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The Necessity of Speech and Language Assessment in "West Syndrome": A case report

Srabanti Khemka[1], Saurav Chandra Sharma[2]

[1]Asssociate Professor, Sri Aurobindo Institute of Medical Sciences, SAU.
[2]Postgraduate (SLP) Student, Sri Aurobindo Institute of Medical Sciences, SAU.

Abstract:
West syndrome is an epileptic encephalopathy accounting for one-fourth of epilepsies occurring in the first year of life and is highly associated with cognitive impairment. Autism spectrum disorders refer to a group of developmental disorders that are characterized by a wide range of impairments in social and communicative abilities, stereo-typed behaviors, and restricted range of interests with an onset of initial symptoms present before three years of age. In children with West syndrome, autism symptoms arise frequently. The present case study reports the speech and language characteristics of a 7-year-old boy followed with the emphasis and the association of West syndrome and autism.

Keywords: West syndrome, Autism, com-DEALL

INTRODUCTION:

West syndrome (WS) is a unique epilepsy disorder characterized by a triad of infantile spasms, hypsarrhythmia, and arrest of psychomotor development. The syndrome was first described in 1841 by an English physician, William West, and the term "West Syndrome" was given in the early 1960 (Cowan & Hudson,1991). 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 (Riikonen, 2001).West syndrome is classified into three main categories as symptomatic, idiopathic, and cryptogenic based on associated etiological factors (Lux & Osborne, 2004).WS is now understood to be an age-dependent epileptic encephalopathy, an expression of brain injury to any cause; which may be pre-natal, perinatal or postnatal. West syndrome is the most common paediatric encephalopathy that begins at approximately four to seven months of age, and is characterized by infantile spasms and neuro developmental regression (Mytinger, 2021). Common co-morbidities include global developmental delay/intellectual disability, autism spectrum disorder, cerebral palsy, and visual and hearing impairment (Sharma et al., 2021). Children with infantile spasm are at high risk for dysphagia. Aspiration is commonly present in children with infantile spasms (Lawlor,2020). Screening and treating these co morbidities are important for preventing respiratory infection and optimizing growth and quality of life (Murphy, 1999).About 70-80% of children with West syndrome have mental retardation, 20% of patients with associated ASD (Strasser, et al., 2018), and 12-15% has hyperkinetic disorder (Rafnsson, 2007) suggesting that these clinical conditions share a common basis (Chong, et al., 2018).

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by impaired social communication as well as restricted repetitive interests and behaviours. However, the autism spectrum encompasses many other domains, including processing speed, learning and verbal memory, reasoning,

and problem solving (Velikonja, et al., 2019). Despite the recognition of autism spectrum disorders commonly co-occurring with epilepsy, the relation between epilepsy and autism still remains unclear (Tuchman, 2010). Studies have found increased rates of epilepsy (5% to 38.3%) in individuals with autism and autism spectrum disorders (Tuchman & Rapin, 2002). A meta-analysis study indicated the prevalence of epilepsy in people with autism spectrum disorder to be 8% and 20% respectively in the absence and presence of intellectual disability (Amiet et al., 2008). Rafnsson (2007) reported that for children diagnosed with onset of epilepsy in the first year of life, autism spectrum disorder developed in 14% while it developed in 46% of children diagnosed with West syndrome. A recent population based study on the association between seizures in the first year of life (other than infantile spasms) and ASDs reported a relatively high prevalence of ASD (7%) (Saemundsen et al., 2007b). The prevalence of ASDs among both children with infantile spasms and those with other unprovoked seizures in the first year of life seems to exceed the prevalence of ASDs in the general population, which ranges from 0.5% to 1% in recent studies (Chakrabarti & Fombonne, 2005; Baird et al., 2006; Fombonne et al., 2006; Ellefsen et al., 2007; Jonsdottir et al., 2007).

The comorbidity of epileptic syndromes and autism remain to be unclarified. Many questions remain unclear and unsolved even in the domain of speech and language characteristics and imply extensive research based on spectrum of severity present in West syndrome.The present case describes a child with west syndrome with autism symptoms.

MATERIAL AND METHOD

In the present study language skills of the child were assessed using Receptive expressive emergent language scale (REELS) and Communication DEALL developmental checklist (Com-DEALL). Cognitive and social pre-requites skills for language learning was assessed by using cognitive and social development milestone. Autism checklist, Indian scale for assessment of autism (ISAA) and DSM 5 diagnostic criteria for ADHD was administered to differentially diagnose the case. Short sensory profile (SSP) and Montreal children hospital feeding scale (MCH-FS) was used to assess sensory issues and feeding difficulties respectively.

