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Early childhood myoclonic epilepsy: An independent genetic generalized epilepsy with myoclonic seizures as the main seizure type

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HIGHLIGHTS

- We suggest early childhood myoclonic epilepsy (ECME) as an independent epileptic syndrome.
- Seizure types included myoclonic seizures, generalized tonic-clonic seizures and/or absences.
- Seizures were in remission before adolescence with normal development or mild deficits at the most.

ABSTRACT

Objective: To elucidate the characteristics of the myoclonic seizures alone, or predominant myoclonus combined with generalized tonic-clonic seizures (GTCS) and/or absences, in early childhood, and discuss its classification.
Methods: Forty-two children were retrospectively recruited between January 2006 and June 2015.
Results: The mean age of seizure onset was 40.5 months. They were divided into 4 groups: myoclonic seizures alone; predominant myoclonus combined with GTCS; predominant myoclonus combined with absences; predominant myoclonus combined with both GTCS and absences. Interictal EEG showed generalized spike- or polyspike-wave discharges at 2–4 Hz. Seizures were controlled in 22 patients at a mean age of 60.5 months. The psychomotor development was normal (30/37) or mildly delayed (7/37).
Conclusions: We reported a cohort of patients with early childhood myoclonic epilepsy (ECME), with the following characteristics: Seizures started below 5 years old in otherwise normal children; Seizure types included myoclonic seizures alone or combined with GTCS and/or absences; Febrile or afebrile GTCS might appear firstly; Interictal EEG showed generalized spike- or polyspike-wave; Seizures usually were in remission before adolescence with normal development or mild cognitive or behavioral deficits in most.
Significance: ECME might be an independent epileptic syndrome not established by International League Against Epilepsy (ILAE) previously.

1. Introduction

Genetic (idiopathic) generalized epilepsies (GGEs) constitute one-third of all epilepsies (ILAE, 1989; Engel and ILAE, 2001). They are genetically determined and affect otherwise normal individuals of both sexes. GGEs manifest with absences, myoclonic jerks or generalized tonic-clonic seizures (GTCS), alone or in varying combinations and severity (Panayiotopoulos, 2005). According to the age at onset, GGEs proposed by International League Against Epilepsy (ILAE) include: myoclonic epilepsy in infancy (MEI), epilepsy with myoclonic absences (MAE), epilepsy with myoclonic atonic seizures also named as Doose syndrome, childhood absence epilepsy (CAE), juvenile absence epilepsies (JAE), and juvenile myoclonic epilepsy (JME) and so on (Berg et al., 2010). Among them, absence seizures are the cardinal presentations of CAE and JAE, which appear in childhood and adolescent respectively. Absence seizures with onset in the first 3 years of life were reported and

Abbreviations: CAE, childhood absence epilepsy; ECME, early childhood myoclonic epilepsy; EOAE, early onset absence epilepsy; GGEs, genetic (idiopathic) generalized epilepsies; GTCS, generalized tonic-clonic seizures; ILAE, International League Against Epilepsy; IPS, intermittent photic stimulation; JAE, juvenile absence epilepsies; JME, juvenile myoclonic epilepsy; MEI, myoclonic epilepsy in infancy; MAE, epilepsy with myoclonic absences; PME, progressive myoclonus epilepsies; VEEG, video-electroencephalogram.

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were named as early onset absence epilepsy (EOAE) (Fernandez-Torre et al., 2005; Shahar et al., 2007), filling the blank of possible GGE with absences during infancy. Myoclonic seizures are the most important seizure type in two GGEs: (1) MEI has its onset between the age of 6 months and 3 years; (2) JME is onset at age of 5–16 years and myoclonic jerks usually appear around the age of 14–15 years (Panayiotopoulos, 2005). However, little attention was paid to the patients with predominantly myoclonic seizures but seizure started at early childhood, particularly between the age of 3 and 5 years. In addition to well-described GGEs of early childhood that have myoclonic jerks as a predominant seizure type, some other seizure types were indispensable, such as myoclonic atonic seizures for Doose syndrome, and myoclonic absence seizures for MAE (Panayiotopoulos, 2005).

In our clinical practice, we noticed that a number of young children were presented by generalized seizures including myoclonic seizures alone or predominant myoclonic seizures combined with GTCS and/or absence seizures. However, the clinical features such as the seizure onset age and seizure types prevented them to be included into any kind of epileptic syndromes named by ILAE. In this study, we report such a cohort of young children, analyze the electroclinical characteristics of their epilepsy, and then discuss its classification in the current electroclinical syndromes.

2. Patients and methods

This study was approved by the Ethical Committee of Peking University First Hospital. The individuals' parents had given written informed consent to publish this case details. All data were analyzed anonymously.

