

Myoclonic epilepsy syndromes

Ingrid SCHEFFER *PhD FRACP*

Epilepsy Research Centre, Austin Health, University of Melbourne, Australia

Myoclonic seizures are the hallmark seizure type in an increasing number of epilepsy syndromes. It is important to diagnose specific myoclonic epilepsy syndromes in order to optimise investigations and management, and to provide prognostic and genetic counselling. Specific electro-clinical patterns are determined by age of onset, seizure types, developmental changes and EEG abnormalities.

Myoclonic epilepsy syndromes range from severe epileptic encephalopathies to relatively benign seizure disorders such as benign myoclonic epilepsy of infancy. The distinctive pattern of severe myoclonic epilepsy of infancy, or Dravet syndrome, begins at around 6 months of age with febrile convulsive status epilepticus which may be generalized or hemiclonic. Other seizure types evolve between 1 and 4 years. Early developmental milestones are normal until one year when development plateaus and regression occurs. The outcome is usually poor. Generalized spike wave is seen on EEG and often photosensitivity. The majority of patients with severe myoclonic epilepsy of infancy have

recently been shown to have severe mutations of *SCN1A*, the gene encoding the alpha 1 subunit of the sodium channel.

Severe myoclonic epilepsy of infancy can be differentiated clinically from Myoclonic-Astatic Epilepsy of Doose which carries a less severe prognosis. The onset of the seizure disorder is less aggressive and the child usually has myoclonic-astatic drop attacks. The prognosis of this condition is variable. Distinction from Lennox-Gastaut syndrome, where myoclonic seizures may also occur, is important.

In adolescence, the syndrome Juvenile Myoclonic Epilepsy is common and relatively benign. In more severe cases, distinction from the Progressive Myoclonus Epilepsies may need to be considered. Moreover, myoclonic elements are central to the diagnosis of rare absence epilepsy syndromes such as Myoclonic Absence Epilepsy and Absences with Eyelid Myoclonias. Epilepsy with myoclonic seizures is usually genetic in origin. Understanding of the genetics of these disorders is progressing rapidly.