Interesting case report ECARUCA (2014-12)

2q23.1 microdeletion of the MBD5 gene in a female with seizures, developmental delay and distinct dysmorphic features

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http://www.sciencedirect.com/science/article/pii/S1769721212001401

Abstract:
We report a 2-year-old female who initially presented with seizures, developmental delay and
dysmorphic features and was found to have a 0.3 Mb deletion at chromosome 2q23.1 encompassing the critical seizure gene, MBD5. Her distinct physical features include bifrontal narrowing with brachycephaly, low anterior hairline, hypotonic facial features with short upturned nose, flat nasal bridge, hypertelorism, tented upper lip with everted lower lip, downturned corners of her mouth, and relatively coarse facial features including thickened tongue. She also had a short neck, brachytelephalangy, clinodactyly, and hypertrichosis. At 3½ years she developed progressive ataxia and lost vocabulary at the age of 4. Regression has been reported in one other case of MBD5 deletion. MBD5 is a member of the methyl binding gene family and appears to be responsible for regulating DNA methylation in the central nervous system. Our patient was entirely deleted for the MBD5 gene with partial loss of the EPC2 gene, which suggests that haploinsufficiency of MBD5 is responsible for the distinct phenotype observed. This supports the hypothesis that MBD5 is indeed the critical gene implicated for the findings seen in patients with deletions of chromosome 2q23.1. Further studies are necessary to delineate the role that the MBD5 gene plays in the development of the brain and these specific physical characteristics.

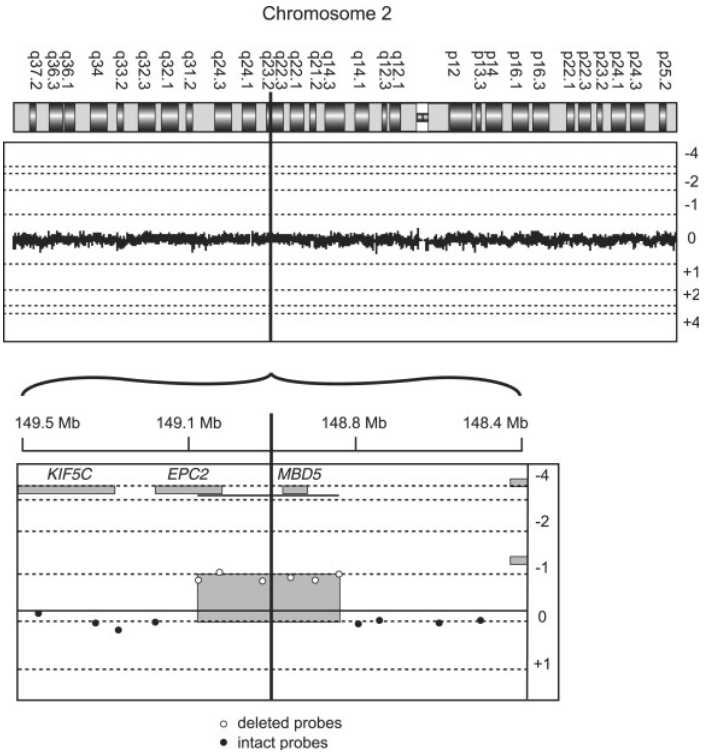

Fig. 1. 0.3 Mb deletion within 2q23.1. Magnified view of deleted region involving 6 oligonucleotides and base pair coordinates 148,867,234–149,172,531 (Human Genome build: hg18), resulting in loss of one copy of the MBD5 gene (OMIM 611472) and partial loss of the EPC2 gene (OMIM 611000).

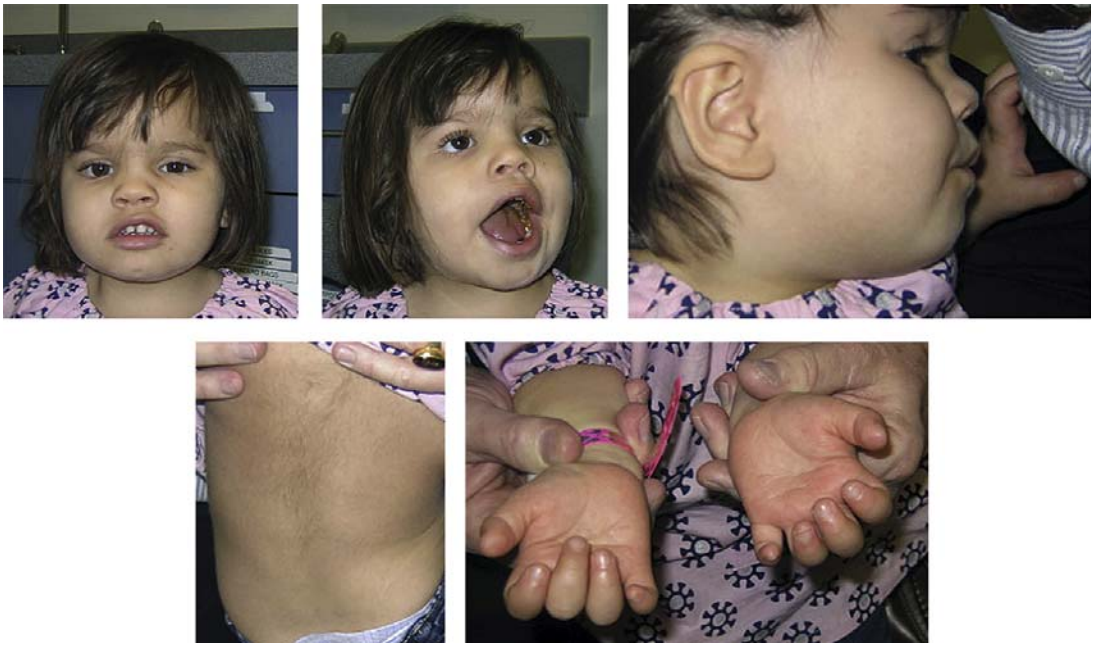

Fig. 2. Frontal and profile view demonstrating square shaped face, coarse facial features including thick tongue, hypertelorism, flat midface, upturned nose, Darwinian tubercle and short neck at 2.5 years of age. Our patient also demonstrated brachytelephalangy and hypertrichosis.