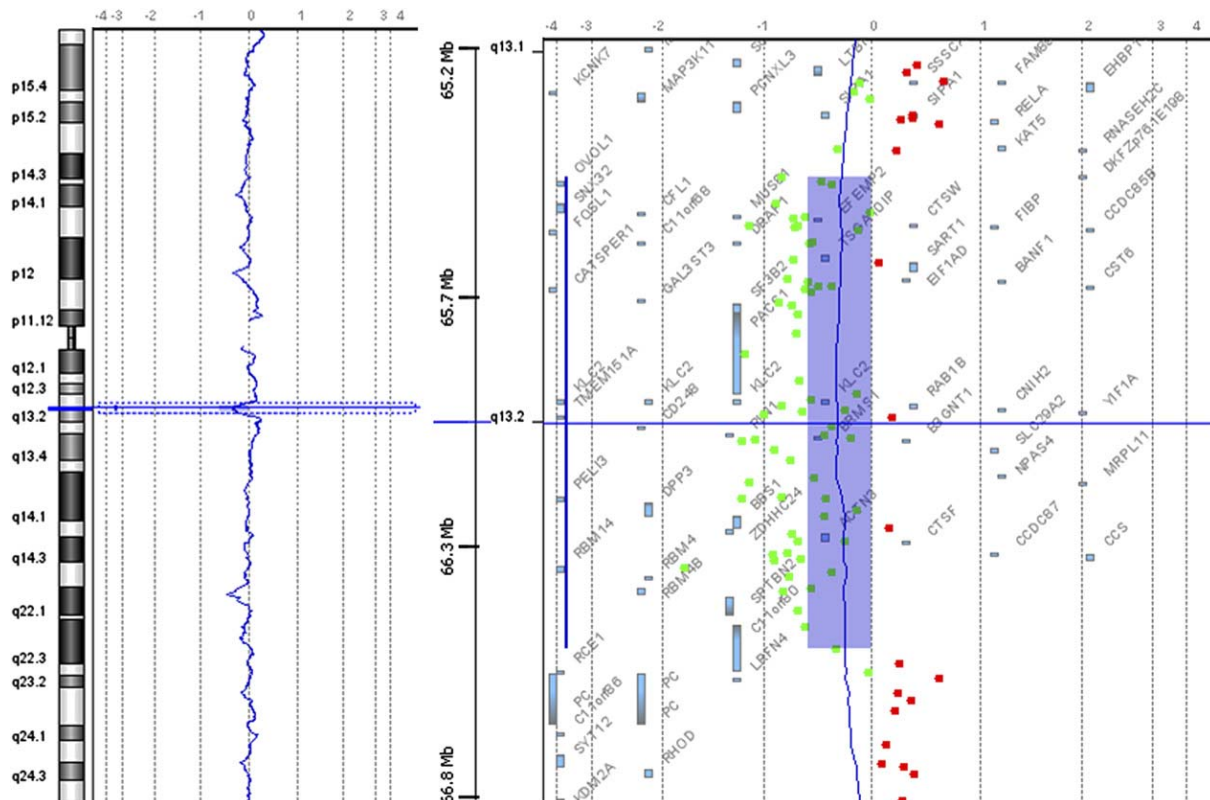


Fig. 1. Graphical output of the aCGH results. On the left an overview of whole chromosome 11; on the right a detailed view of the deleted region (chr11:65544038e66563827, hg 19). Dots represent the oligos along the chromosome. In green the oligos in the deleted region (ratio patient Cy3 to control Cy5 is  $< -0.4$ ); in red oligos with a normal ratio (between  $-0.4$  and  $0.4$ ). The area highlighted in blue indicates the extension of the minimal deletion.

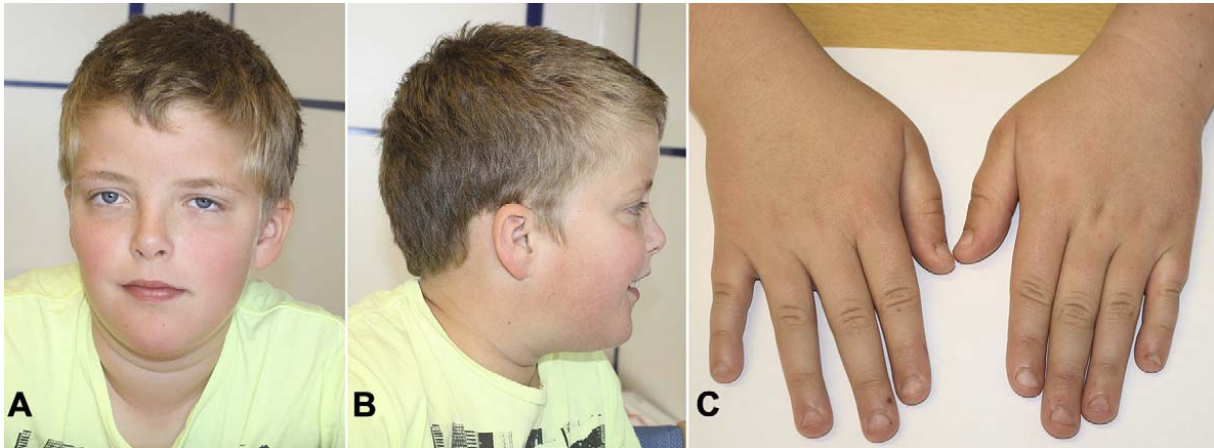


Fig. 4. The patient at the age of 10 years. Shown with written consent from the patient's family. (A) Facial features. Note the small nose, anteverted nostrils and down slanted palpebral fissures. (B) Profile of the patient. Note the thick upper helix of the ear. (C) Hands. Note the small/hypoplastic fingernails.