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Paediatrics

A RARE PRESENTATION OF ADDISON DISEASE IN A PATIENT WITH CEREBRAL PALSY-A CASE REPORT

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ABSTRACT Addison's disease although rare, it is a well-recognized endocrine disorder characterized by adrenal insufficiency. It typically presents with a specific set of signs and symptoms, including weakness, weight loss, hyperpigmentation, and electrolyte imbalances. However, it can occasionally manifest atypically, making its diagnosis challenging. Here, we report an unprecedented case of Addison's disease in a 16-year-old male with cerebral palsy, mental retardation, and a history of seizure disorder. His presentation included generalized weakness, vomiting, hyponatremia, and hypotension, which initially posed a diagnostic conundrum. The detailed evaluation revealed primary adrenal insufficiency and appropriate treatment with hydrocortisone and fludrocortisone resulted in a significant improvement in his clinical condition. This unusual presentation underscores the importance of considering Addison's disease even in patients with complex medical histories and unusual clinical pictures, as timely diagnosis and intervention can be life-saving. This case report emphasizes the need for healthcare providers to remain vigilant for rare and atypical presentations of well-known conditions like Addison's disease.

KEYWORDS : Addison's disease, cerebral palsy, atypical presentation, adrenal insufficiency, hypovolemic shock, hyponatremia.

INTRODUCTION

Endocrine disorders often go unnoticed during their initial stages, particularly in developing nations where healthcare professionals might have limited awareness and access to specialized diagnostic tools may be restricted. [1] Given the low frequency of these disorders and their tendency to manifest atypically, it becomes crucial for healthcare providers to familiarize themselves with the diverse ways in which these conditions can present. [2,3]

Addison's disease or primary adrenal insufficiency, is a rare endocrine disorder characterized by the inadequate production of adrenal hormones, particularly cortisol and aldosterone, due to dysfunction of the adrenal glands. Its clinical presentation can vary, but common features include weakness, fatigue, weight loss, and hyperpigmentation of the skin and mucous membranes. [4,5]

Cerebral palsy (CP) on the other hand, is a non-progressive neurological disorder originating in early childhood and affecting muscle coordination and body movement. It is primarily a motor disorder, and while it may have associated comorbidities, the relationship between cerebral palsy and Addison's disease is exceedingly rare and poorly documented. [6]

This case report highlights the unique clinical encounter of a patient with cerebral palsy who presented with an unusual manifestation of primary adrenal insufficiency. The coexistence of these two conditions in a single individual is exceptionally rare and poses several intriguing questions. Understanding the complex interplay between these disorders and recognizing the distinct presentation of Addison's disease in the context of cerebral palsy is essential for timely diagnosis and effective management. This case underscores the importance of considering unusual clinical presentations in patients with underlying neurological conditions and the value of multidisciplinary collaboration in the diagnosis and care of complex medical cases.

Case presentation

Somnath, a 16-year-old male, was admitted to our hospital with a complex set of symptoms and a history of developmental delay and seizure disorder. The history of his current illness began 15 days prior to admission when he started experiencing generalized weakness and multiple episodes of non-bilious vomiting, with vomitus containing food particles. His appetite decreased significantly during this period, but there were no reported instances of loose stools, fever, abdominal pain, or loss of consciousness.

Upon examination, the patient was conscious but irritable, displaying generalized muscle wasting, hyperpigmentation, and contractures. His vital signs were indicative of hypovolemic shock, with blood pressure as low as 80/40 mmHg. He was immediately transferred to the Pediatric Intensive Care Unit (PICU), where he received intravenous fluid boluses to stabilize his blood pressure. After three boluses, his blood pressure and peripheral pulses improved, and he was transitioned to maintenance fluid.

Further systemic examination revealed hypertonia, exaggerated upper and lower limb reflexes, and the absence of elicitable plantar reflexes. However, there were no signs of meningeal irritation or cerebellar dysfunction. Blood investigations showed normocytic normochromic neutrophilic leukocytosis, hyponatremia (Na 120), hyperkalemia (K 5.3), and a low serum chloride level (Cl 80). The diagnosis pointed toward adrenal insufficiency, so slow sodium correction was initiated with 3% NaCl over 48 hours.

However, after 48 hours, sodium levels remained low, and further investigations were conducted to explore the adrenal pathology. Urine osmolality was below the normal range, and urine sodium was elevated, suggesting a possible renal etiology. Adrenal gland imaging via ultrasound showed that both adrenal glands were of normal size. Serum cortisol levels were found to be abnormally low (20.89 ng/ml) while serum ACTH levels were significantly elevated (1596 pg/ml). This raised strong suspicion of primary adrenal insufficiency, and the patient was promptly started on hydrocortisone treatment (5 mg/tab, 1 tab TID).

