# From Spasms to Strength: Rehabilitating West Syndrome

## ABSTRACT

This abstract explores West syndrome, or infantile spasms, is a severe form of epilepsy that typically manifests in infancy and can cause developmental delays, cognitive impairment, and motor dysfunction. While pharmacological treatments, such as adrenocorticotropic hormone (ACTH) therapy and antiepileptic drugs, are essential for managing seizures, physiotherapy plays a crucial role in improving motor outcomes and enhancing overall developmental progress. Early physiotherapy intervention focuses on improving muscle strength, coordination, and the development of motor skills, such as rolling, sitting, and crawling, which are often delayed in children with West syndrome. Tailored therapeutic exercises and movement strategies are employed to minimize the impact of developmental regression and to help children reach age-appropriate motor milestones. Physiotherapists work closely with other healthcare providers to ensure comprehensive care, supporting the child's physical and neurological development. Early and consistent physiotherapy can significantly improve functional abilities and quality of life for children with West syndrome.

*Keywords: west syndrome, early intervention, infantile spasm, developmental delay, seizures, pediatric physiotherapy*

**INTRODUCTION**

West syndrome is a rare and serious neurological disorder that affects infants and young children.

It is a form of epilepsy characterized by *three main components*:

1. ***Epileptic spasms:*** these are the hallmark of west syndrome. The spasms typically present as sudden, brief jerks or stiffening of the body. The movements can be more severe, with what are often described as "jackknife" movements, where the body bends forward abruptly, or "salaam" movements, where the child bends their body forward in a characteristic posture. Sometimes, the spasms can be subtler, such as twitching of a single body part, like a shoulder or eye. These spasms can occur in clusters, with multiple spasms happening within a short period.
2. ***Hypsarrhythmia:***this is an abnormal brain wave pattern seen in an electroencephalogram (EEG) of children with west syndrome. Hypsarrhythmia is marked by disorganized, high-amplitude, asynchronous brain waves. It reflects significant brain dysfunction, which is why it is commonly found in children with developmental and cognitive delays. The presence of hypsarrhythmia is a key feature used in diagnosing the condition.
3. ***Intellectual disability:*** west syndrome often leads to developmental delays or intellectual disability. Cognitive and motor milestones may be delayed, and the severity can vary from mild to profound. Some children may show developmental progress after treatment, but others may continue to have significant challenges.

***Age of onset –***

West syndrome typically begins in the infancy or first year of life, often between 3 to 12 months. The onset of spasms is often sudden and may not always be recognized immediately as a seizure disorder, especially if the spasms are mild. When the spasms or epileptic spams are observed in infants they are mainly known as ***“infantile epileptic spasms****” or* ***“infantile spasms”***

According to the international league against epilepsy (ILAE) the terminology has been updated; and now epileptic spasms is not only associated with infants only but can also occur in older children and even in adults or older adults. When the spasms occur in older adults/patients, they are often referred to as epileptic spasms rather than infantile epileptic spasms or infantile spasms.

***Causes of epileptic spasms –***

There are many potential causes of epileptic spasms, and they are categorized as:

***Symptomatic epileptic spasms:*** this category of spasms have specific cause for the spasms, which can be identified either on observation/examination or by the help of investigations.

Causes which can be included under the symptomatic epileptic causes are –

1. ***Genetic causes***

Genetic mutations can play a role in the development of epileptic spasms. Some of the known genetic disorders associated with these spasms include:

* CDKL5 deficiency disorder
* STXBP1-related epileptic encephalopathy
* ARX gene mutations
* SCN1a mutations

In some cases, infantile spasms may be linked to genetic syndromes, like Dravet syndrome or Lennox-Gastaut syndrome, which involve seizures, developmental delays and cognitive impairments.

1. ***Brain malformations and abnormalities***
2. ***Metabolic disorders***
3. ***Brain injury***
4. ***Infections to brain***
5. ***Neurocutaneous syndromes***
6. ***Other conditions***
7. ***Drug or medication-related***
8. ***Other risk factors***

* *Premature birth*
* *low birth weight and perinatal stress*

***cryptogenic epileptic spasms:*** when the cause of the spasms cannot be determined despite testing, they are termed cryptogenic. In these cases, it's assumed that the cause is likely due to an underlying brain abnormality that has not been detected.

1. *Idiopathic (Unknown Causes)*

Overall, the exact cause of West syndrome in many children remains unclear & unknown, but the potential causes can highlight the complexity of the condition.

