A rare metabolic disease causing infantile spasm: Argininosuccinate lyase deficiency

Nursan Yeniay Sut, Miraç Yıldırım, Ömer Bektaş, Ayşe Tuğba Kartal, Serap Teber
Department of Pediatric Neurology, Ankara University Faculty of Medicine, Ankara, Turkey

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Argininosuccinate lyase deficiency(ASLD) is the most common urea cycle defect after ornithine carbamoyl deficiency. The incidence of ASLD is 1:70000 live births. It usually presents with neonatal onset hyperammonemic coma, and seizure. Various seizure types such as tonic-clonic seizures, absence seizures, myoclonic jerks and atonic seizures can be observed.

OBJECTIVE

Argininosuccinate lyase deficiency (ASLD) is an urea cycle disorder that can cause refractory epilepsy. We aimed to present a patient who was diagnosed with ASLD in the neonatal period, had epileptic spasms at the age of 5 months and was safely treated with adrenocorticotropic hormone (ACTH).

CASE REPORT

A 3-day-old boy was admitted to our emergency outpatient clinic due to lethargy, seizure, and feeding difficulties. He was born after uneventful pregnancy and delivery, with non-consanguineous marriage of his parents. His neurological examination was normal except for altered consciousness and hypotonia. Biochemical tests were normal except for hyperammonemia. Plasma amino acid analysis revealed a high level of citrulline. Next generation sequencing identified a homozygous mutation of c.1251C>A (p.Ser417Arg) in the ASL gene. He was treated with phenobarbital and levetiracetam for focal clonic seizures in the neonatal period. However, epileptic spasms started when the patient had an infection at the age of 5 months. Electroencephalogram (EEG) revealed a hypsarrhythmia pattern. Additional antiseizure medications such as topiramate and vigabatrin were started. Despite receiving appropriate doses of antiseizure medications, there was no improvement in the number and duration of seizures. Finally, ACTH was administered and the seizures ceased immediately. No side effects related to ACTH were occurred except mild bradycardia.

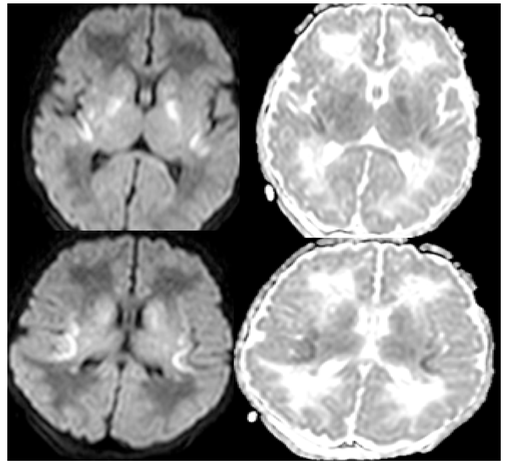

Figure 1. Diffusion restriction in perirolandic, periinsular cortices, and globus pallidus

CONCULSIONS

To our knowledge, this case is the first report describing the epileptic spasm in a child with ASLD. We also observed significant improvement in seizures with ACTH. We suggest that ACTH can be used safely in patients with ASLD.

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