Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Medical Science

Ohtahara Syndrome: Case Report and Literature Review

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DOI: 10.36347/sjmcr.2023.v11i04.046 | **Received:** 04.03.2023 | **Accepted:** 17.04.2023 | **Published:** 21.04.2023

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Abstract | **Case Report**

Ohtahara syndrome or Early Infantile Epileptic Encephalopathiy (EIEE) is a rare form of neonatal epileptic encephalopathy. It has particular electro-clinical, therapeutic and outcome features. Its etiologies remains obscure and are essentially secondary to a congenital or acquired structural malformation of cortical development. We report a case of a term newborn presented with seizures on week two of live. Seizures were intractable. Metabolic workup for seizures was normal. Brain MRI did not reveal any structural abnormality. Electroencephalogram showed suppression pattern suggestive of ohtahara syndrome.

Keywords: Ohtahara syndrome, Seizures, Suppression burst.

INTRODUCTION

Ohtahara syndrome is a rare form of epilepsy characterized by seizures and developmental delays that usually occur within the first three months of live (most often within the first 10 days). Some cases are caused by a brain abnormality, metabolic disorder, or gene mutation, while other cases have no known cause. We report a case diagnosed in the department of pediatrics in Sidi Bernoussi CNSS Polyclinic.

CASE REPORT

The patient was delivered by caesarean section at 38 week of gestation to a healthy 29 year-old G2P2 mother. Antenatal scans showed no abnormalities and the mother had no medical history and there was no personal or family history of seizures. The baby weighted 3150 grams at birth and immediately cried after delivery. Breastfeeding was initiated. On day twelve of live, baby was noted to have tonic seizures. He was admitted to our unit for management. Blood glucose, calcium and magnesium levels were normal. Lumber puncture and sepsis screen showed no abnormalities. MRI Brain did not reveal any structural abnormality (Figure 1).

