Case Report

Electroencephalographic characterization of a case of infantile spasm with atypical presentation

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ABSTRACT

Infantile spasm is one of the common seizure disorders with serious consequences. It is generally characterized by a muscle spasm of various intensity and distribution, salaam spells, and infantile myoclonic seizures with typical electroencephalography (EEG) features of hypsarrhythmia. Here, a unique case is being reported in which breath holding spells for few seconds were the sole presentation of infantile spasm. The EEG features of the case included typical high voltage spike and slow wave pattern followed by very poor suppression representing modified hypsarrhythmia, a classical feature of infantile spasm. Fast Fourier transformation of hypsarrhythmic spells confirmed the delta dominance with the highest power spectral density of delta waves. The breath holding spells being the sole presentation of infantile spasm makes it an important case clinically due to the possibility of misdiagnosis due to its inconspicuous presentation.

Key words: *Breath-holding spells, Electroencephalography, Hypsarrythmia, Infantile spasm*

Infantile spasm is a seizure disorder of childhood and probably the very first seizure described. It is a kind of catastrophic disease due to no effective treatment available till date. Classical clinical presentation of infantile spasm shows complete heaving of the head forward toward knees, and then immediately relaxing into the upright position. These bowings and relaxing spells would be repeated alternately at intervals of a few seconds, from 10 to 20 times or more at each attack [1]. As the name suggests, the onset of this disorder occurs usually in the 1st year of life with a mean age of onset at 3–5 months of age. The incidence of infantile spasm is 1 in 2000–4000 live births worldwide [2]. Although west syndrome is used synonymously with infantile spasm though, unlike infantile spasm west syndrome is a classic triad of three features, spasm, characteristic EEG findings, and developmental delay or mental retardation [3].

Characteristic clinical presentation of infantile spasm has been described as salaam spells with head nodding movement along with generalized contraction of all limbs which relaxes gradually in few seconds. It may repeat several times within a brief period and occurs in bursts of multiple episodes. Though, the common clinical presentation of an infantile spasm is a generalized or focal abnormal sudden jerky movement of trunk or limbs, an atypical presentation like only little flexion movement of the head has also been reported occasionally. A presentation like isolated breath holding spells is very rare if ever reported [4,5].

Here in the present case report, we summarize the case of a child with "breath-holding spells" being the only apparent clinical symptom with EEG features of hypsarrhythmia, which is a characteristic feature of infantile spasm and west syndrome.

CASE REPORT

A 5-month-old baby visited the pediatric outpatient department of the institute with chief complaints of breath-holding spells of 2 months duration. The spells usually occurred while crying and lasting for around 1–2 s followed by spontaneous recovery. There was no history of loss of consciousness, abnormal body movements, bluish discoloration or pallor of the body. He also had a history of a non-productive cough of 3 days duration which was not associated with tachypnea, fever, and difficulty feeding or chest retraction. Baseline general examination cardiorespiratory workout has been done by the department to rule out any cardiac or respiratory abnormality. General physical examination did not show any feature suggestive of syndromic features (including the Down's syndrome) known to be associated with early seizure disorders and infantile spasm in an infant [6] (Fig. 1). The baby was kept under prophylactic antitubercular treatment apparently because his mother was on antitubercular treatment for 6 months during pregnancy but did not complete the treatment due to subsequent deranged liver profile. The peripartum period was uneventful except for the history of decreased fetal movements at the time delivery.

He is a singleton term baby born with lower segment cesarean section having a birth weight of 2.625 kg. The child got

hospitalized 3 months back to the institute for 4–5 days with a history of jaundice. He is being breastfed besides supplemental nutrition. He is also receiving all the immunization as per the recommended schedule.

His developmental history shows that the baby attained neck holding at 3½ months, coos present, laugh loud at 4 months of age, monodextrous reach present. As per his pedigree, he is the third child born; the first sibling died at the age of 1 week after birth, second is alive, and of 3 years of age. At presentation, his weight was 8.06 kg, height was 65 cm, and body mass index was 19.07 kg/m^2. Respiratory rate was 30/min, temperature was 98° F, and pulse was recorded as 128 beats/min. No signs of pallor, icterus, cyanosis, clubbing, lymph nodes, or edema observed. On chest examination, no chest retraction was seen, bilateral air entry present, vesicular breath sound was there, no any added sound has seen. Apex impulse was localized in the 5th intercostal space, lateral to midclavicular line. Heart sound was heard normally, no murmur had been noticed as per the records. Mantoux test was performed, and it showed in the duration of 2 cm×2 cm. Complete hemogram showed a hemoglobin of 9.6 g/dl, neutrophils 12% (significantly decreased), lymphocytes 80% (significantly increased), and erythrocyte sedimentation rate 55 mm at 1st h.

Figure 1: The 5-month-old baby boy with no apparent tell-tale sign of any syndrome usually associated with infantile spasm

The electroencephalography (EEG) of the patient was done in the clinical neurophysiology department of the institute. The EEG showed generalized multifocal spikes with very high voltage (250–450 μv) delta range slow waves followed by suppression pattern. These bursts of high voltage were present for 2–3 s followed by 2–3 s of suppression period (a characteristic hypsarrhythmic pattern, classically associated with infantile spasm) (Fig. 2a-d).