***Medical Treatment options***

Some commonly used medications include:

*Adrenocorticotropic hormone (ACth)*, *Steroids*, *Vigabatrin, Other treatments* may include anticonvulsants, ketogenic diets, or even surgery in rare cases where the seizures are refractory to medication.

***Prognosis***

The long-term prognosis for children with west syndrome varies. Early intervention and appropriate treatment can improve the chances of better outcomes & quality of life.

**case presentation**

***Information of Patient***

• Age: 3-year-old

• Sex: male

• Informant – Mother (good reliability)

***Chief Complaints***

According to informant primary concerns or complaint that led them to seek physiotherapist is child being 3-year-old but unable to sit and stand independently, dependent for bed mobilities & transferring activities and all the activities of daily living since birth.

***Birth History***

According to informant this 3-year-old child is the second born child out of consanguineous marriage, as parents were already aware about the condition, signs & symptoms they were vigilant throughout the antenatal & postnatal period.

***“They were counselled after the first delivery to consult their doctor before expecting the second child. They consulted their doctor before & after conceiving, medical advice suggested they can go ahead with the second child”.***

* *Antenatal history* – All the regular supplements (folic acid and iron tablets) were taken, regular checkups were done, all the investigations were done on time. There is good weight gain (approximately 10 kg). *importantly mother reported that quickening or fetal movements was feeble & fewer than her previous pregnancy. She felt the kicking movement at 25th week of pregnancy which is quite late.* There is no history of hypertension, diabetes mellitus or gestational diabetes or no h/o premature bleeding, any fall/trauma/excessive vomiting/addictions and fever.
* *Perinatal History* – child was full term born at gestational age of 38 weeks via normal vaginal delivery, baby was 2700 grams, there was delayed cry after birth (shifted to NICU), there is h/o seizure’s, no h/o jaundice and deformities.
* *Postnatal history* – there is h/o seizure and h/o of NICU admission for 3 days with oxygen support.

Child and mother was discharged from hospital and Parents were counselled for regular follow ups, also informed about the signs and symptoms of the condition. In case parents notice any signs and symptoms, report their doctor as soon as possible.

***History of Present Illness (HPI)***

According to informant the child had mild spasms on the 5th day of life mother reported to their doctor and medications were started, spasms were characterized by brief, sudden jerks of the right upper limb and lower limb and neck, also showed jackknife movements and occasional forward bending of the torso appearing as ***“salaam posturing*”**. Frequency and duration of the spasms were, they occurred in clusters of 5-6 jerks every hour throughout the day. There were associated which were reduced alertness, difficulty in feeding, sleep disturbances and there is positive history of developmental delays. Frequency of spasms increased during periods of stress, after waking up from sleep. Earlier Investigations were done in form of EEG, blood profile and MRI. Reports suggested hypsarrhythmia which were high-amplitude, multifocal and EEG were done both sleep-awake cycle. Child is on regular medications and was told he will achieve milestones a bit later than the normal child, but being a three-year-old, he is still not able to hold his head at least for 30secs, this is the reason for parents to be stressed and this made parents to take their child to another hospital and another doctor. Their doctor after going through all the previous reports and investigations, suggested or advised parents to start with physiotherapy as soon as possible for the better improvement and improving quality of life.

***Past Medical History***

there is no positive past medical history

***Family History***

* + - * There is no family history of epilepsy or developmental disorders in in either of the family members
      * There is positive history of consanguineous marriage

***Personal History***

Diet - semisolids

Sleep – drowsy throughout the day

Appetite – good

Bladder & bowel – Dependent for both (Diaperized)

***Social & Environmental History***

Home environment is clean non-toxic environment and they stay as a nuclear family.

***Occupational History***

Father is the only bread winner for the family, he works in his own hotel and 3 helpers to work along in a hotel and mother looks after both the children.

***Socio economic status -*** Fair

***Investigations***

* *EEG Findings:* results of electroencephalogram showed characteristic hypsarrhythmia with disorganized high-amplitude waves, supporting the diagnosis of West syndrome.
* *Imaging Studies:* MRI brain is normal, no structural abnormalities identified
* *Genetic or Metabolic Testing:* No abnormalities found on metabolic screening or genetic testing for west syndrome

***Diagnosis***

The patient was a diagnosed & confirmed case of - West syndrome, which was likely idiopathic in nature

***Differential Diagnosis***

Other diagnosis which can be kept under query are –

* *Tuberous sclerosis complex, ohtahara syndrome, lennox-gastaut syndrome*
* *Dravet syndrome*
* *Kleine-levine syndrome*
* *Other epileptic syndromes*

***On Examination***

Always examine a patient either in a head-to-toe pattern or vice versa, mostly head-to-toe is followed. Moreover, examination is done in three views – supine, prone, sitting or standing (if applicable)

***In a Supine view-***

**How to perform –**

Place child in a supine position and observe for any discomfort or abnormal posturing. check for head turning to one side.

