Interesting case report ECARUCA (2014-01)

A de novo 3.57 Mb microdeletion in 8q12.3q13.2 in a patient with mild intellectual disability and epilepsy

Willem M.A. Verhoeven, Jos I.M. Egger, Ilse Feenstra, Nicole de Leeuw


Abstract:
A female patient, nine years of age, is reported with a history characterized by delay of psychomotor and speech development, mild to moderate intellectual disability and persistent sleep disturbances since the age of two. The patient showed facial dysmorphisms, a pectus excavatum and a sandal gap. Apart from lowered intelligence, neuropsychological functioning disclosed impaired attentional capacities and executive control as well as weak motor skills. Genome wide SNP array analysis revealed a 3.57 Mb de novo microdeletion in band q12.3 of chromosome 8. The long lasting sleep disorders turned out to originate from a rare juvenile epilepsy, continuous spike-waves during slow sleep (CSWS) syndrome, that includes the electrical status epilepticus in sleep (ESES) phenomenon. MRI-scanning of the brain showed no abnormalities. To the authors knowledge, this is the first report of a de novo 8q12.3q13.2 microdeletion syndrome that presents with ESES/CSWS.

Fig. 1. A. log2 intensity ratios of the SNP probes on chromosome 8 obtained with 250k SNP array analysis, showing the interstitial microdeletion in 8q12.3q13.2 (arr 8q12.3q13.2(65,236,018–68,801,113) × 1 dn) detected by significantly lowered values of 221 consecutive probes. B. Screen shot of the UCSC Genome Browser (http://genome.ucsc.edu/; Human Genome, February 2009 (GRCh37/hg19) assembly) showing the deleted region 8q12.3q13.2 encompassing a total of 27 coding genes, including one known recessive disease gene CYP7B1 in 8q12.3.
Fig. 2. Female patient at the age of 9 1/6 years, presenting several characteristic facial features including hypertelorism, downslanting palpebral fissures, a long, pear shaped nose, low-set, posteriorly rotated ears, and a full lower lip. The extremities show a bilateral sandal gap and flat feet.