CASE REPORT

"West Syndrome—Infantile Spasms": A Pediatric Case Report

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Abstract

West syndrome is a rare, severe form of epilepsy with onset in infancy and early childhood. It combines episodes of epileptic spasms that occur in a cluster, an abnormal pattern of interictal electroencephalogram termed as hypsarrhythmia and neuropsychomotor delay. The syndrome mainly results from brain dysfunction in the prenatal, perinatal, or postnatal period and focal lesions early in life may secondarily affect other sites in the brain presenting with some degree of developmental delay and mental retardation. The oral manifestations vary to a large extent presenting as generalized tooth wear, gingival enlargement, multiple white spot lesions, and a high-arched palate. This case report highlights the importance of early diagnosis, various clinical features, and management in a pediatric patient with West syndrome.

Keywords: Hypsarrhythmia, Infantile spasm, West syndrome.

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Introduction

West syndrome is a unique epilepsy disorder characterized by a triad of infantile spasms, hypsarrhythmia, and arrest of psychomotor development.[1] The syndrome was first described in 1841 by an English physician, William West, and the term "West Syndrome" was given by Dr Gastaut, Dr Poirier, and Dr Pampiglione in the early 1960.[2] The incidence of West syndrome ranges from 2 to 3.5 per 10,000 live births and occurs more commonly in boys in comparison to girls with the ratio being 60:40.[3–5] The characteristic infantile epileptic spasms occur in clusters and particularly on arousal from sleep. The interictal electroencephalogram (EEG) pattern is termed "hypsarrhythmia" and was first described by Gibbs and Gibbs in 1952.[6] West syndrome is classified into three main categories as symptomatic, idiopathic, and cryptogenic based on associated etiological factors.[7] The long-term prognosis is related to the etiological cause. Children with idiopathic West syndrome have a better prognosis than those with symptomatic infantile spasms.[6] Genetic causes that have been linked to an increased occurrence of infantile spasms include mutations in the ARX gene and cyclin-dependent kinase-like 5 (CDKL5) gene on the short arm of the X chromosome.[6] The ARX gene codes for ARX protein which regulates other genes that contribute to normal brain development. Mutations in this gene reduce the amount and function of the ARX protein, which disrupts normal brain development, leading to seizures and intellectual disability.[6] The CDKL5 gene is a serine/threonine kinase that is expressed in abundance in the neurons of the cerebral cortex and regulates morphogenesis and signaling pathways across the brain. Mutations in this gene cause an abnormal pattern of signaling pathways from the cerebral cortex leading to epileptic spasms.[8]

The orodental findings may vary from case to case and have not been widely studied. This case report highlights the various clinical features and management in a pediatric patient diagnosed with a symptomatic form of West syndrome.

Case Description

An 8-year-old female patient reported to the Department of Pedodontics and Preventive Dentistry, Maulana Azad Institute of Dental Sciences, New Delhi, with a chief complaint of multiple decayed teeth. The patient presented with an inability to communicate with poor motor skills and displayed stranger anxiety. The child was born prematurely via prolonged delivery with an associated history of maternal dehydration, fever, and vaginal infection just before delivery. The patient presented with seizures from as early as 20 days of age, which occurred initially on awakening. The frequency of seizures progressed to multiple times in a day occurring almost daily. The seizures were associated with violent jerking of the upper and lower limbs. The EEG depicted abnormal signs of hypsarrhythmia and diffuse cerebral atrophy was noticeable on MRI. History of delayed milestones such as sitting without support at two and a half years, neck holding at 9 months was also present. The patient was initially on adrenocorticotropic hormone (ACTH) therapy, which was later replaced by anticonvulsant medication. Family history revealed one healthy female sibling of the patient with no significant medical history.

Examination

Presently, at the age of 8 years, the patient appeared underweight with a lean body type. The general physical examination revealed neuropsychomotor delay (Fig. 1) with atrophy of upper and lower limbs.

The extraoral examination revealed arched eyebrows, a wide nasal bridge, and hypertelorism. The lips were incompetent with a normal mouth opening (Fig. 2).