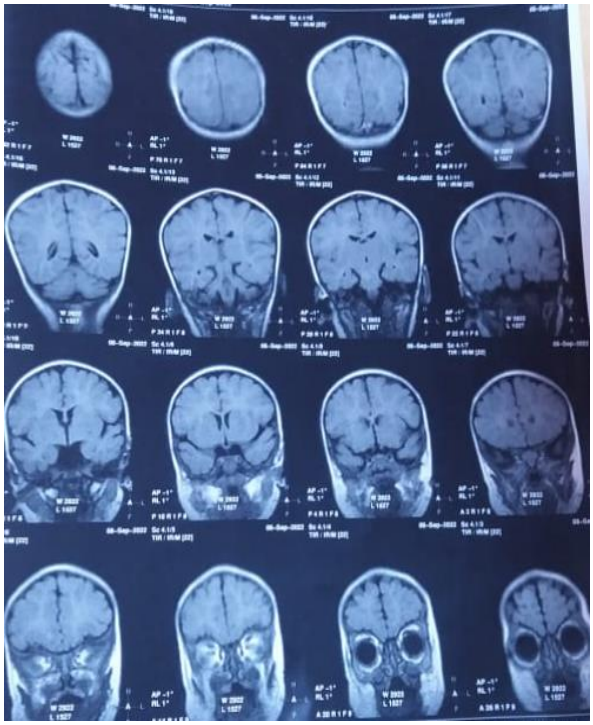

Figure 1: MRI brain of the patient showing no anomaly

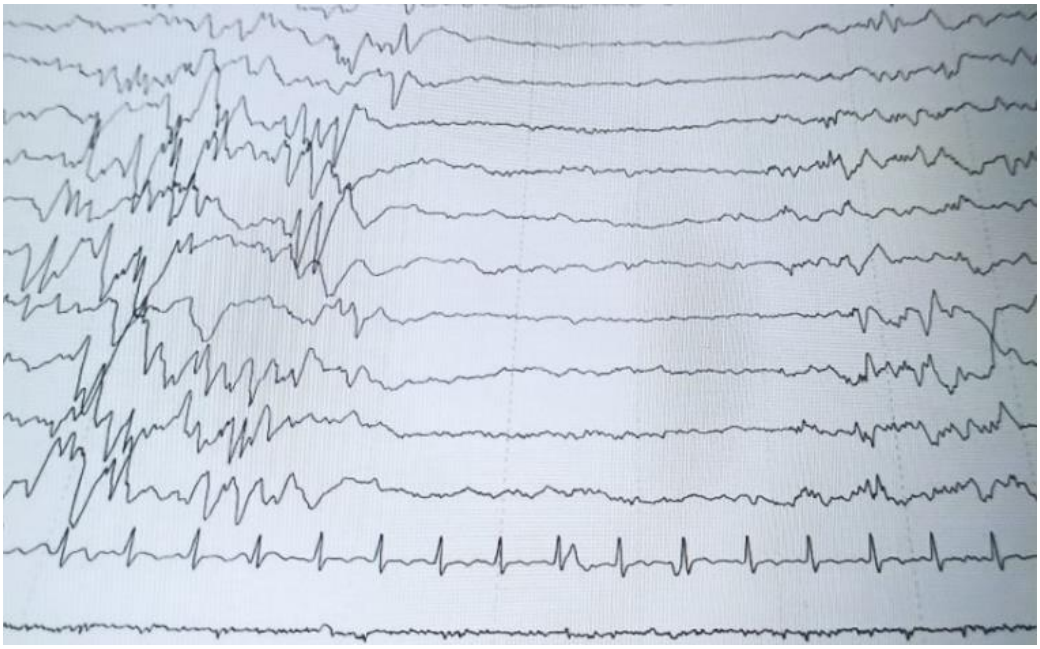

Figure 2: EEG OF the newborn showing suppression burst pattern

The neonate was managed with maintenance Phenobarbital and vigabatrin, but intermittent seizures persisted until topiramate was finally effective in controlling them. Neurological examination at discharge revealed abnormal findings, including axial and appendicular hypotonia, and early intervention therapy was initiated. Follow-up at 9 months showed a marked reduction in the frequency of seizures and the baby continued topiramate and phenobarbital.

DISCUSSION

Ohtahara syndrome, also known as early infantile epileptic encephalopathy with burst suppression, is a complex and debilitating condition that manifests in infancy. Named after the pioneering work of Dr. Shunsuke Ohtahara, who first described it in 1976, this syndrome poses significant challenges for affected infants and their families [1].

The exact cause of this syndrome remains elusive, although it is believed to be primarily driven by genetic mutations. While most cases of Ohtahara syndrome occur sporadically, meaning they arise randomly and are not inherited, specific genetic mutations, such as those in the ARX gene or other genes associated with brain development, can be implicated in some cases [2]. Ohtahara syndrome typically emerges in infancy, with seizures often presenting within the first few weeks or months of life [3].

The hallmark symptom of Ohtahara syndrome is early onset seizures, which can be frequent and diverse in type, including tonic seizures characterized by muscle stiffening, focal seizures affecting only one part of the body, and spasms involving sudden contractions of muscles. These seizures are often refractory to medications, posing significant challenges in seizure control. In addition to seizures, infants with Ohtahara syndrome may exhibit developmental delay, intellectual disability, and other neurological abnormalities such as abnormal muscle tone, feeding difficulties, and respiratory challenges [4, 5].

Diagnosing can be intricate, as it is a rare condition that shares similarities with other types of epilepsy syndromes. A thorough medical history, physical examination, and comprehensive neurological evaluation are crucial in the diagnostic process. EEG is a key diagnostic tool for Ohtahara syndrome, typically revealing a distinctive pattern known as “burst suppression” [6-8]. This pattern is characterized by bursts of high-voltage activity followed by periods of low activity on the EEG. Brain imaging studies such as magnetic resonance imaging (MRI) may also be employed to rule out other potential causes of seizures.

As of now, there is no cure for Ohtahara syndrome, and management is mainly supportive, focusing on controlling seizures and addressing associated symptoms. Antiepileptic drugs are commonly used to manage seizures, although their effectiveness may vary. Other treatment options may include ketogenic diet, vagus nerve stimulation, and other forms of neuromodulation techniques. Early intervention with physical, occupational, and speech therapies may also be beneficial in managing developmental delays and improving the quality of life for affected infants [5, 7].

The prognosis of Ohtahara syndrome is generally bleak, with most infants experiencing severe developmental delays and intellectual disability. The majority of affected infants do not achieve

developmental milestones, and the mortality rate is high, with some studies reporting mortality rates of up to 50% within the first two years of live. However, there have been rare reports of patients who have survived into adulthood with varying degrees of developmental progress [5, 9].

CONCLUSION

Ohtahara syndrome is a rare and severe form of epilepsy that presents in infancy and is characterized by early onset seizures, developmental delay, and a challenging prognosis. Despite the complexities surrounding this condition, ongoing research and advancements in medical management provide hope for improved outcomes in the future.

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