European Journal of Clinical Pharmacy

Print ISSN: 2385-409X **Online ISSN**: 3105-0409

Website: https://farmclin.com/

Original Research Article

CLINICO-ETIOLOGICAL SPECTRUM OF INFANTILE SPASM

Article History:

Name of Author:

Dr Ramswaroop Gurjar,[1] Dr Neha Sharma,[2] Dr Vikash Gurjar,[3*] Dr S. Sitaraman[4]

Affiliation: *[1]Senior Resident, Department of paediatrics, RUHS, Jaipur*
[2]Senior Resident, Department of paediatrics, SMS Medical College and Hospital, Jaipur
[3]Medical Officer, Department of Medical, Health and Family Welfare, Kotputli–Behror, Rajasthan
[4]Senior Professor and Unit Head, Department of Pediatrics, SMS Medical College and Hospital.

Corresponding Author:
Vikash Gurjar

Received: 07-11-2025
Revised: 26-11-2025
Accepted: 18-12-2025
Published: 31-12-2025

Abstract: infentile spasm (IS) is a distinctive epileptic disorder of infancy and early childhood and represents one of the most severe forms of childhood epilepsy, often reflecting a true epileptic encephalopathy. AIM: To Study the Clinico-etiological Spectrum of Infantile Spasms. METHODOLOGY: The present study was conducted in the Department of Paediatrics, SMS Jaipur. It was designed as a hospital-based cross-sectional study. The study duration extended from June 2024 to may 2025, carried out over a period of 1 year or until completion of the required sample size, whichever occurred earlier. RESULT: The majority of children with infantile spasms presented within the first year of life and showed significant neurodevelopmental impairment, with hypoxic-ischemic encephalopathy being the most common aetiology. Classical hypsarrhythmia on EEG and flexor spasms were the predominant electroclinical features, and neuroimaging abnormalities were observed in most patients. CONCLUSION: Infantile spasms are largely associated with identifiable and often preventable perinatal and neonatal causes, particularly hypoxic and metabolic insults. Early diagnosis, optimal perinatal care, and prompt initiation of treatment are essential to improve neurological outcomes in affected children.

Keywords: Infentile, spasm, neurological,

INTRODUCTION

Infantile spasm (IS) is a distinctive epileptic disorder of infancy and early childhood and represents one of the most severe forms of childhood epilepsy[1], often reflecting a true epileptic encephalopathy. Children may present with isolated infantile spasms or as part of West syndrome, which is classically defined by the triad of epileptic spasms, characteristic electroencephalographic (EEG) abnormalities in the form of hypsarrhythmia or modified hypsarrhythmia, and progressive neurodevelopmental regression.[2] The condition was first described in 1841 by West, who observed these seizures in his own child. Clinically, infantile spasms are characterized by sudden, brief contractions involving flexion and/or extension of the neck, trunk, and limbs, with flexor, extensor, or mixed patterns that may occur singly or in clusters[3]. IS is a relatively rare disorder, with an estimated incidence ranging from 1.6 to 4.5 per 10,000 live births, accounting for approximately 2000–2500 new cases annually. The age of onset varies from the first

week of life up to 4.5 years, though most cases present between 3 and 7 months of age.[4] While gender predilection remains unclear, some studies suggest a slight male predominance with a male-to-female ratio of about 60:40[5]. The aetiology of infantile spasms is highly heterogeneous and includes a wide spectrum of prenatal, perinatal, and postnatal causes such as neurocutaneous syndromes, metabolic disorders, cortical malformations, hypoxic–ischemic brain injury, perinatal insults, postnatal infections, and head trauma[6]. In resource-limited settings, neonatal asphyxial brain injury is frequently reported as the most common acquired cause, while neonatal hypoglycaemic brain injury has also emerged as an important contributor.[7] Genetic abnormalities constitute another significant etiological category.[8] The pathophysiology of IS is not fully understood; proposed mechanisms include a nonspecific cerebral insult occurring at a critical stage of brain development and dysfunction of the hypothalamic–pituitary–adrenal axis related to early-life stress or immunological disturbances. Infantile spasms are broadly classified as symptomatic or cryptogenic, with an additional idiopathic subgroup proposed by the International League Against Epilepsy (ILAE).[9,10] Epileptic spasms were formally recognized as a distinct seizure type by the ILAE in 2001 and are typically associated with characteristic ictal EEG changes, although classic hypsarrhythmia may not always be present. Early recognition and prompt initiation of treatment—most commonly hormonal therapy with ACTH or steroids, along with antiseizure medications or dietary therapy—are crucial, as shorter lead time to treatment is associated with improved seizure control and better developmental outcomes.[11] Despite this, delays in diagnosis and treatment remain common.