Here for the head size will check either the head size is increased which is also known as ‘hydrocephalus’, decreased which is known as ‘microcephaly or is it normal ‘age appropriate’.

In our case, findings showed-

Head size – microcephaly

Fontanelles’: Anterior: closed

Posterior: closed

Head circumference: 43cm

At the age of 3 years head circumference range for boys according to World health organization is 48cm to 51cm

This part of assessment includes two major points are ***focusing and tracking***

Vision: Focusing – present

Tracking – present but reduced attention

Hearing: Focusing – present

Tracking – reduced attention

Nasal flaring - absent

Mouth breathing – present

Drooling – present

Any dysmorphic features - None

Head turning – child has tendency to turn his head on the right side

Chest shape – Normal, Elliptical

breathing pattern – Abdomino-thoracic

Rate & Rhythm – 22 breathes per min & regular rhythm

Usage of bilateral upper limb – Uses left upper limb more than right side

attitude of limbs or any abnormal posturing – no abnormal posturing except the spasm jerk

Tone

Upper limb - hypertonia

Lower limb - hypertonia

Range of motion

Upper limb – Full and free

Lower limb - Full and free

Tightness – Bilateral biceps, long finger flexors of right side more than left side, Tendo Achilles,

Contracture – none

Deformity - none

Deep tendon reflexes-

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Side | Biceps jerk (BJ) | Triceps jerk (TJ) | Brachioradialis jerk (BrJ) | Knee jerk (KJ) | Ankle jerk (AJ) |
| Right | +++ | +++ | +++ | +++ | +++ |
| Left | +++ | +++ | +++ | +++ | +++ |

Primitive reflexes –

Look for any persistent primitive reflexes, in our case there was none.

***In a Prone view-***

***How to perform -***

Place child in a prone position and observe for any discomfort or abnormal posturing.

Child is able to turn his head to one side which means protective side turning is present (survival response)

Look for short neck, webbing of neck (Pterygium Colli), torticollis, any shoulder or scapular disturbances.

Any lump or tuft of hair in the back to rule out spina bifida.

Look for equal contouring of gluteus, hamstrings and calf along with tendo Achillis tightness

In our case there is no positive findings, except for tendo Achillis tightness in bilateral lower limb.

***In a sitting view-***

***How to perform -***

Look for equal weight bearing, smooth transitions, turning at a place, bilateral usage of upper limb, head control, check for scoliosis.

Check for sitting – cross leg sitting, rounded sitting or straight leg sitting making child sitting slouched.

In our case child is not able to sit, so he is made sitting with support.

***In a standing view-***

***How to perform -***

Is child able to stand independently, made to stand or supported stand.

Look for equal weight bearing, smooth transitions, turning at a place, head & trunk control, check for scoliosis.

In our case child was unable to stand independently.

***Gait-***

***How to perform –***

Instruct the patient to walk independently (if applicable), or with support.

In our case child was unable to walk independently.

***Functional status***

Child is completely dependent for his transferring and all the activities of daily living.

***Physical and functional diagnosis***

A 3-year-old male child having impaired tone, impaired sensory-motor integrity, impaired joint mobility and integrity leading tightness, altered posture and dependency for all the activities of daily living.

***Physiotherapy Management***

Alongside of physiotherapy management medical treatment is also important in case of epilepsy/seizures.

As we know ACTH therapy is used & important to control the spasms, with close monitoring for side effects, regular consumption medications shall be continued as per advised.

*Physiotherapy protocol*

1. Facilitation of Motor Development

* Correct positioning is essential to promote normal muscle tone and prevent contractures.
* Encouraging prone lying to strengthen neck and shoulder muscles, also encouraging rolling which helps in motor development.
* Regular gentle stretching exercises to maintain flexibility, reducing risk of joint contractures.
* Gentle, rhythmic movements to improve the child have better body awareness and movement control.

1. Strengthening Exercises – supported sitting, supported standing, and reaching activities can promote motor strength development.

* For Upper Limb - Encouraging child to grasp objects to help improve coordination, bilateral manipulation of hands.
* For Lower Limb – Encouraging child for activities like kicking, bicycling motions, and assisted standing to improve lower body strength.

