Pyridoxine-Dependent Epilepsy. Importance of EEG findings as predictors of response to treatment

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CLINICAL IMAGE

In the last 10 years the study of epileptic encephalopathies has received considerable attention, especially those of genetic origin [1]. The purpose of this study is to show the importance of electroencephalographic findings in a case of pyridoxine-dependent epilepsy for both diagnosis and evaluate treatment response. We present the case of a newborn girl who manifested epileptic seizures since birth with poor response to antiepileptic drugs. Within a few hours of life she started with tonic attacks of the left arm, compatible with asymmetrical spasms, which are accompanied by automatisms of opening and closing of the mouth. The patient presented hypertonia of both upper limbs as well as clonic rhythmic movements of lower limbs, of approximately 10 seconds duration. An initial EEG showed disorganized basal brain activity as well as frequent epileptiform multifocal anomalies (Figure 1). Pipecolic acid in blood and cerebrospinal fluid was elevated. A cerebral MRI study was performed with epilepsy protocol, with tractography and single-voxel spectroscopy that showed: Signs of generalized atrophy in both cerebral hemispheres, corpus callosum and cortico-spinal tract in addition to reduction of n-Acetylaspartate. Phenobarbital treatment was initiated without being able to control epileptic seizures. Based on EEG findings treatment with pyridoxine was initiated. In the control EEG after initiation of pyridoxine, a significant improvement in brain activity was observed. At 48 hours after the onset of pyridoxine, the patient improved from the seizures and the level of consciousness. Currently epileptic seizures have virtually disappeared. We are waiting for the genetic study, hoping to find the existence of mutations in the
Pyridoxine-dependent epilepsy is a hereditary autosomal recessive disorder characterized by epileptic seizures that respond poorly to traditional antiepileptic drugs. However, they do respond favorably to pyridoxine. This disease appears early after birth or during the stage of childhood. Intellectual disability is a common symptom that these patients usually present [2]. Recently, high urinary excretion of α-aminoadipic semialdehyde, which is a reliable biomarker of this disorder, has been demonstrated and mutations have been demonstrated in the ALDH7A1 gene, which encodes α-aminoadipic semialdehyde dehydrogenase, in the vast majority of Patients with PDE [3]. The EEG features are not specific for this entity, though clinical and corresponding EEG improvement with pyridoxine is key (Figure 2). It is important to include in the differential diagnosis pyridoxal phosphate dependent epilepsy, which is a distinct but related entity. The early consideration of a pyridoxine therapy remains the most important in a newborn or in a child with refractory early onset seizures [4]. Actually, there are few existing studies that highlight the importance of EEG in assessing response to treatment.

REFERENCES