A Case of CDKL5 Mutation-Associated Epileptic Encephalopathy

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INTRODUCTION

CDKL5 (Cyclin-dependent kinase-like 5) mutation causes a clinical syndrome called 'Developmental and epileptic encephalopathy 2', which is characterized by treatment-resistant seizures, psychomotor developmental delay, cortical visual impairment, dystonic movements and Rett Syndrome-like stereotypical hand movements starting in the first year of life. is happening.

In this article, we present a female patient who started having seizures at 4 months of age, had psychomotor developmental delay and stereotypical movements, diffuse spasticity, and normal brain MRI. Whole exome sequencing (WES) identified a heterozygous mutation in the CDKL5 gene.

CASE

HISTORY: The patient's seizures started in the form of contractions and jerks when she was 4 months old. She was diagnosed West syndrome and received 5 doses of ACTH therapy. Carbamazepine and topiramate were added to her treatment because of the continuation of the seizure. Various treatments such as clonazepam, oxcarbazepine, vigabatrin, and phenytoin were tried, but seizure control could not be achieved. The patient is still on valproic acid, lamotrigine and clobazam treatments.

BACKROUND & FAMILY HISTORY: Parental consanguinity + (cousins)

PHYSICAL EXAMINATION: Head circumference 53.3 cm (normal), orientation, cooperation reduced for age, no eye contact, object tracking and speech, stereotypical movements, axial hypotonia, DTR normoactive, can walk 2-3 steps with support. (Figure 1.)

LABORATORY

- Cranial magnetic resonance imaging (MRI): Normal (12 years and 14 years).
- Electroencephalography (EEG): Occasionally generalized, high-amplitude spike-wave discharges in the bilateral fronto-temporal region (16 years). (Figure 2.)
- Chromosome analysis: 46, XX.
- Microarray analysis: Normal.
- Whole exome sequencing (WES): Heterozygous c.404-1G>T mutation (XLD) in the CDKL5 (NM_001323289.2) gene.

Figure 1.

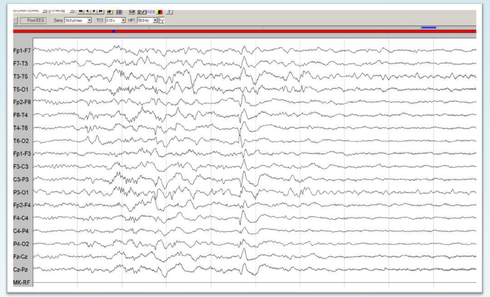

Figure 2.

CONCLUSIONS

The CDKL5 (Cyclin-dependent kinase-like 5) gene is located on the X chromosome (Xp22.13) and its mutations are inherited as an X-linked dominant. It causes a clinical syndrome called 'Developmental and epileptic encephalopathy 2', which is characterized by treatment-resistant seizures, psychomotor developmental delay, cortical visual impairment, dystonic movements and Rett Syndrome-like stereotypical hand movements that start in the first year of life. The vast majority of patients are girls. Cerebral, cerebellar atrophy, and corpus callosum abnormalities can be seen on crainal MRI. Although CDKL5 mutation is rare, it should be considered in the differential diagnosis of patients with psychomotor developmental delay and stereotypical hand movements.

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