During the course of treatment, the patient's sodium levels gradually improved, and his vital signs stabilized. His medical history included a diagnosis of global developmental delay (GDD) with mental retardation and a seizure disorder. Notably, he had a history of seizures beginning at the age of 4 years and was diagnosed with a form of epilepsy that resembled juvenile myoclonic epilepsy. He had been on medication (Valproate 300 mg OD) for this condition.

Regarding birth history, he was born full-term via normal vaginal delivery and had a birth weight of 2 kg. He required admission to the Neonatal Intensive Care Unit (NICU) for 5 days due to feeding difficulties. His developmental history revealed that he achieved developmental milestones with mild delays up to the age of 4 years, at which point he experienced a regression of milestones.

As for his current developmental status, the patient displayed the ability to walk with support and engage in scribbling activities. In terms of language development, he had reached the stage of bi-syllabic speech and could ask for food. The comprehensive clinical evaluation of the patient's complex medical condition is ongoing, with a focus on understanding the interplay of his underlying conditions and his recent presentation with adrenal insufficiency.

DISCUSSION

Addison's disease is reported to have an incidence of 0.8 per million

and a prevalence ranging from 40 to 110 per million in the United States and various European nations. [2] Unfortunately, there is a lack of data regarding its incidence and prevalence in India. The diagnosis of Addison's disease is often delayed due to a failure to suspect it, primarily because the signs and symptoms can be subtle in many cases. Additionally, delays in seeking expert medical care contribute to this diagnostic lag. [1]

Typical initial symptoms of Addison's disease include weakness and weight loss, observed in more than 90% of cases, gastrointestinal complaints in over 80%, body aches in about 18% of cases, and symptoms such as salt cravings, syncope, and disorientation are present in approximately 12-15% of cases. Common clinical signs include hyperpigmentation observed in 94% of patients, and hypotension affecting 90% of those with the condition. Frequently noted laboratory findings comprise electrolyte imbalances (92%), hyponatremia (88%), hyperkalemia (64%), hypercalcemia (6%), and anemia (40%). [2,7]

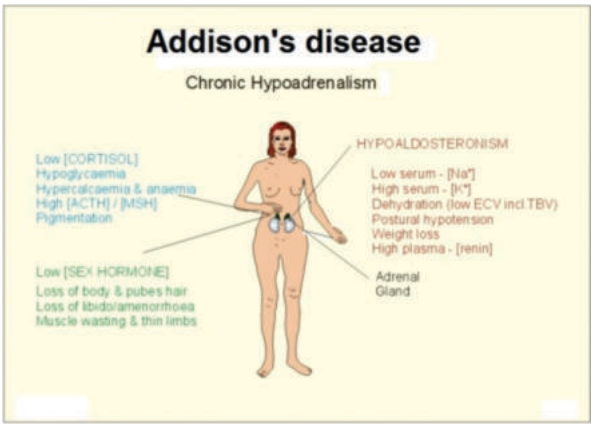

Figure 1 signs and symptoms

The case of a 16-year-old male with cerebral palsy, mental retardation, seizure disorder, and an atypical presentation of primary adrenal insufficiency (Addison's disease) presents a diagnostic challenge due to the coexistence of multiple medical conditions, complex clinical features and a rare mode of presentation.

The patient's initial presentation with generalized weakness, severe vomiting, hypovolemic shock and pronounced hyponatremia was alarming. The presence of cerebral palsy and mental retardation added layers of complexity to the diagnostic process. Considering the patient's medical history which included a past diagnosis of juvenile myoclonic epilepsy and the use of Valproate broadened the scope of potential etiologies for his current symptoms.

The patient's persistent hyponatremia, low normal serum cortisol levels, elevated serum ACTH levels and inadequate cortisol response to ACTH stimulation led to the diagnosis of primary adrenal insufficiency. This rare presentation of Addison's disease underscores the importance of considering endocrine disorders in complex clinical scenarios even in patients with pre-existing medical conditions.

Swift intervention and the initiation of hydrocortisone and fludrocortisone replacement therapy resulted in rapid clinical improvement. The patient's response to treatment as evidenced by corrected hyponatremia, normalization of electrolyte levels and blood pressure stabilization supports the accuracy of the diagnosis and the importance of prompt management.

In this case, the presence of hyperpigmentation and hypotension initially raised suspicion regarding the possibility of Addison's disease. Routine tests including electrolyte measurements further supported this diagnostic hypothesis. Specifically, hyponatremia was detected and correcting this electrolyte imbalance led to the alleviation of symptoms. His electrolyte levels and blood pressure continued to improve with the initiation of specific treatment. Such a presentation occurring in a patient with cerebral palsy is remarkably unusual for cases of Addison's disease and has not been previously documented. This case underscores the importance of healthcare providers being attentive to such atypical presentations and recognizing that basic investigations like assessment of electrolyte imbalances can offer crucial diagnostic insights.

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