Interesting case report ECARUCA (2014-04)

Delineating the 17q24.2eq24.3 microdeletion syndrome phenotype

Jodi M. Lestner, Richard Ellis, Natalie Canham

http://www.sciencedirect.com/science/article/pii/S176972121200242X

Abstract:
We present an 11-year-old girl with a 2.3 Mb de novo interstitial deletion in chromosome 17q24.2eq24.3 identified by array CGH. The phenotype in this case includes skeletal malformations (lower limb bowing, progressive scoliosis and dental abnormalities), feeding problems, mild learning difficulties, and a characteristic facial appearance. Much of the phenotype is attributable to the deletion of KCNJ2, which causes Andersen Tawil Syndrome (ATS), but the facial appearance is not typical. We hypothesise that the presence of mild channelopathy-related features seen in ATS may be explained by haplo-insufficiency, leading to a reduced number of functionally normal Kir2.1 channels. Comparison is made to previous reports describing overlapping 17q deletions, and potential candidate genes which account for the specific phenotypic similarities with this case are highlighted.

Fig. 1. A: Representative array CGH data profile for chromosome 17 showing deletion. B: Chromosome 17 region q24.2eq24.3 adapted from the database of genomic variants (http://projects.tcag.ca/variation, genome build hg19) showing genes within the region

and C: Copy number variants identified in normal individuals (red deletion, blue gain, brown combined deletion and gain). Genes KCNJ2 (associated with ATS) and PRKAR1A (associated with Carney complex) are highlighted. D: Deletion in present case, and deletions reported by Blyth et al. and Vergult et al.

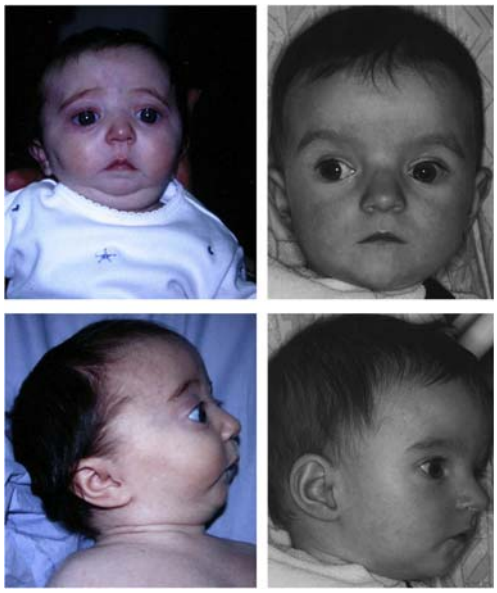

Fig. 2. Facial appearance of our patient (left) and patient described by Blyth et al. (right) at six and five months, respectively

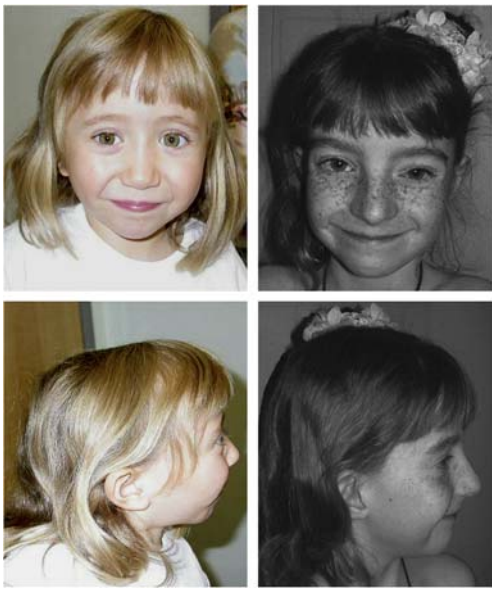

Fig. 3. Appearance of our patient (left) and patient described by Blyth et al. (right) in childhood