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Fig. 1: General physical appearance

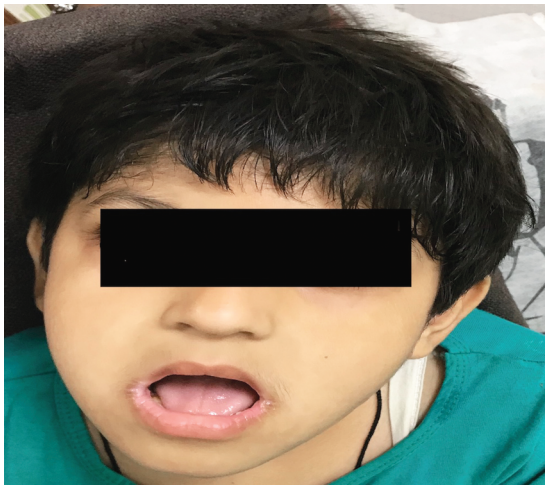

Fig. 2: Extraoral picture

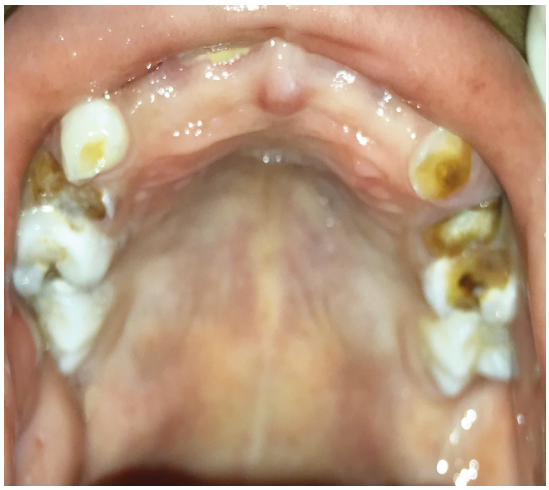

Fig. 3: Intraoral picture depicts high-arched palate

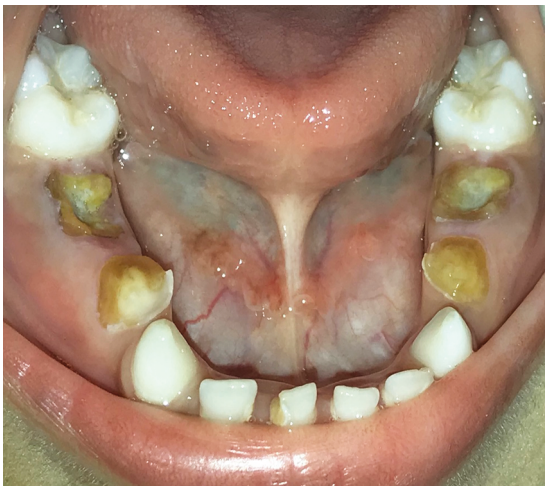

Fig. 4: Intraoral picture depicts mandibular arch and lingual tongue-tie

On intraoral examination, mixed dentition was present with Angle's Class I malocclusion. The gingiva appeared healthy with a pale pink hue and without any visible signs of enlargement. A high-arched palate (Fig. 3) and lingual tongue-tie (Fig. 4) were noticeable. Root stumps of upper central and lateral incisors, upper left canine, upper and lower first molars bilaterally, and lower second molars were present. The upper second molars depicted occlusal caries (Figs 4 and 5).

The dental treatment plan focused on providing preventive as well as restorative care while simultaneously catering to the behavior and intellectual deficit of the patient. The parents were educated about the importance of brushing twice daily and following healthy oral hygiene practices. The dietary counseling included advice regarding the use of a sticky form of carbohydrates such as candies and food items and beverages with high sugar content and limiting the frequency of the same. It was emphasized that oral home care should support professional care for the best outcome. The treatment plan included oral prophylaxis, fluoride application in the form of varnish to prevent accidental ingestion of fluoride gel, pit and fissure sealant application, and extraction of root stumps of deciduous upper central and lateral incisors under local anesthesia. The patient had reduced ability to cooperate with even simple procedures and had poor communication skills,