AIM

To Study the Clinico-etiological Spectrum of Infantile Spasms.

MATERIAL AND METHODS

The present study was conducted in the Department of Pediatrics at….. .. It was designed as a hospital-based cross-sectional study. The study duration extended from ….., carried out over a period of …. year or until completion of the required sample size, whichever occurred earlier. The study population included all children aged between 3 months and 5 years who were diagnosed with infantile spasms at our centre during the study period.Children were included in the study if they were between 3 months and 5 years of age and had a confirmed diagnosis of infantile spasms based on a detailed history of spasms either reported by caregivers or directly witnessed by a pediatrician, along with documentation of classical or modified hypsarrhythmia on electroencephalography (EEG) at any time during evaluation. Inclusion was subject to the availability of informed written consent from parents or legal guardians.

Children were excluded from the study if consent was refused by parents or caregivers, if a primary caregiver was not available at the time of enrolment, or if significant clinical data or medical records required for the study were unavailable or incomplete.

RESULTS

Table 1 : Distribution of participants according to age group

Age group	Frequency	Percent
≤1 year	28	45.2%
1-2 years	19	30.6%
2-5 years	175	24.2%
Total	**62**	100%

The age-wise distribution showed that the majority of children with infantile spasms were aged ≤1 year (45.2%), followed by those between 1–2 years (30.6%). Children in the 2–5 years age group constituted 24.2% of the study population, indicating that infantile spasms predominantly present in the first year of life.

Table 2 : Distribution of Participants according to Need of Resuscitation

	Frequency	Percent
Cried delayed after birth	19	30.6%
Cried immediately after birth	43	69.4%
Total	**62**	100%

A history of delayed cry after birth was present in 30.6% of children, while the majority of patients (69.4%) cried immediately after birth. This finding suggests that a significant proportion of cases had no apparent perinatal asphyxia at birth.

Table 3 : Distribution of participants according to history of NICU stay

NICU stay	Frequency	Percent
Yes	37	59.7%
No	25	40.3%
Total	62	100%

A history of NICU stay was present in 59.7% of the study subjects, whereas 40.3% did not require NICU

admission. This reflects a high burden of perinatal or neonatal complications among children with infantile spasms.

Table 4: Distribution of participants according to aetiology

Aetiology	Frequency	Percent
Hypoxic-ischemic encephalopathy (HIE)	15	24.9%
Hypoglycaemia	11	17.74%
Structural brain abnormalities	9	14.52%
NNJ	7	11.3%
CNS infections	6	9.68%
Cryptogenic	4	6.45%
Genetic / chromosomal disorders	3	4.84%
Trauma	2	3.23%
Cerebrovascular disease	2	3.23%
Vit-B-12 deficiency	2	3.23%
PKU	1	1.61%

Hypoxic-ischemic encephalopathy was the most common aetiology, accounting for 24.9% of cases, followed by hypoglycaemia (17.7%) and structural brain abnormalities (14.5%). Other causes included neonatal jaundice, CNS infections, genetic and metabolic disorders, while 6.5% of cases remained cryptogenic.

Table 5: Distribution of participants according to clinical spectrum

Clinical spectrum	Frequency	Percent
Developmental delay	62	100%
Abnormal movement	62	100%
Seizure	62	100%
Microcephaly	24	38.7%
Increased tone	16	25.8%
Feeding difficulties	9	14.5%
Hypotonia	5	8.1%
Sleep disturbance	2	3.2%
Recurrent infection	2	3.2%

All children in the study had developmental delay, abnormal movements, and seizures at presentation (100% each). Microcephaly was observed in 38.7% of cases, while abnormalities of tone, feeding difficulties, hypotonia, sleep disturbances, and recurrent infections were seen in a smaller proportion of patients.

