Clinical and developmental profile of children with West Syndrome in a tertiary care hospital in South India: A prospective observational study

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Abstract

Objectives To describe the etiological, clinical and developmental profile of children with West Syndrome.

Methods A detailed history, clinical examination, DP-3 developmental assessment, EEG and MRI were conducted in the initial encounter. Children were followed up at 6 months to assess seizure control, E-CHESS scores and neurodevelopmental status.

Results Out of 35 children, 62.9 % had Symptomatic West Syndrome. No significant differences were found between Symptomatic and Cryptogenic groups in demographics, seizure type, response to treatment, or E-CHESS scores. Neurodevelopment was adversely affected in all groups influenced by time lag in initiating treatment and E-CHESS scores.

Conclusion The Symptomatic group dominated over the cryptogenic group with potentially preventable perinatal asphyxia as the commonest cause. Neurodevelopment was greatly affected with effects persisting despite seizure control. Delayed treatment and the severity of epilepsy can adversely affect the neurodevelopmental outcomes.

Key Notes Symptomatic predominance in West syndrome is noted. Early intervention is needed to improve developmental outcomes. Seizure control alone does not guarantee good neurodevelopment.

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- West syndrome
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Introduction

West syndrome is a devastating age dependent epileptic encephalopathy characterized by a triad of epileptic spasms (IS), hypsarrhythmia and developmental retardation. It can impair the brain maturation, cognition, and development of the child. It most commonly presents in the second to twelfth months of life. It is often caused by an organic brain dysfunction whose origins may be prenatal, perinatal, or postnatal. This study aimed to explore the etiological and clinical spectrum of children with West syndrome, with a special emphasis on their developmental outcomes. It seeks to enhance paediatricians' understanding of this devastating, age-dependent epileptic encephalopathy.

Methods

This prospective observational study was conducted at our institution from September 2018 to May 2020. Children aged 1 month to 5 years who presented to the emergency, inpatient, or outpatient departments either as known cases of West syndrome or newly diagnosed based on the International League Against Epilepsy (ILAE) definition were included in the study. The study was approved by the institutional ethics committee.

At the initial encounter, a detailed history, clinical findings, developmental assessment (DP-3 scoring), electroencephalogram (EEG), neuroimaging (MRI), and other relevant investigations were performed. Children were followed up six months after the initiation of treatment, with assessments focused on:

- Seizure control
- Adverse outcomes
- Early Childhood Epilepsy Severity Scale (E-CHESS) scoring
- Developmental status based on DP-3 scoring.

The Developmental Profile 3 (DP-3) scoring system evaluates five domains: physical, adaptive, social-emotional, cognitive, and communication. A total standard score and a composite General Development Standard (GDS) score were calculated, with minimum scores of 50 for individual domains and 40 for the composite GDS.

The Early Childhood Epilepsy Severity Scale (E-CHESS) is a tool designed to assess epilepsy severity, aid in evaluating treatment efficacy, and investigate the impact of epilepsy severity on development. It includes five key measures:

1. Duration of seizure occurrence
2. Seizure frequency
3. Number of seizure types
4. Number of anticonvulsant medications used
5. Response to treatment

Each variable was scored from 0 to 3, with higher scores indicating greater severity. Children in the study were categorized into two groups based on their E-CHESS scores: <10 and ≥ 10.

Statistical Analysis

Data analysis was conducted using SPSS 22.0 and R environment version 3.2.2. Descriptive and inferential statistical methods were applied.

- Continuous variables were presented as Mean ± SD (Min–Max).
- Categorical variables were presented as Number (%).
- Statistical significance was set at $P < 0.05$.

Clinical trial number: Not applicable.

Results

A total of 35 children were recruited for the study, of whom 19 (54.3%) were male. The children were categorized into symptomatic (22, 63%) and cryptogenic (13, 37%) groups.

Among the symptomatic group, 13 children (59.1%) were born preterm. Both groups were comparable, except for the significantly higher prevalence of prematurity in the symptomatic group ($P = 0.003$) (Table I). There was no significant association between gender, age distribution, consanguinity, or mode of delivery across the two groups. CNS malformations were observed in 20 children (90.9%) within the symptomatic group.

Seizure type analysis showed that 28 children (80%) presented with flexor spasms. The relationship of spasms with sleep was evaluated, and 21 (60%) had no association with sleep, while 12 (35%) experienced spasms upon awakening.

Most children did not exhibit specific features preceding or following the spasms, and 22 (62.8%) had no additional seizure types apart from infantile spasms. No significant difference was observed in these characteristics between the symptomatic and cryptogenic groups.

