CDKL-5 Encephalopathy in an Indian Girl: Partial Response to the Modified Atkins Diet

Dear Sir,

CDKL5-epileptic encephalopathy (EE) is an infantile-onset, X-linked catastrophic epilepsy, with intractable seizures accompanied by severe developmental delay, deceleration of head growth, and autistic traits in some patients. We describe a 3-years-old female child with CDKL5-EE who showed partial response to the modified Atkins diet.

This child presented at 2 years of age with a history of global developmental delay and seizures since 2 months of age. The perinatal period was uneventful. Seizures were generalized tonic and hypomotor, multiple times a day. She had failed phenobarbitone, phenytoin, valproate, carbamazepine, levetiracetam, topiramate, clobazam, and zonisamide. She had also failed therapeutic trials of pyridoxal phosphate, folic acid, and biotin. She had not achieved social smile, head control, grasping, or vocalization. She also had poor eye contact and had stereotypies, microcephaly, and central hypotonia.

Investigations revealed a normal brain magnetic resonance imaging. The electroencephalogram showed multifocal spike-wave discharges. The metabolic testing (blood ammonia, arterial lactate, blood acylcarnitine profile, urinary organic acids) revealed no abnormalities. There was no cerebrospinal fluid hypoglycorrhachia.

In view of the clinical profile, a possibility of CDKL5 mutation was considered. Genetic testing for CDKL-5 mutations was performed as previously described.[1] All coding exons of CDKL5-gene were polymerase chain reaction amplified from genomic DNA and cycled sequenced in both forward and reverse directions. Mutation analysis revealed a novel heterozygous c.529T>C nucleotide substitution leading to an amino acid change (p.Tyr177His), confirming the diagnosis of a CDKL5-encephalopathy. This mutation was not reported either in the HGMD professional 2016.1 database (Biobase, Qiagen, Hilden, Germany) or in the ExAC dataset (http://exac.broadinstitute.org).

As the child had failed multiple antiepileptic drugs (AEDs), she was started on the modified Atkins diet with carbohydrates limited to 10 g/day. There was no calorie, protein, or fluid restriction. Fats were actively encouraged. Within 1 week of starting the diet, there was a 60% reduction in seizure frequency (moderate urinary ketosis) and some improvement in alertness as well. At 1 year on the diet, the seizure frequency has significantly reduced from several times a day to 2–3 per week. However, there has been no improvement in autistic features or developmental milestones. There have been no significant side effects and she has tolerated the diet well.

Recently, Müller et al. reported the results of a retrospective evaluation of low long-term efficacy of AEDs and ketogenic diet (KD) in 39 patients with CDKL5-EE.[2] In the study, the authors found an overall low response to both AEDs and KD in the long-term. Only 2/12 patients had a significant seizures reduction during KD for more than 6 months and one of them...
Dear Sir,

Although firearm injuries are rare among penetrating traumas in children, they are very important in terms of their complications. Many thoracic complications may arise during spinal trauma caused by gunshot wounds. Cerebrospinal fluid (CSF) leak is one of the serious complications of gunshot wounds. Here, we present a 10-year-old girl admitted to our Pediatric Intensive Care Unit with spinal trauma after being shot on the shoulder. She was admitted in critical condition. The patient's Glasgow Coma Score was six with hemiplegia in her legs. She had anisocoria and reduced lung sounds on the left side of the chest. She was intubated and stabilized in Pediatric Intensive Care Unit. Chest tube inserted because of pneumothorax and hemothorax in the left lung. The entry hole of the bullet was in the left shoulder, and the exit hole was in the interscapular region. In computerized tomography of the chest, there was partial fracture in T2, T3, and T4 vertebrae and almost complete transection for more than 1 year. The modified Atkins diet, a less restrictive alternative to KD, may be a more feasible option for resource-constraint settings. This report highlights the potential benefit of the modified Atkins diet in some children with refractory seizures, secondary to CDKL5-encephalopathy.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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