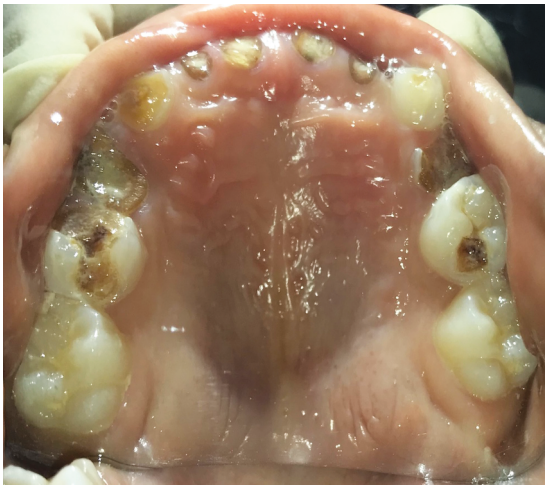

Fig. 5: Intraoral picture depicts maxillary arch

which required the presence of the mother in the operatory and the procedures to be carried out with utmost patience and compassion. The procedures were carried out cautiously and the patient was repetitively pacified with the help of the mother to gain more

cooperation. The recall appointments were planned and preventive measures were reinforced at every appointment.

Discussion

West syndrome is a unique disorder that affects individuals during infancy and early childhood. At least 76 different synonyms have been used to describe infantile spasms such as massive spasms, infantile massive spasms, flexion spasms, jack-knife convulsions, nodding convulsions, and lightning fits.[6]

West syndrome is a severe form of epilepsy having an onset in early childhood and is composed of a triad of infantile epileptic spasms occurring in the cluster, characteristic EEG abnormality called "hypsarrhythmia" and delayed psychomotor development.[1] The onset of infantile spasms varies from the first week of life to 3 years of age, with a peak at 6 months and usually cease before 5 years of age.[6] This characteristic triad has been reported by Regis et al.,[9] Khatri et al.,[10] and Dantas-Neta et al.[11] in children presenting with West syndrome. In the present case, seizures were present from as early as 20 days of age, which occurred initially on awakening and later on, progressed to occur multiple times in a day, almost daily. The seizures were associated with violent jerking of the upper and lower limbs. There were abnormal signs of modified hypsarrhythmia on EEG and delayed milestones such as delayed cry, sitting without support, not before 2 years of age, neck holding at 9 months, monosyllabic speech, and dysarthria were also present. The upper and lower limbs depicted atrophy to some extent with poor motor skills.

West syndrome is classified into three main categories as symptomatic, idiopathic, and cryptogenic based on associated etiological factors.[7] Symptomatic West syndrome is associated with an identifiable cause which may be prenatal, perinatal, or postnatal. The prenatal causes include toxemia during pregnancy, trisomy 21, intrauterine infection, or organic academia. Among the perinatal causes, persistent neonatal hypoglycemia and hypoxic–ischemic encephalopathy may lead to focal lesions in the brain and the postnatal causes mainly include trauma, meningitis, herpes simplex encephalopathy, and the various inborn errors of metabolism such as maple syrup urine disease and phenylketonuria.[9] Idiopathic West syndrome includes cases in which spasms occur without any identifiable cause and other neurological signs or symptoms whereas the term cryptogenic is reserved for cases suspected of being symptomatic with an unidentified underlying cause.[9] The long-term prognosis is related to the etiological cause. Rantala et al.[12] have reported that the prognosis of idiopathic West syndrome is better as compared with symptomatic infantile spasms, whereas Matsuo et al.[13] reported that West syndrome has an unfavorable prognosis regardless of the etiology. The present case was diagnosed as a symptomatic form of West syndrome as a history of maternal toxemia, dehydration, fever, and vaginal infection during the course of pregnancy and an episode of preterm prolonged delivery was present.

The diverse etiological factors of West syndrome generate a stress response causing increased production of corticotrophin-releasing hormone (CRH) from the immature brain resulting in epileptic spasms. The action of CRH is inhibited by ACTH via a negative feedback mechanism and is an established treatment modality for the syndrome.[14] The present case was initially on ACTH therapy which was later replaced by anticonvulsant medication.

