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West syndrome and autism: a case report

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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, who is a 7-year-old boy followed with the diagnosis of West syndrome and autism, is presented to emphasize this association.

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Introduction

West syndrome (WS) is an epileptic encephalopathy which accounts for 4-10% of childhood epilepsies and 25% of epilepsies occurring in the first year of life. Three main features of WS are as follows: 1. Infantil spasms, 2. Cessation and regression of psychomotor development, 3. The finding of hypsarrityhmia in EEG [1,2].

West syndrome is classified into symptomatic, cryptogenic and idiopathic depending upon the cause. Approximately 75% of the patients are in symptomatic WS group in which convulsions develop due to cortical malformations, prenatal events, neurocutaneous syndromes (tuberosclerosis, sturge-weber), chromosomal impairment and metabolic diseases. In criptogenic group, the underlying cause can not be clarified, while in idiopathic WS, the psychomotor development of the patients prior to convulsion attacks is normal [3].

Course primarily rests on etiology. Criptogenic/idiopathic etiology, late onset and rapid response to treatment is associated with good prognosis. In 50% of all cases, spasms disappear before the age of two. In 70%, severe mental retardation, autism, conduct disorder, develop and in 25-50% Lennox-Gastaut syndrome is seen. Only 5-10% of the cases may show normal or near normal mental development [4].

Autism spectrum disorders represent a group of neurodevelopmental disorders characterized by marked impairment in social and communicative

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skills, restricted interest and stereotypical behavior in which first symptoms arise before the age of three [5]. The term autism spectrum disorders is used to include children with Autistic disorder, Asperger disorder and with those Pervasive Developmental Disorder-not- otherwise-specified [6]. Despite decades of recognition autism spectrum disorders commonly co-occurs with epilepsy, the relation between epilepsy and autism still remains unclear [7]. Based on a meta-analysis of Amiet et al, the prevalance of epilepsy in people with autism spectrum disorder was found to be 8% and 20% respectively in the absence and presence of intellectual disability [8]. In a prospective study, it was reported that of 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 [9]. In another community-based study, autism spectrum disorder was detected in 5% of newly diagnosed epilepsy cases. In the same study, West syndrome and mental retardation were found to be independently associated with autism spectrum disorders [10].

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Case Presentation

The case, a 7-year-old boy, referred to our clinic due to health council report. He has received special education and physical therapy for last six years. There was severe impairment in language development and social-emotional reciprocity, and repetitive behaviours in his psychiatric evaluation. He had no verbal expression and limited non-verbal communication with poor eye contact and his response to instructions was inconsistent. He had no pretend play, and he had an unusual preooccupation with holding saliva in his mouth and playing with it. He was not able to join his peers or interact with them. He was born at term after a normal pregnancy by an unremarkable delivery. He developed normally until he was five months old and after five months spasms and convulsions started and his neuro-motor development was stopped. He was diagnosed with west syndrome and valproic acid and topiramate were instituted. He is still on valproic acid. Following a period with convulsions for about 6 months, no new convulsion has occurred, but valproic asid was continued as epileptiform anomaly continued to be seen in frontal and parieto-temporal regions in EEG. When he was at the age of 2-3, it was established that he had no eye contact with people, did not look when his name was called, was not interested in people and rocked forward and backward in sitting position and lived in his own world. He was diagnosed with WS+ Autistic disorder and special education was initiated. He started to walk at the age of five, never became continent and language was not developed. There was no evidence of autistic spectrum disorders in his family history.

His weight and height were below the 3rd percentile, and his head circumference was in the 3rd percentile. Blood chemistry analysis yielded normal values. His hearing and eyesight were found to be normal in previous tests and there was no pathology in cranial MR.

His total score in the Childhood Autism Rating Scale (CARS) [11] was 52 / 60, indicating severe autism and total score on the Autism Behavior Checklist (ABC) [12] was 91 / 158 points. He was diagnosed with AD according to DSM-IV criteria [13].

Discussion

Although the course of disease depends on the underlying cause in West syndrome, intellectual disability at various levels occurs in 80% of the patients. Prognosis is better in children in idiopathic group [11]. Considering the developmental process of the present case, it was determined that he was in idiopathic/cryptogenic group, convulsions were controlled with drug treatment within 5-6 months after onset, but retardation in psychomotor development continued and autism symptoms emerged in later years. His score in childhood autism rating scale was 52, "extremely autistic" and

his score in autism behavior checklist was 91. Although there is no specific finding, 17-60% of autistic children display various EEG abnormalities. Findings are usually observed in frontal, temporal, parieto-temporal, and centro-temporal regions. In a study comparing video-EEG recordings and ages of WS patients with or without autism, it was thought that the continuation of hypsarrhythmia in later ages and the presence of dominant spike and wave findings in frontal regions were associated with the development of autism and it was concluded that hypsarrhythmia led to permanent damage in cognitive functions and behavioral skills [14]. In the present case, although convulsions did not continue, epileptic discharges continued to be seen in frontal and parieto-temporal regions, and in parallel he had severe autism symptoms, which is consistent with the literature. However, as his history was evaluated retrospectively, and some of his medical records could not be reached, it was not known how long hypsarrhythmia in his EEG lasted and to what degree it contributed to his autism.

In a study in which children diagnosed with WS were screened with autistic behavior checklist and children with high probability of being autistic were compared with those with low probability, it was established that in those with WS+high probability autism, cryptogenic and symptomatic groups constituted 5.9% and 94.1% of the cases. In the symptomatic group, congenital causes were the leading ones [15]. In a study of Saemundsen, 83% of the cases with autistic WS displayed in symptomatic group [9]. The present case was a patient with WS belonging to criptogenic/idiopathic group with severe autism symptoms.

Mental retardation is present in 75% of autistic cases [8]. In another study, psychomotor retardation was detected in 86% of WS group while in 100% of WS and high probability autism group [15]. Consistent with the findings in the literature, our case had severe autism symptoms and marked psychomotor retardation.

WS is frequently associated with autism. Autism is a chronic disorder lasting lifelong and the most important treatment modality is education. As early onset of education is of critical importance, care should be taken in order not to overlook these cases and to refer them to treatment.

Informed Consent

Written informed consent was obtained from the patient for the publication of this case report.

Competing interests

The authors declare that they have no competing interests with respect to the authorship and/or publication of this article.

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