A total of 22 children (62.8%) had at least one comorbidity associated with West syndrome, with 17 (77.3%) in the symptomatic group and 5 (38.5%) in the cryptogenic group. Among these, visual impairment was the most common comorbidity in the symptomatic group, while feeding difficulties were more frequently observed in the cryptogenic group (Table 1).

Children with symptomatic West syndrome had significantly higher rates of developmental delay prior to seizure onset than those with cryptogenic West syndrome ($P < 0.01$). In contrast, developmental regression following seizure onset was observed more frequently in the cryptogenic group, which was statistically significant ($P = 0.046$) (Table 1).

The response to treatment was assessed based on complete seizure cessation, partial cessation, or no improvement. Both groups showed comparable responses, with 14 children (63.6%) in the symptomatic group and 8 children (61.5%) in the cryptogenic group achieving complete seizure cessation. Girls demonstrated a better response to treatment compared to boys.

The E-CHESS score at six months post-treatment initiation was comparable between the symptomatic and cryptogenic groups.

At the six-month follow-up, DP-3 scores across different domains and overall developmental scores showed no significant difference between the two groups, except in the physical domain, where the cryptogenic group had better outcomes (Table II). Within each DP-3 domain, developmental scores varied significantly based on treatment response (*ANOVA analysis*). Children with no response to treatment had significantly lower developmental scores compared to those who showed partial or complete cessation.

A significant association was found between higher E-CHESS scores and lower developmental scores at follow-up, with the social-emotional domain being the most affected. Furthermore, children with higher E-CHESS scores demonstrated greater declines in developmental scores, particularly in the social-emotional and communication domains (Table 2).

Early initiation of treatment was associated with better developmental outcomes at follow-up compared to delayed treatment initiation, with the difference in General Developmental Score being statistically significant ($P = 0.017$) (Table 2).

Discussion

A prospective observational study was conducted at Aster Medcity, Kochi, from September 2018 to May 2020 to evaluate the clinical spectrum, aetiology, and outcomes of West Syndrome in children. A total of 35 children meeting the inclusion criteria were enrolled. The mean age at presentation was 10.6 months (range: 3–24 months), comparable to Sehgal R et al.[1] (median: 12 months). The mean age of spasm onset was 4.8 months, aligning with findings by Kalra et al.[2] (5 months). No significant association was observed between seizure onset and gender or preterm gestation. The mean delay in presentation was 5.7 months, likely due to delayed recognition and referral. The male-to-female ratio was 1.18:1, consistent with prior studies [1–3].

A symptomatic aetiology was identified in 62.9% of children, while 37.1% were cryptogenic. Perinatal risk factors were present in 22 children, with antenatal risks in 11. Birth asphyxia was the predominant perinatal factor, followed by neonatal hypoglycaemia and meningitis. Preterm birth was significantly associated with symptomatic West Syndrome. CNS malformations were found in 20 children, including cerebral atrophy, white matter injury, and polymicrogyria. Two children had tuberous sclerosis. No genetic or metabolic causes were identified, possibly due to limited investigations. These findings are consistent with previous studies [1–3].

Flexor spasms were the most common type (80%), followed by extensor and mixed. The mean number of spasms per cluster was 10.5, with a mean of 6.6 clusters per day. Sleep–wake cycle-related spasms were seen in 40%, most commonly on awakening.

Associated seizures developed in 37.1% of children, with two cases evolving into Lennox-Gastaut Syndrome.

Microcephaly was present in 36.4% of symptomatic cases, and 90.9% had pre-existing developmental delay, consistent with findings by Kaushik et al[3]. and Singhi et al[4]. At least one comorbidity was seen in 62.9% of children. Vision impairment was most frequent, followed by hearing impairment, feeding difficulties, and sleep disturbances. Vision impairment was significantly higher in symptomatic cases, as also reported by Sehgal et al[1]. and Kaushik et al[3]. One child had Down syndrome, aligning with literature highlighting increased incidence of infantile spasms and risk of autism spectrum disorder in these children[5–7].

A favourable epilepsy outcome (≥50% reduction in spasms) at six months was achieved in 85.7% of children, while complete cessation occurred in 62.9%. ACTH resulted in complete cessation in 66.6% and partial response in 24.2%. Vigabatrin showed no significant benefit in two children with tuberous sclerosis but offered partial improvement in three ACTH non-responders. These seizure outcomes are consistent with previous studies on epilepsy management and treatment response in West Syndrome [8,9].

