Case Report

Brain MR imaging in acute hyperammononemic: Case report✩,✩✩

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ABSTRACT

Acute hyperammononemic encephalopathy is rare and generally is not widely known; only a few pediatric cases were found in the literature. These lesions’ clinical presentation differs significantly so they can mimic other lesions. In this case report, we discuss a 5-year-old boy who presented with generalized seizures and was unconscious in an apyretic context, for which she had a cranial computed tomographic and magnetic resonance imaging, both objectified an acute hyperammononemic encephalopathy resulting from an enzyme deficiency. Magnetic resonance imaging revealed lesions throughout the cortex, with the periolandic and occipital cortices spared. This distribution of cerebral signal abnormalities on magnetic resonance imaging with an abrupt and profound neurological disorder is secondary to hyperammononemic. The knowledge of the magnetic resonance imaging results of this entity is essential to accelerate the diagnosis, and treatment, also to prevent sequelae.

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Background

To prevent the accumulation of toxic nitrogenous metabolites in the body, the urea cycle absorbs excess nitrogen into urea, a water-soluble waste product [1]. The accumulation of precursors of urea, primary ammonium, causes neurology, which is defined by signs and symptoms of encephalopathy. Progressive lethargy, hypothermia, and dyspnea are the most typical symptoms, which are all linked to a very high plasma ammonium level.

Symptoms such as vomiting, altered mental status, ataxia, seizures, and developmental delay, as seen in our case, are possible.

The magnetic resonance imaging (MRI) finding of the brain diseases is an abnormal signal of cortical with sparing of the periolandic and occipital cortices [2].

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We describe our patient’s brain MRI findings and discuss how they relate to clinical manifestations.

Case presentation

A 5-year-old boy with statu-ponderal delay as medical history was brought to the emergency department after doing generalized seizures and a disorder of consciousness in an apyretic context. Glasgow coma scale score on arrival in the emergency department was 9 out of 15.

In the preceding month, his mother had reported an accidental fall from stairs with normal cranial computed tomographic (CT). No medication had been prescribed.

The physical examination revealed dysmorphic facies, global developmental delay, polypnea, and oculomotor apraxia. The cerebrospinal fluid examination was normal and the CT scan revealed normal findings. The evolution after 2 days was marked by status epilepticus, drowsiness, anisocoria, tetraparesis, and sialorrhea. The control CT scan did not reveal any abnormality. MRI with cross-sectional axial of the brain revealed an extensive symmetric and bilateral cortical parenchymal abnormality of the frontotemporoparietal and insular cortices. It also showed signs of diffusion restriction on axial diffusion-weighted images (Fig. 1), hypersignal in T2 Flair-weighted sequence (Fig. 2), and no gadolinium enhancement. The signal abnormalities were sparing of the perirolandic and occipital cortices. The deep gray matter structures, white matter, and cerebellum were normal.

A urea cycle disorder was suspected after a metabolic acidosis (pH: 7.17) with an increased anion gap on blood gas, associated with increased ammonia levels (120 μmol/l) [14-53 μmol/l].

The rest of the blood and urine workup showed abnormalities in favor of diabetes insipidus (hypernatremia [158 mmol/l] [135-145 mmol/l], low urine specific gravity [1015 g/l] [1030-1050], and urinary/plasma osmolarity < 1) associated with hypokalemia (2.7 mmol/l) [3.5-5.2 mmol/l].

Renal and hepatic function, cortisol levels, and TSH were normal.

Over the next week, the patient’s health deteriorated, with rising cerebral edema, and he was certified brain dead.

Discussion

Common causes of altered consciousness are substance abuse, medication side effects, trauma, cerebrovascular disease, infections, seizures, and metabolic disturbances [2].

Hyperammonemia is a metabolic disturbance in which an excessive amount of ammonia is accumulated in the bloodstream. This usually occurs as a result of a liver disorder or failure. Other common causes include gastrointestinal bleeding, portosystemic shunt, vesico-rectal fistulas, and medications. While in children, the most common cause is a urea cycling enzyme deficiency [3].

The diagnosis is based on the presence of characteristic clinical features (abrupt neurological disorder) as well as the MRI finding of encephalopathy and hyperammonemia.

MRI findings are characterized by cerebral atrophy and bilateral, symmetric hyperintensities on T2-weighted images without corresponding signal intensities in T1-weighted images and by restricted diffusion involving cortices with sparing of the perirolandic and occipital cortices [4,5].

![Fig. 1 – Brain MRI in diffusion sequence (A) with ADC mapping (B): shows diffusion restriction with low ADC.](image-url)
Acute hyperammonemic encephalopathy is a disorder of brain development that may affect many parts of the body and may be caused by mutations in any of many genes of the urea cycle.

**Conclusion**

In this study, we described brain MRI findings in a boy-child patient with acute hyperammonemic encephalopathy due to enzymopathy. The cortex was injured, but the perirolandic and occipital cortices were spared, according to MRI. The ability to understand the MRI findings may help in the diagnosis and management of hyperammonemic encephalopathy.

**Authors’ contributions**

All the authors contributed to the conduct of this research work. The authors have read and approved the final version of the manuscript.

**Patient consent**

I confirm that the legal representative of the patient whose case is reported in this article, gave consent for publication.

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