Oral manifestations have not been widely explored and may present as mouth breathing, deep palate, gingival hyperplasia, severe gingivitis, anterior open bite, alteration in the chronology of tooth eruption, widespread tooth wear, fissured tongue, and lingual interposition between arches.[9,11] Furthermore, the presence of multiple white spot carious lesions, poor oral hygiene, and a diet high in carbohydrates have also been reported.[9] Gingival enlargement may be related to long-term usage of anticonvulsant medication and poor oral hygiene due to a lack of manual dexterity in such children. Regis et al.[9] reported multiple white spot lesions on labial surfaces, generalized tooth wear, gingival enlargement, and altered chronology of teeth in a case of West syndrome. Similar findings including white spot lesions, bruxism, and high-arched palate were reported by Khatri et al.[10] Dantas-Neta et al.[11] reported other oral findings like an anterior open bite with tongue interposition in-between arches, fissured tongue, and a low caries experience among a sample of children with West syndrome. The present case presented with a high-arched palate, lingual tongue-tie, root stumps of upper central and lateral incisors, upper left canine, upper and lower first molars, and lower second molars bilaterally and carious upper second molars. The gingiva appeared healthy with a pale pink hue and without any visible signs of enlargement.

A wide spectrum of disorders such as Rett's syndrome, Angelman's syndrome, and Lennox–Gastaut syndrome presents with similar orofacial findings as West syndrome. A thorough knowledge about the specific pattern of infantile spasms and associated neuropsychomotor deficit in such children can help differentiate this condition from others.

Riikonen et al.[5] studied the life expectancy, intellectual coefficient, and professional activities of 214 individuals diagnosed with West syndrome. They reported that 147 subjects survived beyond the age of 10, 25 of these survivors completed their education at a normal school, and 36 had a professional occupation. This study signified that despite the neurological deficit, with early diagnosis and prompt intervention, the life expectancy of children with West syndrome can be majorly improved. This puts an even greater emphasis on the provision of efficient dental care to such children along with lifelong medical care.

A thorough knowledge about the clinical features and potential implications of West syndrome should be acquired to provide high-quality oral healthcare to these patients. As pediatric dentists, behavioral problems should be dealt with patience and motivation for good oral health should be regularly reinforced. The use of restraints such as papoose board, towel straps can be done for additional support and stabilization of the child during dental procedures. Such supportive measures can improve patient cooperation and lead to superior behavior management. The lack of manual dexterity in children with West syndrome leads to an inability to practice the various oral hygiene measures correctly resulting in high caries activity. The provision of dental care to such patients is further jeopardized due to impaired psychomotor and social development, limited communication, difficulties in cooperation with the treatment, and fear of strangers. This brings out the importance of preventive care in such children, beginning with the eruption of deciduous teeth to continued care throughout life. The parents should be instructed about preventive strategies such as brushing, dietary alterations, and early identification of oral diseases such as dental caries so that any delay in the provision of treatment can be prevented at large.

Individual-targeted, preventive programs should be formulated at the earliest to reduce the occurrence of dental problems in such patients and a collaborative team of both medical professionals and dentists should work hand in hand to improve the quality of life in such patients.

As a healthy mouth is a mirror to a healthy body, with increased life expectancy these children demand the best dental care in the current scenario.

Conclusion

West syndrome is a rare epileptic syndrome with appreciable psychomotor impairment. The burden of the diseased state is itself crippling for the parents leading to a neglect of oral health of the patient. The lack of manual dexterity due to the neuropsychomotor impairment puts these children at a high risk of caries development. The behavioral and developmental deficit and associated muscular incoordination further pose a unique challenge in the management of this syndrome. As pediatric dentists, behavioral problems should be dealt with patience and motivation for good oral health should be regularly reinforced. The parents should be educated about the importance of preventive care, right from early childhood so that various oral diseases can be reduced at large. Dental treatment should be provided at the earliest to limit the occurrence of any other disability. Lastly, commitment to providing dental care with compassion and life-long counseling to the parents should be the sole aim in the management of children with West syndrome.

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