Table 6 : Distribution of participants according to type of spasm

Spasm	Frequency	Percent
Flexor	29	46.8%
Extensor	18	29%
Mixed	15	24.2%
Total	62	100%

Flexor spasms were the most common type, observed in 46.8% of children, followed by extensor spasms in 29.0% of cases. Mixed spasms accounted for 24.2% of the study population.

Table 7 : Frequency of spasm per day distribution among study participants

Mean	8.21
Median	8.0
Std. Deviation	2.62
Minimum	4.02
Maximum	18.0

The mean age of the study subjects was 8.21 months with a median age of 8.0 months and a standard deviation of 2.62 months. The age ranged from a minimum of 4.02 months to a maximum of 18.0 months.

Table 8: Distribution of EEG findings among study participants:

Findings	Frequency	Percent
Classic Hypsarrhythmia	37	59.7%
Modified hypsarrhythmia	14	22.6%
Focal abnormalities	11	17.7%
Total	62	100%

Classic hypsarrhythmia was the most common EEG finding, observed in 59.7% of children, followed by modified hypsarrhythmia in 22.6% of cases. Focal EEG abnormalities were seen in 17.7% of the study population.

Table 9: NEUROIMAGING (MRI/CT) FINDINGS

Neuroimaging Findings	Number of Cases (n)	Percentage (%)
Normal	12	19.4
Cortical malformations	14	22.6
Hypoxic-ischemic changes	20	32.3
Tuberous sclerosis	6	9.7
Other abnormalities	10	16
Total	62	100

Neuroimaging revealed hypoxic-ischemic changes as the most common finding, present in 32.3% of cases, followed by cortical malformations in 22.6% of children. Normal imaging was seen in 19.4% of patients, while tuberous sclerosis and other abnormalities accounted for the remaining cases.

DISCUSSION

The age-wise distribution of children with infantile spasms in the present study shows a clear predominance of younger age groups. The majority of cases, 28 children (45.2%), were aged ≤ 1 year, indicating that infantile spasms most commonly present during early infancy. This was followed by 19 children (30.6%) in the 1–2 years age group. A smaller proportion of children belonged to the 2–5 years age group, accounting for 24.2% of cases. More than three-fourths of the patients were below two years of age at presentation.Infantile spasms predominantly manifest in the first two years of life, emphasizing the importance of early recognition and timely intervention.Surana M et al12 did a similar kind of study and a total 113 included in the study. Mean (±SD) IS onset age was 6.86 months (±4.25).
In the present study, a majority of children, 43 out of 62 (69.4%), cried immediately after birth, indicating an uncomplicated immediate postnatal adaptation in most cases. However, 19 children (30.6%) had a delayed cry after birth, suggestive of possible perinatal compromise. Delayed crying at birth is often associated with perinatal asphyxia and subsequent neurological insult. Such perinatal factors are known to play an important role in the development of epileptic encephalopathies, including infantile spasms. The relatively high proportion of delayed cry highlights the contribution of adverse perinatal events in the etiopathogenesis of infantile spasms. This finding emphasizes the importance of optimal perinatal care and early neonatal resuscitation to prevent long-term neurological sequelae.Ibrahim S et al13 did a similar study and As per parental recall, the mean time for cry after birth was 7.5 ± 3.5 minutes.

More than half of the study population required NICU admission, with 37 children (59.7%) having a history of NICU stay. A substantial proportion of patients (40.3%) did not require NICU care in the neonatal period. The high rate of NICU admission reflects the presence of significant perinatal or neonatal complications among affected children. Conditions such as perinatal asphyxia, neonatal hypoglycaemia, and neonatal jaundice commonly necessitate intensive care and are known risk factors for infantile spasms. This observation highlights the role of adverse neonatal events in the etiopathogenesis of infantile spasms. Early identification and optimal management of neonatal complications may help reduce the subsequent burden of epileptic encephalopathy.

