Epilepsy with Myoclonic-Atonic Seizures (Doose Syndrome)

Myoclonic-atonic seizures are the defining feature.

Frequent tonic seizures as the main seizure type is not usual in this syndrome.

The syndrome is considered an epileptic encephalopathy.

The EEG usually has a normal background particularly at onset of syndrome, with frequent generalised discharges of 2 to 3 Hz spike/polyspike waves.

Non-convulsive status epilepticus is a frequent feature.

Up to half of patients have a good outcome despite early refractory symptomatology.

Treatment usually involves high dose valproate in combination with other anticonvulsants.

Consider early referral to Paediatric Neurology.

DEMOCRAPHICS

Prevalence: ~1% to 2% of all childhood epilepsies.
Age at Onset: 6 months to 6 years, peak 2-4 years.
Genetics: There is often a strong family history of epilepsy including other epilepsy syndromes. A proportion of these patients are SCN1a positive. SLC2A1 has also been found (Glucose Transporter gene)

SYMPTOMS AND SIGNS

Seizure Semiology: Myoclonic astatic seizures are the defining symptoms (100%), manifesting with symmetrical myoclonic jerks immediately followed by loss of muscle tone (atonic component). They cause lightning-like falls, head nodding, or bending of the knees.
- More than half of patients have brief absence seizures, often together with myoclonic jerks, facial myoclonias, and atonic events.
- Atonic and absence seizures are frequent and sometimes many occur each day.
- **Tonic seizures are not a major feature.**
- Non-convulsive status epilepticus for hours or days affects 1/3 of patients.
- In 2/3's of patients, febrile and non-febrile generalised tonic clonic seizures appear first, several months prior to myoclonic astatic seizures.
- The seizure frequency can become very severe and sometimes refractory to treatment. However, despite this, the outcome for most is favourable.

Neurological and Mental State: Normal prior to the onset of seizures.

INVESTIGATIONS
EEG:
- Usually normal background particularly at onset of syndrome, with frequent generalised discharges of 2 to 3 Hz spike/polyspike wave. With time background may show generalised slowing.
- Epileptogenic activity increases in sleep.
- Photic stimulation may trigger generalised spike/wave or myoclonic astatic seizures.
- Non-convulsive status leads to continuous or discontinuous and repetitive 2 to 3 Hz spike/wave.

Neuro Imaging:
- Symptomatic causes need to be excluded.

DIFFERENTIAL DIAGNOSIS
- Lennox Gastaut Syndrome.
- Atypical childhood epilepsy with centrotegmental spikes
- Dravet syndrome

PROGNOSIS
- Half of patients (probably with the idiopathic form) achieve seizure freedom and normal development.
- The others (probably symptomatic cases) continue with seizures, severe impairment of cognitive functions, and behavioural abnormalities.
- Ataxia and motor linguistic disturbances may emerge.

MANAGEMENT OPTIONS
- Early referral to paediatric neurology.
  - Sodium valproate.
  - Ethosuximide.
  - Lamotrigine.
  - Levetiracetam.
- Refractory cases may respond to the ketogenic diet.

DISCUSSION WITH FAMILY
- Safety
- Epilepsy Medical Record
- Potential of performing baseline educational assessment (through school counsellor)
- Drug Handout

LINK TO USEFUL RESOURCES
- Epilepsy Action (UK) have information for Parents on Epilepsy with Myoclonic Astatic Seizures (Doose Syndrome)

This page was created in March 2012 and last modified on October 24th 2014.

REFERENCES: