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More than a decade of misdiagnosis of alternating hemiplegia of childhood with catastrophic outcome: A case report

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Alternating hemiplegia of childhood (AHC) is a distinct clinical disorder characterized by recurrent episodes of hemiplegia, abnormal ocular movement, and progressive developmental delay. It is an extremely rare genetic disorder related to ATP1A3 gene mutations with an estimated prevalence of 1/1,000,000 children. It is believed that this number could be an underestimate due to variability in clinical presentation, lack of knowledge about the disease, and lack of advancement in the diagnostic laboratory and radiologic test that will confirm the diagnosis. A thorough literature search yielded only one case study reported from Saudi Arabia. In this paper, we present a case of AHC in which the diagnosis was missed for many years until severe hypoxic brain insult occurred from prolonged status epilepticus. We are not only presenting an interesting clinical entity and radiological images, but we are also shedding the light on a rare genetic disease with catastrophic sequelae. Since the original description of AHC, many endeavors have been made to understand the pathophysiology of the disease which resulted in linking the disease with mutations in the gene ATP1A3. Despite this substantial progress in the understanding of the disease, no curative treatment has been discovered, and the disease continues to be challenging to treat. All the current treatments are focused on reducing the frequency, duration, and severity of AHC episodes. The challenges in diagnosis and treatment lead to a poor outcome as seen in our case. Early recognition and accurate diagnosis of the disease with the suitable treatment may lead to improved outcome. Referral to a center with expertise in genetic disorders and access to genetic labs is of paramount importance in the diagnosis of this disease. The complexity and severity of this disorder make more research crucial to find the curative therapy and further understand the disease.

Biography

Hussein Algahtani is the Associate Dean of clinical affairs and the Head of the simulation center in the College of Medicine at King Saud bin Abdulaziz University for Health Sciences in Jeddah, Saudi Arabia. He is also an Assistant Professor in Neurology and the neurosciences block coordinator. In addition, he is the Neurology section Head and the Head of Neurophysiology laboratory at King Abdulaziz medical city in Jeddah, Saudi Arabia. He is a well-known researcher with more than 50 publications in the literature.

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