**Epileptic Spasms**

"Epileptic spasms" (also known as West Syndrome) is an age related epilepsy syndrome characterized by epileptic spasms, and chaotic EEG abnormalities, associated with modification of behaviour and cognitive decline.

ILAE Commission on Classification and Terminology (2010) classified this group as Epileptic Spasms because spasms may continue past or even occur de novo after infancy.

Epileptic spasms may be classified as focal, generalized or unknown.

It is the prototype of an epileptic encephalopathy - a condition in which epileptic abnormalities themselves contribute to the progressive disturbance in cerebral function.

There can be a profound impact on neurological development, particularly cognition and behaviour.

**DEMOGRAPHICS**

Prevalence: 0.25-0.4 / 1000
Age of onset: Peak age: 3-7 months, 90% present in first year, rare after 18 months of age
Genetics: There are rare single gene epilepsies. Genetic consultation is required to explore this aetiology.
Aetiology: The etiologies in about 50% of cases are prenatal (malformations, intrauterine insults, neurocutaneous syndromes - including Tuberous Sclerosis, metabolic and genetic disorders)

Most common causes/associations are:
- Hypoxic ischaemic encephalopathy (10%), genetic (8%), Tuberous Sclerosis (7%), other cerebral malformation (8%), stroke, including porencephaly (8%), periventricular leukomalacia (5%).
- **Tuberous Sclerosis:** Epilepsy is the most common neurological symptom of Tuberous Sclerosis (80-90%), with frequent presentation of Infantile Spasms in first year of life (33%).
- **Hypoxic Ischemic Encephalopathy (HIE):** Prolonged depression of EEG over 21 days of age in term or near-term infants with HIE is a valuable predictor of later development of Infantile Spasms.
- **Mitochondrial disorders:** Spasms are the most common presenting seizure type in children with probable and definite mitochondrial disease. In unexplained Infantile Spasms, particularly with developmental delay, investigate for mitochondrial disease.

**SYMPTOMS AND SIGNS**

Seizure Semiology
- Epidemic spasm: Sudden, bilateral and symmetric contraction of muscles of neck, trunk, and extremities. The type of seizure depends on muscles (flexor or extensor) affected and on extent of contraction. The intensity of contractions and number of muscle groups involved vary considerably.
- The episodes are grouped in series or clusters, 5-30 secs apart, mainly on awakening or during transition from NREM to REM sleep.
- Asymmetric spasms may occur.
INVESTIGATIONS
Tailor investigations depending on the clinical phenotype.

EEG:
- Classic hypsarrhythmia: Very high voltage asynchronous, random and independent spike and sharp wave discharges with periods of electro-decrements. Discharges are worse in NREM sleep.
- Modified hypsarrhythmia: Focal or asymmetric discharges, episodes of voltage attenuation and some inter-hemispheric synchronization.

There is no difference in prognosis or treatment with these EEG patterns.

Neuroimaging: MRI/MRS

Genetic: chromosomes, CGH array, and when indicated, gene panel and next generation sequencing.

Ophthalmological evaluation

Hearing assessment

TORCH screen

Metabolic:
- It is important to treat treatable conditions (B12, copper, UMS, CSF glucose)
- First line investigations include Urine metabolic screen, biotinidase, Vitamin B12, Lactate, ammonia, LFTs.
- Second line investigations (often in consultation with a Paediatric Neurologist) include copper, transferrins, CSF analysis including glucose, aminoacids, lactate, neurotransmitters (if clinically indicated), Urine P6C, plasma amino acids
- Glucose transporter disorder has been reported as a cause of epileptic spasms.

PROGNOSIS
A population based, 10 yr follow-up observational study of 18 infants (Trevathan et al., 1999), found:
- 80% of 10 year olds had developmental delay, often severe
- 40% - cerebral palsy
- 94% - active epilepsy, with 50% Lennox Gastaut (other series 15-20%)
- 15% - died before 11 years

In addition, others have shown:
- There is a very high incidence of neuropsychiatric difficulties, and
- There is significant increase in Autistic Spectrum Disorder.

Poor prognostic factors:
- Adverse neonatal history
- Symptomatic aetiology
- Partial seizures
- Asymmetric EEGs
- Age of onset <4months
- Seizures preceding spasms (outside neonatal period)
- Delayed development prior to onset of spasms
- Time delay in treating > 1 month

MANAGEMENT OPTIONS
Aim for:
Cessation of seizures
Resolution of hypsarrhythmia on EEG
Preserved development
Short lag time to treatment leads to better long-term developmental outcome

**Treatment options:**
- Hormone (ACTH / prednisolone)
- Vigabatrin:
  - Considered by many as preferred treatment in Tuberous Sclerosis group.
  - Monitor for visual field defects – Electro retinography (ERG) at base line and every 3 months
  - If patient fails one option, try other (e.g. if patient fails Vigabatrin try steroids and vice versa)

**UK Infantile Spasm Study (UKISS) (2004):**
- Multi centre study compared the treatment effects of Vigabatrin with hormonal treatment (prednisolone and tetracosactide) in infantile spasms
- Cessation of spasms by 2 weeks was more likely with hormonal treatment than vigabatrin
- Generally, development and epilepsy outcomes were not significantly different between the two treatment groups at 4 years of follow up
- Better development was seen at 14 months and 4 year follow up in those with no identified aetiology allocated hormonal treatment.

Patient may respond to one or other. If above fails – there is no data to suggest one drug is superior.

**ICISS: Follow-up to UKISS (2017):**
- Hormonal treatment with vigabatrin is significantly more effective in stopping infantile spasms and resolving EEG than hormonal treatment alone.

**Other options include:**
- Ketogenic Diet - >50% improvement of spasms at 1-2 years
- Consider surgical options in appropriate candidates with intractable spasms
- Other suitable antiepileptics (many other drugs have been tried but are not first line treatment and are used when other options fail).

**DIFFERENTIAL DIAGNOSIS:**
- Other non epileptic events eg: shudders.
- Benign myoclonus of early infancy:
  - Spells begin in less than 1 year, self-limited
  - Clusters of head, trunk or extremity spasms, eye-blinking, brief jerking of upper extremity or trunk, and head nodding episodes
  - EEG invariably normal, neurologic development is not affected
  - Complete resolution 2 weeks to 8 months after onset.

This page was created in March 2012 and last modified in December 2017.

**REFERENCES:**