Forty-two children were retrospectively recruited from approximately 42,814 video-electroencephalogram (VEEG) recordings monitored in our hospital between January 2006 and June 2015 (Fig. 1), and had the following criteria: (1) Normal development before onset of epilepsy. (2) Onset of the seizures before 5 years old. (3) Myoclonic seizures alone or predominant myoclonic seizures combined with GTCS and/or absence seizures throughout the disease course. The occurrences of GTCS were confirmed by careful history-taking from their caregivers and at least two occurrences with a loss of consciousness and subsequent postictal sleep. The myoclonic seizures and/or absence seizures were identified by VEEG with synchronous polyelectromyography monitoring. (4) Presence of interictal generalized spike- or polyspike-wave discharges, without consistent focal spikes. (5) No evidence of structural or metabolic etiology. Brain magnetic resonance imaging (MRI), biochemical studies including lactate, pyruvate, and ammonia were normal in all patients. Plasma amino acids and urine organic acids or mitochondrial genome analysis detected in some patients were all normal. (6) Exclusion of established epileptic syndromes with myoclonic seizures was based on the definitions built by ILAE. The exclusion of MEI was based predominantly on the presence of other seizure types except for myoclonic seizures or seizure onset over 3 years old. Febrile convulsion and preceding afebrile GTCS before the onset of myoclonic seizures and then replaced by myoclonic seizures were not excluded from MEI (Ito et al., 2012). Exclusion of Doose syndrome was based mainly on

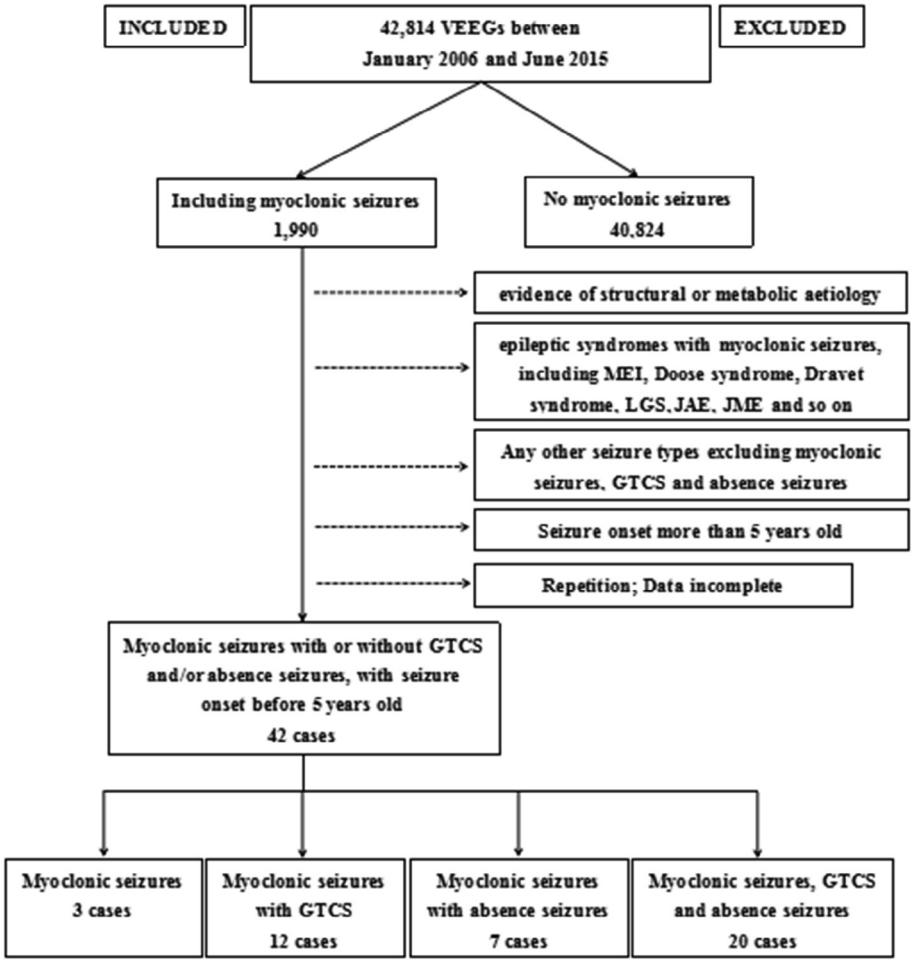

Fig. 1. The screening diagram of 42 patients included in the study. MEI: myoclonic epilepsy in infancy; LGS: Lennox-Gastaut syndrome; JAE: juvenile absence epilepsies; JME: juvenile myoclonic epilepsy; GTCS: generalized tonic-clonic seizures.