The quantitative analysis of EEG findings was found to be as follows: Fast Fourier transformation (FFT) of the T3O1 and T4O2 channels (representative of background activities) of the EEG (Fig. 3a and b) shows a very high highest power spectral density (PSD) for delta waves compared to other waves including theta, alpha, and beta waves.

DISCUSSION

Infantile spasm is a catastrophic pediatric seizure disorder with a poor prognosis [1]. In general, infantile spasm presents with typical clinical features of massive spasm, salaam spells, and infantile myoclonic seizures which is categorized under the class of generalized seizure disorders. Occasionally, focal events and focal seizures have also been reported [7]. Cases of infantile spasm with the asymptomatic presentation are a rare entity.

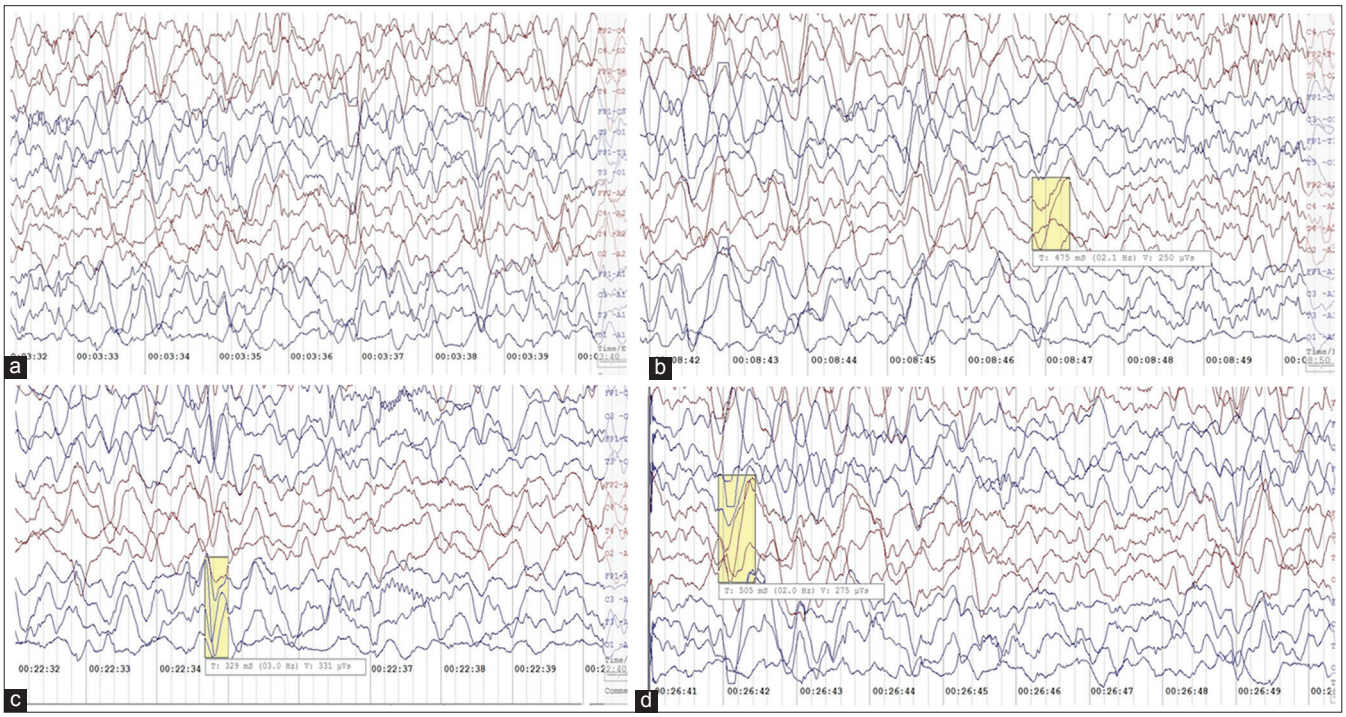

Figure 2: (a-d) The multifocal independent spike discharges in the whole of the record (primarily occipital activities, as depicted in the image) at the different time frame

