ISSN: 2641-1911
DOI: 10.33552/ANN.2022.12.000797
Archives in Neurology & Neuroscience
Iris Publishers

Case Report

Congenital Adrenal Hyperplasia-Associated Encephalopathy: A Case Report

Faruk Incecik* and Ozlem M Herguner

Divisions of Neurology, Department of Pediatrics Çukurova University, Adana, Turkey

Corresponding author: Faruk Incecik, Divisions of Neurology, Department of Pediatrics Çukurova University, Adana, Toros mah. 78186 sok., Yeşilpark Evleri, Kat: 7, no: 13, Adana, Turkey.

Received Date: July 05, 2022

Published Date: July 26, 2022

Abstracts

Congenital Adrenal Hyperplasia is a group of autosomal recessive disorders due to deficiencies of enzymes involved in steroidogenesis. Clinical manifestations depend on the degree of cortisol and aldosterone deficiency. The salt-wasting form can present as a medical emergency with severe hyponatremic dehydration, hyperkalemia, polyuria and hyperpigmentation. However, in recent years, patients with CAH presenting with acute encephalopathy or encephalopathic episodes have been reported. We report here clinical and nueroimaging findings of a case with CAH presenting with acute encephalopathy.

Keywords: Congenital adrenal hyperplasia; Acute encephalopathy; Children

Introduction

Congenital Adrenal Hyperplasia (CAH) is a group of autosomal recessive disorders due to deficiencies of enzymes involved in steroidogenesis. The most common form is a 21-hydroxylase deficiency. Clinical manifestations depend on the degree of cortisol and aldosterone deficiency [1]. Acute encephalopathy is one of the most severe diseases of both infancy and childhood, and patients have symptoms of prolonged seizures and deep coma. Acute encephalopathy is heterogeneously caused by viral infection, drugs, and metabolic disorders [2].

In the literature, a few cases have been reported with acute encephalopathy or encephalopathic episodes in CAH patients [3,4,5]. Here, we report a case with CAH, presented with acute encephalopathy.

Case Report

A 3,5-years-old girl was admitted to our hospital because of unconscious and seizure after 2 day of fever and vomiting. The parents were consanguineous. She was born at term, and she had been diagnosed as salt-wasting 21-OHD in neonatal period and supplemented with oral hydrocortisone and fluorocortisol. On admission, she showed comatose and had focal seizures. Glasgow Coma Scale was 5 at admission. Intravenous hydrocortisone was administered but his level of consciousness did not recover. Levetiracetam treatment was started for focal seizures and her seizures did not recur. The patient regained consciousness over the next 48-72 hour.

In her laboratory examinations, serum sodium was 131 meq/L, serum potassium was 4.7 meq/L, and blood sugar level was 73 mg/ dl. Blood gases showed pH 7.29, pCO2 42.4 mmHg, HCO3 15 mmol/L, and base deficit -13.0 mEq/L. Cerebrospinal fluid (CSF) examination revealed normal. Serological tests for infections are negative. Electroencephalography showed generalized bilaterally synchronous slow waves without epileptiform discharges. Cerebral magnetic resonance imaging (MRI) scan with diffusion-weighted

images (DWI) showed diffusion restriction of the right hemisphere, predominantly in subcortical white matter (Figure 1).

Control DWI images of brain MRI after two weeks showed prominent resolution of the high intensity lesions. She exhibited mild left hemiparesis and epilepsy as sequelae.

Discussion

Congenital adrenal hyperplasia is a collective group of disorders involving the lack of an adrenal corticosteroid-synthesizing enzyme, which leads to variable degrees of excesses or shortages of glucocorticoids, mineralocorticoids, or adrenal androgens, and more than 95% of all cases of CAH are caused by 21-hydroxylase deficiency. Adrenal crisis is the most dangerous complication of CAH, but even if patients develop adrenal crisis, they should recover following the administration of additional corticosteroids and other supportive treatment [1].

Recently, the cases of CAH-related encephalopathy have been reported in literatüre [3,4,5]. Le et al [3] reported acute encephalopathy in three patients with CAH. They exhibited disturbed consciousness or seizures, which did not improve after glucocorticoid administration. Brain MRI disclosed various patterns of white matter lesions. Grunt et al [4] described two patients with classic saltwasting CAH who were treated with glucocorticoids and suffered from an acute encephalopathic crisis in their preschool age and showed focal or multifocal cortical-subcortical lesions on MRI of the brain. In another study, Abe et al [5] reported acute encephalopathic episodes in 15 patients with CAH. Fourteen patients presented with seizures and 12 patients had a refractory course. Twelve patients had deep coma for > 24 hours. Neuroimaging studies showed heterogeneous features. High signal intensity was detected in cerebral MRI (DWI) in all 15 patients in the acute period. In the chronic period, 14 patients survived, 11 of whom had some degree of neurological sequelae. In our patient had disturbance of consciousness and seizure with diffusion restriction of the right hemisphere in cerebral MRI.

Congenital adrenal hyperplasia-associated encephalopathy is a heterogeneous pathological condition. The initial symptoms of CAH-associated encephalopathy resemble those of adrenal crisis, and it is associated with prolonged coma and/or refractory seizures intractable to therapy [5]. In addition, patients often develop neurological sequelae of varying severity. Neuroimaging of patients with CAH-associated encephalopathy revealed heterogeneous findings, including abnormal high signal intensity on DWI appearing in both the cerebral cortex and subcortical or deep white matter [3,4,5].

Different pathophysiologic mechanisms might explain the cerebral lesions in patients with CAH presenting with acute encephalopathy as described in the presented cases. This indicated that the physiological instability of steroids in patients with CAH may have some potential to trigger encephalopathy, even if the patients with CAH are treated with current replacement therapy of corticosteroids. Therefore, previous results indicated that these MRI findings and clinical course were not always generated by the same pathological condition, but the fragility of underlying CAH to some stress and excitotoxicity may often be contributory to both the brain lesions and their clinical conditions in diverse mechanisms [5].

Conclusion

CAH-associated encephalopathy should be considered when there is resistance to standard therapy for adrenal crisis, the presence of persistent seizures, and prolonged deep coma.

Conflict of Interest

No conflict of interest noted by the authors.

Acknowledgment

None.

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