Atypical benign partial epilepsy and a new variant of SLC35A3 gene plus 2p25.1 duplication. Phenotypic-Genotypic correlation?

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Abstract

Background Atypical Benign Partial Epilepsy (ABPE), recognized also as pseudo-Lennox syndrome, is an uncommon form of epilepsy characterized by generalized minor seizures such as atonic, absences, or myoclonic seizures, and electroencephalographic pattern of focal or multifocal sharp waves with activation of epileptiform discharges during sleep. ABPE is indicated as a variant of ESES (ILAE classification 2017). ESES is a clinical entity that is characterized by encephalopathy with cognitive/behavioral regression and EEG pattern of electrical status epilepticus during slow sleep. Fine and gross motor, language and social/behavioral impairment are associated symptoms, which may have reversible or persistent course. ABPE has been ascribed to the group of the “epilepsy aphasia spectrum” disorders, which includes also Rolandic Epilepsy, Landau-Kleffner syndrome, and electrical status epilepticus during sleep/continuous spike-wave during sleep. We report a young boy with a previous mild motor and language delay, who at 2-year-old presented with recurrent atonic seizures and an EEG pattern consisting of continuous spike and waves during sleep. **Methods** Next Generation Sequencing and microarray technology were used to investigate the molecular background of the proband, and the findings were then analyzed and integrated with clinical data. **Results** The child has been followed up to 7 years of age showing a progressive, complete EEG resolution and a normalization of the previous motor and cognitive impairment in association to the levetiracetam treatment. Genetic diagnosis displayed a novel heterozygous mutation c.310G>A of the SLC35A3 gene and a partial duplication of the short arm of chromosome 2, which might have pathogenic correlation with the neurological signs presented by the child. **Conclusions** Data generated by a genomic approach disclose a more comprehensive view of the genotype-phenotype correlation analysis for the novel pathogenic variant and ABPE. The relationship between the phenotypic manifestations of the child and genetic data is discussed.

Full Text

Due to technical limitations, full-text HTML conversion of this manuscript could not be completed. However, the manuscript can be downloaded and accessed as a PDF.

Figures

Figure 1

EEG at 28 months. To note, the continuous spike and waves during the slow sleep CSWS type.

Figure 2

Atonic crisis was registered during EEG record. Spikes and wave prevalent in the fronto-centro-temporal areas during the atonic crisis.

Figure 3

EEG after ethosuximide and levetiracetam with noticeable improvement of the EEG. Isolated ft-onto-central spikes remain.