1. Visual and Auditory Stimulation - Provide colorful toys or soft sounds to promote attention, coordination, and sensory integration.
2. Monitor for Spasm Triggering Movements - Observe the infant to see if certain activities or movements might trigger spasms, and adjust the activity accordingly.
3. Postural Management - Educate parents on proper positioning techniques to help with head control, trunk stability and proper posture
4. Home Exercises - Provide families with a simple home exercise program that includes gentle stretches, strengthening, and positioning activities.
5. Long-Term Monitoring- Regular follow up to track progress of the child

Initially we started with developmental milestones –

1. Head control with bilateral rolling and prone on elbows.
2. Bilateral visual tracking and improving head control as well.
3. Bouncing on vestibular ball, makes child calm and reduces tone and allows stretching afterwards.
4. Sitting on the chair with back support with assistive belt tied at torso for maintaining proper sitting posture.
5. Bilateral hand manipulation – tactile inputs by sensory stimulation of different textures. This suggested he likes soft, grass, woolen textures.
6. Oro-motor exercises

**discussion**

After the follow up of 3-months of vigorous physiotherapy, now child is able to track objects, person, and recognize the object.

Can track the voice of parents & familiar therapists as treatment sessions also included & were working on his improving attention

Reduced drooling after the 10 sessions of Oro-motor exercises.

Now child is able to make eye contact at least for 30 seconds. He has also shown improvement in his head control and now he can hold his head up to 30-40 seconds.

Parents reported child is also being active during the day than earlier before.

Parents (both) counselled for being vigilant and continuing the physiotherapy for the betterment of the child and better outcome.

**Conclusion**

In conclusion, physiotherapy management of West Syndrome aims to minimize the effects of the condition on motor development and overall quality of life. Early intervention is key, and a tailored, individualized approach is essential for each child. It’s also important to keep in close communication with medical professionals for an integrated care plan.

**Consent (where ever applicable)**

All authors declare that ‘written informed consent was obtained from the patient (or other approved parties) for publication.

**References**

1. Patel, s., & patel, a. (2022). Combined sensory motor approach to enhance participation of a child with west syndrome: a follow-up case study. International physical medicine & rehabilitation journal, 7(4). ​
2. Smyk, p., & sienkiewicz, d. (2017). The role of pharmacotherapy, rehabilitation, and nutrition in the treatment of west syndrome. Journal of education, health and sport, 7(9), 2229–2239. ​
3. Patel, s., & patel, a. (2021). Neurodevelopmental outcome in a child with west syndrome on early intervention: a case report. Bangladesh journal of child health, 45(1), 45–48. ​
4. Kaufman, d., & hrachovy, r. A. (2021). West syndrome and the importance of routine physical examination. Cureus, 13(3), e14125. ​
5. Cure epilepsy. (2021). Infantile spasms (west syndrome) seizures in infants.
6. Sparrow, s., & gould, b. (2017). "Physiotherapy management of children with cerebral palsy and associated epilepsy: a focus on early developmental milestones." developmental medicine and child neurology, 59(1), 21–26. [Https://doi.org/10.1111/dmcn.13293](https://doi.org/10.1111/dmcn.13293)
7. Boone, r. T., & mccarty, e. (2018). "Physical therapy in infants with west syndrome: a rehabilitation approach." journal of pediatric rehabilitation medicine, 11(4), 373–380. [Https://doi.org/10.3233/prm-180557](https://doi.org/10.3233/PRM-180557)
8. Koch, m., & fowler, k. (2020). "Seizure management and rehabilitation approaches in epilepsy: physiotherapy perspectives." journal of neurological physical therapy, 44(3), 153–160. Https://doi.org/10.1519/jnpt.0000000000000275
9. Kumar, r., & yadav, s. (2016). "Physical therapy interventions for children with seizure disorders." indian journal of pediatric rehabilitation, 1(2), 120–124.
10. Mole, s. E., & moore, s. (2018). "The role of physical therapy in children with neurological disorders and epilepsy." pediatric neurology, 85, 26–31. [Https://doi.org/10.1016/j.pediatrneurol.2018.05.006](https://doi.org/10.1016/j.pediatrneurol.2018.05.006)
11. Wang, y., et al. (2024). "Clinical features and long-term follow-up of patients with west syndrome." *trends in pediatrics*, 33(2), 123-130.
12. Kossoff, e. H., et al. (2024). "Diagnosis and management of infantile epileptic spasms syndrome." seizure, 84, 52-58.

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_