Hypoxic-ischemic encephalopathy (HIE) was the most common etiological factor identified in the study, accounting for 24.9% of cases, followed by hypoglycaemia in 17.74% of children. Structural brain abnormalities constituted 14.52% of cases, while neonatal neonatal jaundice (NNJ) was observed in 11.3%. Central nervous system infections were responsible for 9.68% of cases, highlighting the contribution of postnatal acquired insults. Cryptogenic infantile spasms accounted for 6.45% of the study population, whereas proven genetic or chromosomal disorders were identified in 4.84%. Other less common etiologies included trauma, cerebrovascular disease, vitamin B12 deficiency, and phenylketonuria, together contributing to a small proportion of cases.Osborne et al14 revealed that of 207 infants, 127 (61%) had proven etiology,

68 (33%) had no identified etiology, and 12 (6%) were not fully investigated. Etiologies were prenatal in 63, perinatal in 38, postnatal in 8, and 18 others. The most common etiologies were: hypoxic–ischemic encephalopathy (HIE) 21 (10%), chromosomal 16 (8%), malformations 16 (8%), stroke 16 (8%), tuberous sclerosis complex (TSC) 15 (7%), and periventricular leukomalacia or hemorrhage 11 (5%). The remaining 32 etiologies were all individually uncommon. Response to

treatment is given for individual etiologies.
All children in the study presented with developmental delay, abnormal movements, and seizures, each observed in 100% of cases, highlighting the severe neurodevelopmental impact of infantile spasms. Microcephaly was noted in 38.7% of patients, indicating significant underlying brain involvement. Abnormal muscle tone was also common, with increased tone seen in 25.8% and hypotonia in 8.1% of children. Feeding difficulties were present in 14.5% of cases, reflecting associated neurological dysfunction. Sleep disturbances and recurrent infections were less frequent, each observed in 3.2% of patients.

Flexor spasms were the most common type observed in the study, accounting for 46.8% of the cases. Extensor spasms constituted 29% of the patients, making them the second most frequent pattern. Mixed spasms were seen in 24.2% of children. The predominance of flexor spasms highlights the typical presentation of infantile spasms in early childhood. The presence of mixed and extensor spasms indicates variability in the clinical manifestation of the disorder. Chandra S et al15 revealed that flexor type of spasm was found in 88% and extensor type was in 8%. Perinatal asphyxia being the most common etiology and present in 72.3%.

The mean age of the children at presentation was 8.21 months with a standard deviation of 2.62 months, indicating that most patients presented within the first year of life. The median age was 8.0 months, which was comparable to the mean, suggesting a relatively symmetrical age distribution. The minimum age at presentation was 4.02 months, while the maximum age was 18.0 months. This highlights that infantile spasms predominantly manifested during early infancy, with fewer cases presenting beyond one year of age. The narrow dispersion around the mean reflects clustering of cases in the typical age range for infantile spasms.
Electroencephalography findings in the present study revealed classical hypsarrhythmia in the majority of cases (59.7%). Modified hypsarrhythmia was observed in 22.6% of the children. Focal EEG abnormalities were noted in 17.7% of cases. The predominance of classical hypsarrhythmia highlights its strong association with infantile spasms. However, a substantial proportion of patients showed modified or focal patterns. This emphasizes the variability of EEG manifestations in infantile spasms.

Neuroimaging abnormalities were observed in a majority of children with infantile spasms in the present study. Hypoxic-ischemic changes were the most common neuroimaging finding, seen in 32.3% of cases. Cortical malformations were identified in 22.6% of children, indicating a significant contribution of structural brain abnormalities. Tuberous sclerosis was noted in 9.7% of patients. Other neuroimaging abnormalities accounted for 16.0% of cases. Normal neuroimaging findings were observed in 19.4% of children.

CONCLUSION

The present study highlights that infantile spasms predominantly manifest during early infancy, with the majority of affected children presenting within the first year of life. A significant proportion of patients had evidence of adverse perinatal and neonatal events, such as delayed cry at birth and requirement of NICU admission, underscoring the important role of perinatal insults in the etiopathogenesis of infantile spasms. Hypoxic-ischemic encephalopathy emerged as the most common etiological factor, followed by hypoglycaemia and structural brain abnormalities, emphasizing that potentially preventable and acquired causes constitute a major burden in this population.

