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Copy number variations and Epilepsy

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Copy Number Variants (CNV) play a key role in genetic determinism of many neurodevelopmental disorders. Several studies have shown the involvement of chromosomal aberrations in genetic epilepsies, which could be identified by new technologies such as Comparative Genomic Hybridization DNA microarray (array-CGH).

This study aimed to describe an Algerian family with epileptic phenotype and other genetic disorders, and to seek for CNV associated with epilepsy, using high-resolution array-CGH.

This family includes two individuals with juvenile myoclonic epilepsy, an individual with abnormal movements and another with intellectual disability (ID) and dysmorphic syndrome. In this family, six different CNVs were identified : 20q13.13 deletion Xp11.22 duplication, deletion 8p23.1, 14q13.2 deletion, 13q12.13 deletion and duplication 22q11.21. This latter was identified in an individual with intellectual disability, dysmorphic syndrome. This phenotype is perfectly correlated to structural chromosomal abnormality identified by array CGH.

Biography

Amina Chentouf has completed her MD in Neurology (2006) at Oran University of Medicine. She is the Head of Epilepsy Unit at Oran University Hospital in Algeria since 2014; and MD PhD in Epilepsy Genetics since April 2016. She is a Reviewer and Editorial Board Member in many journals. She had many publications in international peer reviewed journals. Currently, she leads research projects in the field of Genetics of Epilepsy. She is also interested in Epidemiology of Epilepsy.

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