West syndrome - epileptic encephalopathy at early infancy

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Abstract
West syndrome (WS), synonymously infantile spasm or epileptic spasm, is a rare type of epileptic encephalopathy occurring at early infancy that exists with variable life expectancy. It is the most common form of epileptic encephalopathy. WS presents with spasms marked by extensor or mixed movements with distinct electroencephalogram (EEG) pattern of hypsarrhythmia, flexor and psychomotor arrest. Children with west syndrome always depict abnormal EEG, but sometimes the abnormality is seen only during sleep. The incidence of infantile spasms is found closer to 1 in 2,000 children, that typically begins between 2-12 months of age and peaks between 4-8 months of age. It is observed in otherwise healthy infants and also among infants with abnormal cognitive development. If this spasm happens in older subjects, they are preferably called "epileptic spasms" rather than infantile spasms. The goal for treatment of infantile spasms is to have complete control of spasms. Hormonal therapy with ACTH or vigabatrin is the mainstay of treatment. In spite of the development of new antiepileptic drugs (AEDs), about 35-40% of cases are drug-resistant. Children affected with the west syndrome can be cured, but a successful therapy often depends on the timely diagnosis. This case report is one evidence highlighting the treatment strategy for the west syndrome, and this could be useful for any further study regarding this topic.

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INTRODUCTION
West syndrome is a rare type of epilepsy that most often affects children in early infancy. It is observed in otherwise healthy infants and also among infants with abnormal cognitive development. It is one among the childhood epileptic encephalopathic syndromes, which are a group of conditions in which sensory, cognitive and/or motor functions deteriorate as a consequence of epileptic activity (Vinayan, 2016). About 90% of patients affected with WS had their disease onset during the first year of life, with the incidence ranging from 2 to 3.5 in 10,000 live births. Predominantly, disease onset is between 3 and 7 months. The disease onset after 18 months is rare, but there are cases where onsets up to 4 years of age have been reported (Hrachovy and Frost, 2008).

Typically, the patient gets relieved of spasms by 5 years of age, but around 60% of children diagnosed with WS are affected with other types of seizures even after cessation of spasms (Riikonen, 1982).
This early life epileptic encephalopathy is characterized by a triad (a subset of) infantile spasms, the arrest of psychomotor development and hypsarrhythmia on an EEG (Lux and Osborne, 2004). Diagnosis is made based on EEG, a useful tool in the diagnosis of epilepsy and others such as neuroimaging studies and some laboratory testing (Vinayan and Moshé, 2014). If the west syndrome is suspected, a prolonged awake and sleep video-EEG study is suggested rather than a brief EEG recording (Lux and Osborne, 2004; Tatum et al., 2018). The goal for treatment of infantile spasms is to have complete control of spasms. Recent data states the use of adrenocorticotropic hormone (ACTH; short-term hormonal therapy) or vigabatrin as first-line therapy (Sharma and Viswanathan, 2008). Vigabatrin is effectively used in infantile spasms as a result of tuberous sclerosis (Salar et al., 2018). The use of Vigabatrin is limited, owing to its concerns regarding retinopathic side effects. When the syndrome occurs out of the normal range of age, it’s mostly of poor prognosis and usually suffers from severe physical and cognitive impairments.

CASE REPORT

A ten-month-old female baby, the third child of non-consanguineous parents, presented with occasional sudden jerks to backwards along with eye deviation to one side which lasts for a few seconds in the last one month. She was delivered LSCS at preterm (8 months), with a birth weight of 2.3 kg. She cried immediately after birth and had poor sucking. Her mother had a history of pregnancy-induced hypertension at five months of gestation and was on antihypertensives. The fetal movement was observed at five months of gestation with the history of decreased fetal movement. There was an incidence of uterine insult around 27-30 weeks of pregnancy.

The baby had a history of neonatal jaundice and was treated with phototherapy for 2 days. There was no particular history of excessive cry and was discharged after 10 days of life. She was apparently normal for 3 months of age. Later on, it was observed that the baby was not fixing eye on the face and had a history of partial head control presented with delayed milestones. She responds to sound but not to light with no babbling. She had tried to reach objects at seven months of age and could transfer objects between hands at nine months. On further evaluation with MRI brain at a local hospital showed the possibility of end-stage periventricular leucomalacia (PVL). On further clinical examination in AIMS hospital at Kochi, she was conscious, not fixing, following light and spasticity was present with brisk deep tendon reflexes (DTR).

Based on previous MRI and examinations, the possibility of static encephalopathy with the symptomatic west syndrome was considered probable. Her blood routines were normal, and overnight VEEG showed hypsarrhythmia. As per protocol, the plan was to initiate injection ACTH but her baseline BP was more than 95 percentile, and so it was deferred. 2D ECHO showed tiny posterior muscular VSD (left to right). She was prescribed valproic acid [VALPARIN 5 ml/200 mg syrup: 1ml TID based on 15 mg/kg/day) to treat the west syndrome symptomatically and was advised for discharge. She was further brought to the hospital after 2 months with complaints of persistent spasms and 2-4 episodes of flexor jerk/clausate about 3-4 times a day. EEG repeated on OPD basis showed persistent hypsarrhythmia. Her blood and metabolic investigations have done normal, hence planned to initiate ACTH therapy after explaining the pros and cons to the parents while monitoring for blood sugar and BP, which remained within the normal limits. Gradually the spasms decreased during the hospital stay.

DISCUSSION

ACTH plays a vital role in the neuroendocrine cascade of responses as a result of stressful stimuli, including insults to the brain. As a result of neuroendocrine stress, native ACTH that is synthesized in the pituitary stimulates the adrenal cortex to release steroids (mainly cortisol in human) into the bloodstream. ACTH functions in a similar manner when given as therapy also. Steroids readily cross the blood-brain barrier and bind to receptors widely distributed in the brain. ACTH helps to stimulate parts of the brain to release other hormones. The potentiation of nerve growth-promoting activity can also facilitate the therapeutic action of ACTH (Riikonen, 2001).

Moderate evidence showed short term or low dose treatment with ACTH helped to treat infantile spasm (Wilmshurst et al., 2015). ACTH given for shorter duration or at low doses might help in decreasing the risk of side effects. Further, studies needed to be carried out to put forward evidence regarding more effective treatment.

CONCLUSION

West syndrome is a rare type of epilepsy observed with poor outcome. Hormonal therapy with ACTH or vigabatrin is the mainstay of treatment. The administration of adequate therapy is aimed at bal-
ancing the seizure control along with developmental improvement. In spite of the development of new antiepileptic drugs (AEDs), about 35-40% of cases are drug-resistant. There is an absolute necessity for advance studies that can provide early diagnostic measures and more effective treatment.

**Conflict of Interest**

The authors declare no potential conflict of interest for this study.

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