PROCEDURE

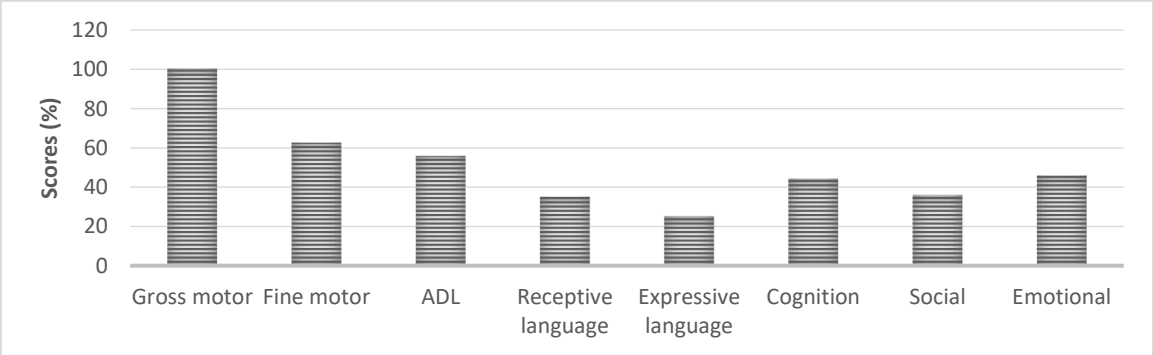
The case report concerns a 5-year-old girl of unrelated healthy Hindi speaking parents.The parents reported to the department of Audiology and Speech Language Pathology with a chief complaint of unable to speak age appropriately. This case was born at 39 weeks of gestation after an uneventful pregnancy. Parents reported of delayed birth cry and birth weight of 2.5 kgs. The patient presented with seizures from 8months of age which progressed to multiple times in a day with a duration of 5-6 seconds occurring almost daily. The seizures were associated with violent jerking of the upper and lower limbs. EEG suggested of generalized seizure disorder. Paediatric impression based on clinical symptoms, EEG and MRI report led to a diagnosis of symptomatic west syndrome. Parental perception indicated delayed motor developmental milestonesof neck holding at 8 months, sitting without support at 11 months and walking alone at the age of 18 months. Language development milestone was reported to be wholistically delayed as acquisition of babbling was at the age of 9 months, first word at 22 months and sentence was developed even at the age of 5 yrs. Behavioural problems were observed during clinical evaluation.The child predominantly used non-verbal mode of communication such as simple & contact

gestures along with vocalization and reduplicated babbling to express her needs. Comprehension of common words, few common phrases and simple sentences used in daily life were present. All the oral peripheral structures were normal in appearance and function. Prelinguistic skills of nonverbal imitation were present however eye contact and turn taking skills were inadequate and attention was single channeled.

RESULTS

The results of REELS used to assess language skills demonstrated a receptive language age ranging from 20-22 months whereas lower expressive language age of range 12-14 months. The standardized developmental checklist of com-DEALL assessed the child in eight different developmental skills. The results showed that the language scores (25% to 35%) and social scores (35.8%) of the child lagged behind in comparison to motor, emotional and the cognitive scores.

Graph 1. Scores of Communication DEALL developmental checklist



Cognitive pre requisites for language learning results indicated a piagetion stage of tertiary circular reaction and social prerequisites for language learning were at an early childhood level. These results of developmental profiles suggested an error play pattern which showed a deficit in understanding of conventional object use or meaning by brief recognitory gestures. Further the child fulfilled 11 out of 18 behaviours in the 50 % criteria of autism checklist. Following this ISAA was administered which depicted a total score of 115, suggestive of moderate autism with 70% disability however thechild did not fulfil DSM 5 diagnostic criteria for ADHD ruling out the existence of attention deficit hyperactive disorder as a comorbid condition. The interpretation of the scores of short sensory profile (SSP) suggested of deficit in tactile sensitivity, movement sensitivity, under responsive sensation and an overall low energy among the total seven domains of the test (Table 1). Total score in Montreal children hospital feeding scale was 63, indicating mild feeding difficulties.

Table 1. Scores of Short sensory profile

Domain	Score	Interpretation
Tactile sensitivity	19	Definite differences
Taste/smell sensitivity	20	Typical performance
Movement sensitivity	03	Definite difference
Underresponsive/seeks sensation	11	Definite difference
Auditory filtering	23	Typical performance

Low energy/weak	21	Definite difference
Visual/auditory	19	Typical performance

DISCUSSION

West syndrome is a severe form of epilepsy having an onset in early childhood and is composed of a triad of infantile epileptic spasms; characteristic EEG abnormality called "hypsarrhythmia" and delayed psychomotor development. (Goswami & Sharma,2021). This characteristic of West syndrome is supported by the features of the child of the present study which exhibited with epilepsy with an abnormal EEG pattern. Considering the delayed motor, cognitive and language developmental milestones of the present case in the initial years it was noted that emergence of autism symptoms was perceived by the parents in the later years.The case of the present study was labelled with moderate autism after a clinical evaluation using ISAA. This finding coincided with research by Besag, et al., (2016) which stated that West Syndrome was associated with one of the most important neurodevelopmental disorders i.e. autism spectrum disorder (ASD). Kayaalp (2007) researched that a number of patients develop autism after an initial diagnosis of West syndrome. Autism was found to persist even after the seizures were controlled in WS (Chiron et al.,1997). Speech and language manifestations have not been widely explored among West syndrome. Based on the results of the speech and language disorders the case was provisionally diagnosed as having a delay in speech and language development associated with moderate autism secondary to west syndrome. Sharma & Parmar (2023) in their case study also noted a delay in the speech and language development associated with west syndrome and was advised for speech therapy supporting the findings of the present study.

CONCLUSION

West syndrome is a rare epilepsy syndrome characterized by onset of epileptic spasms in infants. Infants may have no medical history before the onset of epileptic spasms, or may have a medical history reflecting the underlying cause. Detailed assessment of audiological and speech language evaluation is of critical importance for early identification and intervention which helps a child to improve on impaired skills. Absence of effective treatment of speech and language deficits will result in developmental plateauing and regression. Multidisciplinary approach of intervention is required for rehabilitation of children with WS.

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