Despite seizure control, developmental stagnation or regression persisted in most children, emphasizing the importance of early and aggressive intervention strategies [10]. Symptomatic cases had significant pre-existing developmental delay, whereas cryptogenic cases showed higher post-seizure regression. Delayed treatment initiation significantly impacted communication outcomes. Only 14.3% of children had favourable neurodevelopmental outcomes, and 37.1% had profound delay, similar to Sehgal et al[1]., who reported 8.4% favourable outcomes. In contrast, Partikian et al[11]. reported better cognitive outcomes in US-based populations.

This study highlights the clinical spectrum, aetiology, and outcomes of West Syndrome. The predominance of symptomatic cases aligns with prior studies [1–4], emphasizing the roles of perinatal and structural risk factors. The association between preterm birth and symptomatic cases underscores the need for enhanced neonatal care and early neurodevelopmental assessment.

Although seizure control was achieved in most children, neurodevelopmental outcomes remained poor, with only 14.3% showing improvement. This highlights the need for early intervention programs, including rehabilitation and special education, to reduce long-term disability. Multidisciplinary care involving neurology, developmental paediatrics, and rehabilitation services is essential to improve quality of life.

Study limitations include the small sample size, single-centre design, and short-term follow-up (six months). In addition, genetic and metabolic evaluations were limited, possibly underestimating these aetiologies. Future studies should involve larger multicentre cohorts with longer follow-up to better define prognostic factors and optimize therapeutic strategies[12].

Table 1. ***Demographic and Baseline Clinical Characteristics of the Study Groups***

Variable	***Symptomatic (n=22)*** ***Number (Percentage)***	***Cryptogenic (n=13)*** ***Number (Percentage)***	***P value***	***Interpretation***
Gender (Male/Female)	14 / 8	5 / 8	0.149	Not significant
Age <6 months at onset	4 (18.2%)	6 (46.2%)	0.083	Earlier onset in cryptogenic group
Preterm birth	13 (59.1%)	1 (7.7%)	0.003	Significant
Mode of delivery (LSCS)[b]	8 (36.4%)	2 (15.4%)	0.259	Not significant
Consanguinity	0 (0%)	2 (15.4%)	0.131	Not significant

Variable	*Symptomatic (n=22)* *Number (Percentage)*	*Cryptogenic (n=13)* *Number (Percentage)*	*P value*	*Interpretation*
Family history of IS[c]	2 (9.1%)	1 (7.7%)	1.000	Not significant
Pre-existing developmental delay	20 (90.9%)	4 (30.8%)	<0.01	Significant
Developmental regression	6 (27.3%)	8 (61.5%)	0.046	Significant
Microcephaly	8 (36.4%)	2 (15.4%)	0.259	Not significant
Facial dysmorphism	6 (27.3%)	0 (0%)	0.064	Borderline significance
Neurocutaneous markers	6 (27.3%)	8 (61.5%)	0.075	Trend toward cryptogenic group
Comorbidities (any)[d]	17 (77.3%)	5 (38.5%)		More common in symptomatic group
Associated seizures	8 (36.4%)	5 (38.5%)	0.901	Not significant

P <0.05 considered significant. Statistical tests used: Chi-square test or Fisher's exact test,

[b] Lower Segment Caesarean Section, [c] Infantile Spasm, [d] comorbidities assessed: visual impairment, hearing impairment, sleep disturbances, feeding difficulties, and recurrent infections

Table 2. ***Developmental and Clinical Outcomes at 6-Month Follow-Up***

Variable	*Symptomatic* *Mean score (SD)*	*Cryptogenic* *Mean score (SD)*	*P value*	*Interpretation*
[a]DP3 – Physical domain	58.27 (9.55)	66.46 (12.90)	0.039[b]	Significant difference
[a]DP3 – Adaptive domain	60.45 (11.23)	66.84 (11.98)	0.122	Not significant
[a]DP3 – Socio-emotional domain	60.60 (11.55)	64.84 (13.77)	0.344	Not significant
[a]DP3 – Cognitive domain	60.68 (14.07)	66.92 (16.15)	0.239	Not significant
[a]DP3 – Communicative domain	65.86 (14.61)	73.23 (16.27)	0.176	Not significant
[c]General Development Score (GDS)	48.50 (12.94)	56.00 (17.00)	0.150	Not significant
[d]E-CHESS score	8.27 (2.45)	7.85 (2.97)	0.649	Similar severity
	Symptomatic *Number (Percentage)*	*Cryptogenic* *Number (Percentage)*		
Complete seizure cessation	14 (63.6%)	8 (61.5%)		Comparable
Partial response	5 (22.7%)	3 (23.1%)		Comparable
No improvement	3 (13.6%)	2 (15.4%)		Comparable

[a]DP3: Developmental Profile-3, [b]P <0.05 considered significant. Statistical analysis used: Student's t-test (continuous), Chi-square or Fisher's exact test (categorical). [c]GDS: General Development Score (maximum = 100). [d]E-CHESS: Early Childhood Epilepsy Severity Scale (range: 0–15; higher = greater severity).

