West's Syndrome (Infantile Spasms)

Synonyms: salaam spasms

The syndrome was classically described by Dr W J West in a letter to the editor of The Lancet in 1841[1]. He described the events as:

'... bobbings that cause a complete heaving of the head forward towards his knees, and then immediately relaxing into the upright position ... these bowings and relaxings would be repeated alternately at intervals of a few seconds, and repeated from 10 to 20 or more times at each attack, which attack would not continue more than 2 or 3 minutes; he sometimes has 2, 3 or more attacks in the day.'

Today the disease is classified as having three components, although diagnosis can still be made in the absence of one:

- Infantile spasms.
- Hypsarrhythmia (classical very abnormal electroencephalograph (EEG) that occurs even between attacks).
- General learning disability.

Epidemiology

The peak incidence is between 4 and 7 months of age, with 90% starting under 1 year of age. It is confined to infants and very small children. A small number have idiopathic infantile spasms with normal growth and development prior to the onset of infantile spasms, and no known aetiology[2].

Aetiology

Causes can be divided into symptomatic or idiopathic.

Symptomatic

Almost any disorder that can produce brain damage can be associated with infantile spasms.

- Prenatal conditions (including hydrocephalus, microcephaly, Sturge-Weber syndrome, tuberous sclerosis); genetic syndromes (such as Aicardi's syndrome and Down's syndrome), hypoxic or ischaemic brain damage, congenital infections, and trauma.
- Perinatal disorders include hypoxia or ischaemia of the brain, meningitis, encephalitis, trauma and intracranial haemorrhage.
- Postnatal disorders include pyridoxine dependency, maple syrup urine disease, phenylketonuria, meningitis, degenerative diseases, biotinidase deficiency, and trauma.

Idiopathic

Idiopathic infantile spasms are diagnosed if normal psychomotor development precedes the onset of symptoms, no underlying disorders or definite presumptive causes are present and no neurological or neuroradiological abnormalities exist. Some people use the terms ‘idiopathic’ and ‘cryptogenic’ as synonymous.

Presentation

Spasms begin with a sudden, rapid, tonic contraction of the trunk and limb muscles, with gradual relaxation over 0.5 to 2 seconds. Contractions can last 5 or 10 seconds. They may range from a gentle nodding of the head to a powerful movement of the body.

- The spasms tend to occur in clusters. There may be dozens of them with a lapse of 5 to 30 seconds between each. They tend to occur just before sleep or on awaking. They can occur in sleep but this is unusual.
- Spasms can be flexor, extensor, or a mixture of both.
- Flexor spasms consist of brief contractions of the flexor muscles of the neck, trunk and limbs, causing a brief jerk. They may resemble a self-hugging motion and often there is a cry. The child relaxes and the jerk repeats. These attacks occur in clusters throughout the day and last anywhere from less than 1 minute to 10 or 15 minutes, or more in some patients.
- Extensor spasms are contractions of the extensor muscles, with sudden extension of the neck and trunk with extension and abduction of the limbs.
- Mixed spasms are the most common type, with flexion of the neck and arms with extension of the legs, or flexion of the legs with extension of the arms.
- Various series give the incidence of mixed spasms as around 40-50%, flexor spasms at about 35-40% and extensor spasms at 20-25%.

Between 70% and 90% have psychomotor delay or reversal[2]. Physical examination usually reveals no abnormality, although it may help with an underlying diagnosis like the adenoma sebaceum of tuberous sclerosis.
Examination with Wood's lamp is often helpful to see the hypopigmented skin lesions of this condition.

Mid to moderate growth restriction is common.

Neurological examination reveals nothing pathognomonic, although there may be vague and nonspecific features reflecting brain damage, and the effect of fits or medication.

Ophthalmic examination may reveal evidence of one of the syndromes associated with the condition.

Investigation

- Blood tests include FBC, LFTs, renal function, glucose, calcium, magnesium and phosphate.
- Blood cultures, urine examination and cerebrospinal fluid analysis if infection is suspected.
- Urine amino acids and organic acids and serum biotinidase.
- Brain MRI has the highest yield initial study in determining the aetiology in infantile spasms.
- EEG is essential, as hypsarrhythmia is crucial to the diagnosis:
  - A prolonged study to obtain waking and sleeping traces may be required.
  - Hypsarrhythmia is a characteristic interictal pattern of chaotic, high to extremely high-voltage polymorphic delta and theta rhythms with superimposed multifocal spikes and wave discharges.
  - Multiple variations of this pattern are possible, including focal or asymmetric hypsarrhythmia.
  - The ictal pattern has 11 different variations, although one is present in about 40%.
  - There is no correlation between the ictal patterns and the type of fit.

Management

This is aimed at control of seizures, with the fewest side-effects and the best quality of life. Adrenocorticotropic hormone (ACTH), prednisolone and vigabatrin have the best evidence as first-line medications.

Several other anti-epileptic drugs (levetiracetam, nitrazepam, sodium valproate, topiramate, zonisamide) are usually used as add-on or adjunctive treatment in refractory cases. Pyridoxine (or pyridoxal phosphate) and the ketogenic diet are also established treatment options in refractory cases.

First-line of treatment

- ACTH: this is effective in about 50-65% of cases. It involves a daily intramuscular (IM) injection. The precise dose is not established, although it seems that higher doses may have more side-effects with no increased efficacy.
- Prednisolone may be useful due to its low cost, ready availability, ease of administration, and growing evidence that it may be similar in efficacy to ACTH and vigabatrin.
- Vigabatrin: this has a success rate of about 50%; however, vigabatrin is the treatment of choice in tuberous sclerosis.

Second-line treatment

This includes:

- Benzodiazepines, valproate, lamotrigine, topiramate or zonisamide.
- In a few patients, resection of part of the brain can give relief, especially in tuberous sclerosis.
- A ketogenic diet has shown good outcomes in some studies but the role of ketogenic diet in the management of infantile spasms has not been established.

Prognosis

This very much depends on aetiology. An attempt has been made to get unified case definitions and outcome measures in studies of infantile spasms and West's syndrome. Neurodevelopmental outcome usually tends to be poor. The prognosis for idiopathic infantile spasms tends to be better than for symptomatic infantile spasms.

Early detection and prompt effective treatment have been shown to improve neurodevelopmental outcomes, especially in idiopathic cases. Those with the idiopathic form usually do better than those whose condition is symptomatic. However, the prognosis of infantile spasms in children with Down's syndrome and neurofibromatosis type I, is relatively good.

In a follow-up of 25-35 years, one third of the patients died, the intellectual outcome of the remaining patients was normal or slightly subnormal and between one quarter and one third of the patients were seizure-free.

Further reading & references


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