Clinically, all children exhibited developmental delay, abnormal movements, and seizures, reflecting the severe neurodevelopmental impact of infantile spasms. Flexor spasms were the most common seizure type, and classical hypsarrhythmia was the predominant EEG finding, though modified and focal patterns were also frequently observed, indicating variability in electroclinical presentation. Neuroimaging revealed abnormalities in the majority of cases, further supporting the association of infantile spasms with underlying brain injury or malformations.The study demonstrates that infantile spasms are largely associated with identifiable etiologies, many of which are related to perinatal and neonatal factors. Early recognition, timely neurophysiological evaluation, and prompt management of neonatal complications may play a crucial role in reducing the incidence and improving the outcomes of infantile spasms. The findings reinforce the need for strengthened perinatal care and early referral to tertiary centers for optimal diagnosis and management.

REFERENCES

1)Go CY, Mackay MT, Weiss SK, Stephens D, Adams-Webber T, Ashwal S, Snead OC., Child Neurology Society. American Academy of Neurology.
2)Evidence-based guideline update: medical treatment of infantile spasms. Report of the Guideline Development Subcommittee of the American
3)Academy of Neurology and the Practice Committee of the Child Neurology Society. Neurology. 2012 Jun 12;78(24):1974-80.
4)Petroff OA, Rothman DL, Behar KL, Collins TL, Mattson RH. Human brain GABA levels

rise rapidly after initiation of vigabatrin therapy. Neurology. 1996 Dec;47(6):1567-71.
5)Shields WD, Shewmon DA, Chugani HT, Peacock WJ. Treatment of infantile spasms: medical or surgical? Epilepsia. 1992;33 Suppl 4:S26-31.
6)Kramer U, Sue WC, Mikati MA. Focal features in West syndrome indicating candidacy for surgery. Pediatr Neurol. 1997 Apr;16(3):213-7.
7)Moseley BD, Nickels K, Wirrell EC. Surgical outcomes for intractable epilepsy in children with epileptic spasms. J Child Neurol. 2012 Jun;27(6):713-20.
8)Glaze DG, Zion TE. Infantile spasms. Curr Probl Pediatr. 1985 Nov;15(11):1- Sharma S, Kaushik JS, Srivastava K, et al. Association of Child Neurology (AOCN) - Indian Epilepsy Society (IES) Consensus Guidelines for the Diagnosis and Management of West Syndrome. Indian Pediatr. 2021;58(1):54-66.
9)Nawaz S, Gajre MP, Adhikari A, Shagufta. West syndrome: Clinic etiological profile, and response to treatment in Western India. Int J Contemp Pediatr 2020;7:1367-72.
10)Liao J, Huang T, Srour M, Xiao Y, Chen Y, Lin S, Chen L, Hu Y, Men L, Wen J, Li B, Wen F, Xiong L. Status Epilepticus Manifested as Continuous Epileptic Spasms. Front Neurol. 2020 Feb 14;11:65.
11)Barbarrosa EP, Ferrer IDLCP, Tovani-Palone MR. West Syndrome: Clinical Characteristics, Therapeutics, Outcomes and Prognosis. ELECTRON J GEN MED. 2020;17(2):em190.
12)Surana P, Symonds JD, Srivastava P, Geetha TS, Jain R, Vedant R et al. Infantile spasms: Etiology, lead time and treatment response in a resource limited setting. Epilepsy Behav Rep. 2020 Oct 17;14:100397.
13)Ibrahim S, Gulab S, Ishaque S, Saleem T. Clinical profile and treatment of infantile spasms using vigabatrin and ACTH--a developing country perspective. BMC Pediatr. 2010 Jan 15;10:1.
14)Osborne JP, Lux AL, Edwards SW, Hancock E, Johnson AL, Kennedy CR et al. The underlying etiology of infantile spasms (West syndrome): information from the United Kingdom Infantile Spasms Study (UKISS) on contemporary causes and their classification. Epilepsia. 2010 Oct;51(10):2168-74.
15)Chandra S, Kumar R. Clinico-aetiological Profile and Outcome of West Syndrome from North India. HK J Paediatr (new series) 2016;21:262-265.