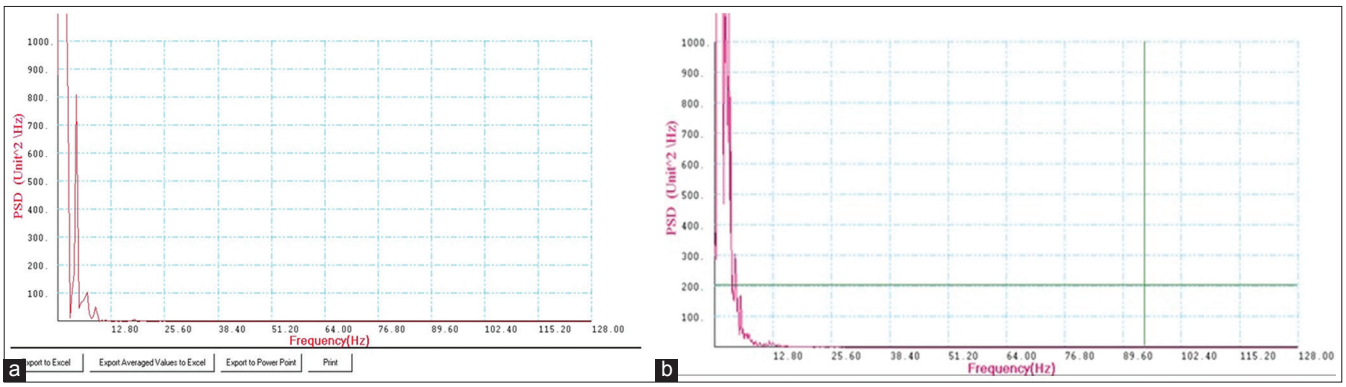

Figure 3: (a and b) Fast Fourier transformation inch. T3O1 and T4O2 depicting the highest power spectral density of delta (much more than what usually observed during usual normal delta sleep of the infants)

The present case reported here, interestingly, had apparently no symptoms related to generalized, focal or spasm type of seizure disorder at presentation and breath-holding spell was the sole presenting complaint besides short-term cough and cold.

The EEG findings showed multifocal independent spike discharges (MISD) usually designated as a modified hypsarrythmic pattern in most of the montages but very prominent in the background occipital montages in the whole of the record. The FFT of the occipital waves showed a very PSD for delta waves, much more than that found during the normal delta sleep of infants.

By definition, hypsarrhythmia is a very high-voltage, disorganized pattern of EEG abnormality. A less chaotic pattern, called modified hypsarrhythmia, is reported to be a more commonly encountered pattern than typical hypsarrhythmic pattern [8]. Hypsarrhythmia or modified hypsarrhythmia is seen in about two-thirds of cases of the infantile spasm. Other patterns, such as MISD, are present in the remainder. Although infantile spasms may be associated with other EEG abnormalities, hypsarrhythmia virtually never occurs in other epilepsy syndromes [9]. The present case shows both MISD and some typical hypsarrythmic discharges pathognomonic of infantile spasm. As for as, infantile EEG concerns delta wave voltage may reach up to 100 μv in usual circumstances. In some cases, a high voltage bursts of up to 200 μv may be noticed, especially during deep sleep, but voltage above 200 μv is never a normal finding [7]. As far as PSD is concerned, for delta waves, it is usually higher during sleep, more so in infants. In the present case, PSD of delta wave is in the range of 800–1000 unit^2/hertz which is much more than the usual PSD value expected during sleep in an infant.

Earlier, few researchers had reported cases which were devoid of typical presentations of seizure with uneventful pregnancy and birth [10,11], normal development before the onset of spasm and absence of neurological abnormalities at the onset of spasm [12] including absence of any other types of seizure before the onset of spasm, and normal laboratory, computerized tomography (CT), or magnetic resonance imaging findings at onset [13,14]. Another report on the similar patient described a case in which the patient presented with "lack of speech, frequent fits of idiotic laughter, and rolling of the head, delighted by music, and gray colors." This description leads to the suspicion that the subject probably had features of autism [15]. Later reports on infantile spasm suggest that there is a correlation between other disorders such as tuberous sclerosis, down syndrome, and genetic associations along with autistic spectrum disorders [16,17].

Hypoxic ischemic encephalopathy has been identified as one of the important predisposing factors for the development of infantile spasm. Although as per an estimate, 25% of all live births are affected with hypoxic-ischemic injury, all cases may not lead to seizure disorders. Probably this hypoxic-ischemic injury acts a second hit for the development of infantile spasm, not the sole causal factor. There must be some other factors acting as the first hit in cases of hypoxic injury leading to infantile spasm development [18,19].

Infections whether prenatal, perinatal, or postnatal have also been noted a very important factor in the development of infantile spasm. Although the exact correlation between infection and development of infantile spasm has been much clear, there are shreds of evidence supporting the role of infections as a causal factor.

The term "breathing holding" is apparently a misnomer as the breathing actually stops in expiration and is apparently an involuntary phenomenon. Although definite provoking factor for breath holding is yet to be known, breath-holding spells with associated loss of consciousness are reported in almost 5% of infants [20]. One of the postulated theories behind the phenomena is the disturbance in central autonomic regulatory features [21].

Various other factors such as metabolic syndromes, encephalopathies, developmental anomalies, and various genetic associations such as *CDKL5* and *SPTAN1* mutations have also been identified as very important identifiable causes for infantile spasm [22]. A future study with EEG based bigger database of breath-holding spells with similar presentation may through some light in the field in the larger interest of the society.

CONCLUSION

This case report could be one of the atypical and rare presentations of infantile spasm with a high chance of clinical misdiagnosis due to its inconspicuous nature of the presentation. A higher degree of suspiciousness, meticulous clinical examination and liberal use of EEG as a diagnostic/screening tool may be suggested as the possible tools to avoid such misdiagnoses.

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