KEY MESSAGE
WHAT THIS STUDY ADDS

This study underscores symptomatic predominance in West Syndrome and highlights the need for early intervention to improve developmental outcomes.

Funding

No funding was received for this study.

Competing Interests

The authors declare no competing interests.

Ethics Approval

The study was approved by the Institutional Research and Ethics Committee, Aster Medcity, Kochi (Ref: AM/EC/67-2018; Date: 03/09/2018).

Consent to Participate

Informed consent was obtained from the parents or legal guardians of all individual participants included in the study.

Human Ethics and Consent to Participate Declaration

The study adhered to the principles of the Declaration of Helsinki. Institutional ethics approval was obtained, and informed consent was secured from parents or legal guardians.

Data Availability

The datasets generated and/or analyzed during the current study are available from the corresponding author on reasonable request.

References

1. Sehgal R, Gulati S, Sapra S, Tripathi M, Kabra M, Pandey RM. Neurodevelopmental and epilepsy outcome in children aged one to five years with infantile spasms—A North Indian cohort. Epilepsy Res [Internet]. 2014 Mar 1 [cited 2025 Apr 9];108(3):526–34. Available from: https://www.sciencedirect.com/science/article/pii/S0920121113003410
2. Kalra V, Gulati S, Pandey RM, Menon S. West syndrome and other infantile epileptic encephalopathies – Indian hospital experience. Brain Dev [Internet]. 2001 Nov 1 [cited 2025 Apr 9];23(7):593–602. Available from: https://www.brainanddevelopment.com/article/S0387-7604(01)00288-1/abstract
3. Kaushik JS, Patra B, Sharma S, Yadav D, Aneja S. Clinical spectrum and treatment outcome of West Syndrome in children from Northern India. Seizure. 2013;22(8):617–21.
4. Singhi P, Ray M. Profile of West syndrome in North Indian children. Brain Dev. 2005;27(2):135–40.
5. Daniels D, Knupp K, Benke T, Wolter-Warmerdam K, Moran M, Hickey F. Infantile Spasms in Children with Down Syndrome: Identification and Treatment Response. Glob Pediatr Health. 2019; 6:2333794X18821939.
6. Sanmaneechai O,Sogawa Y, Silver W, Ballaban-Gil K, Moshé SL, Shinnar S.Treatment outcomes of West syndrome in infants with Down syndrome. Pediatr Neurol. 2013;48(1):42–7.
7. Berg AT, Plioplys S, Tuchman R. Risk and correlates of autism spectrum disorder in children with epilepsy: a community-based study. J Child Neurol. 2011;26(5):540-7.
8. Eisermann MM, DeLaRaillère A, Dellatolas G, Tozzi E, Nabbout R, Dulac O, et al. Infantile spasms in Down syndrome--effects of delayed anticonvulsive treatment. Epilepsy Res. 2003;55(1–2):21–7.
9. Pavone P, Polizzi A, Marino SD, Corsello G, Falsaperla R, Marino S, et al. West syndrome: a comprehensive review. Neurol Sci Off J Ital Neurol Soc Ital Soc Clin Neurophysiol. 2020;41(12):3547–62.
10. O'Callaghan FJK, Lux AL, Darke K, Edwards SW, Hancock E, Johnson AL, et al. The effect of lead time to treatment and of age of onset on developmental outcome at 4 years in infantile spasms: Evidence from the United Kingdom Infantile Spasms Study. Epilepsia [Internet]. 2011 [cited 2025 Apr 9];52(7):1359–64. Available from: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1528-1167.2011.03127.x
11. Partikian A, Mitchell WG. Major adverse events associated with treatment of infantile spasms. J Child Neurol. 2007;22(12):1360–6.
12. Riikonen R. Long-term outcome of patients with West syndrome. Brain Dev [Internet]. 2001 Nov 1 [cited 2025 Apr 9];23(7):683–7. Available from: https://www.sciencedirect.com/science/article/pii/S0387760401003072