Reflex Myoclonic Epilepsy of Infancy: Seizures Induced by Tactile Stimulation

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Myoclonic epilepsy with reflex seizures in infancy is an extremely rare condition, in which seizures are provoked mainly by auditory or auditory-tactile stimuli. To increase the awareness of pediatricians regarding this underrecognized condition, we describe a child with seizures provoked only by the tactile stimulation of specific areas of the head and face. *(J Pediatr 2016;■:■-■).*

Myoclonic epilepsy in infancy (MEI) was initially described in 1981[1] and, even though several reports appeared afterward, no triggering factors were noted except photosensitivity in some subjects.[2] A variant with reflex seizures (reflex myoclonic epilepsy of infancy [RMEI]) was reported and described as a separate condition several years later.[3] These conditions are both classified as MEI.[4] In the reflex variant, myoclonic seizures may be provoked more frequently by auditory stimuli or by the combination of both auditory and tactile stimuli. However, some cases with sensitivity to tactile-only stimulation have also been reported in the literature.[5-7] We describe a child who exhibited RMEI with both spontaneous seizures during drowsiness and sleep, and reflex seizures during wakefulness and sleep. The reflex seizures were provoked only by the tactile stimulation of the vertex of the head and of the midline zones of the face, but there was no response to auditory stimuli.

Case

A 15-month-old boy presented with a 3-month history of episodes of diffuse myoclonic jerks. These episodes were triggered by the unexpected tactile stimulation of the vertex of the head. Myoclonia were more evident in the eyelids, neck, and upper limbs, causing loss of objects from the hands. However, these movements sometimes also involved the lower limbs, leading to the child's falling to the ground. He was the only child of unrelated healthy parents. Family history was unremarkable. Psychomotor development had been completely normal and no interruptions in the acquisition of developmental milestones or regression were reported.

At the first evaluation, general and neurologic examinations were normal. The Bayley Scale of Infant and Toddler Development (Bayley-II) showed fully normal scores (Mental Developmental Index = 93; Performance Developmental Index = 105). An ophthalmologic evaluation including the fundus oculi was unremarkable as were the electrocardiogram, echocardiogram, and abdominal ultrasound scan. Blood screening, array comparative genomic hybridization, and neurometabolic investigations (blood lactate and ammonia, plasma and urine amino acids, urine organic acids, and serum acylcarnitine profile) were normal. Brain magnetic resonance imaging was normal.

Repeated polygraphic video-electroencephalograms (EEGs) showed normal background activity with sporadic interictal epileptic anomalies in the frontocentral regions. Furthermore, generalized spike-wave and polyspike-wave discharges with anterior predominance were recorded, both spontaneously in drowsiness and sleep, as well as with tactile stimulation in wakefulness and sleep (**Figures 1** and **2**). Spontaneous generalized discharges were not always accompanied by clinical manifestations. The seizures could vary in intensity from time to time, being mild (with only blinking or head nodding or drop) or severe (with upward-outward movement of the upper limbs and sudden projection or loss of objects from the hands and, sometimes, flexion of the lower limbs with falls to the ground; **Video**; available at www.jpeds.com). The only type of tactile stimulation provoking reflex myoclonic seizures was the sudden tapping of the mouth, nose, glabella, and vertex of the head. Both acoustic stimuli and tactile stimulation of other parts of the body (eg, masseter muscles, hands, or feet) were evaluated and resulted in no response. Only one time during follow-up intermittent photic stimulation evoked a photoparoxysmal response (type I and III),[8] without any clinical correlate.

Antiepileptic therapy with sodium valproate was proposed, but not started because of parental refusal. Both reflex

EEG	Electroencephalogram
MEI	Myoclonic epilepsy in infancy
RMEI	Reflex myoclonic epilepsy of infancy

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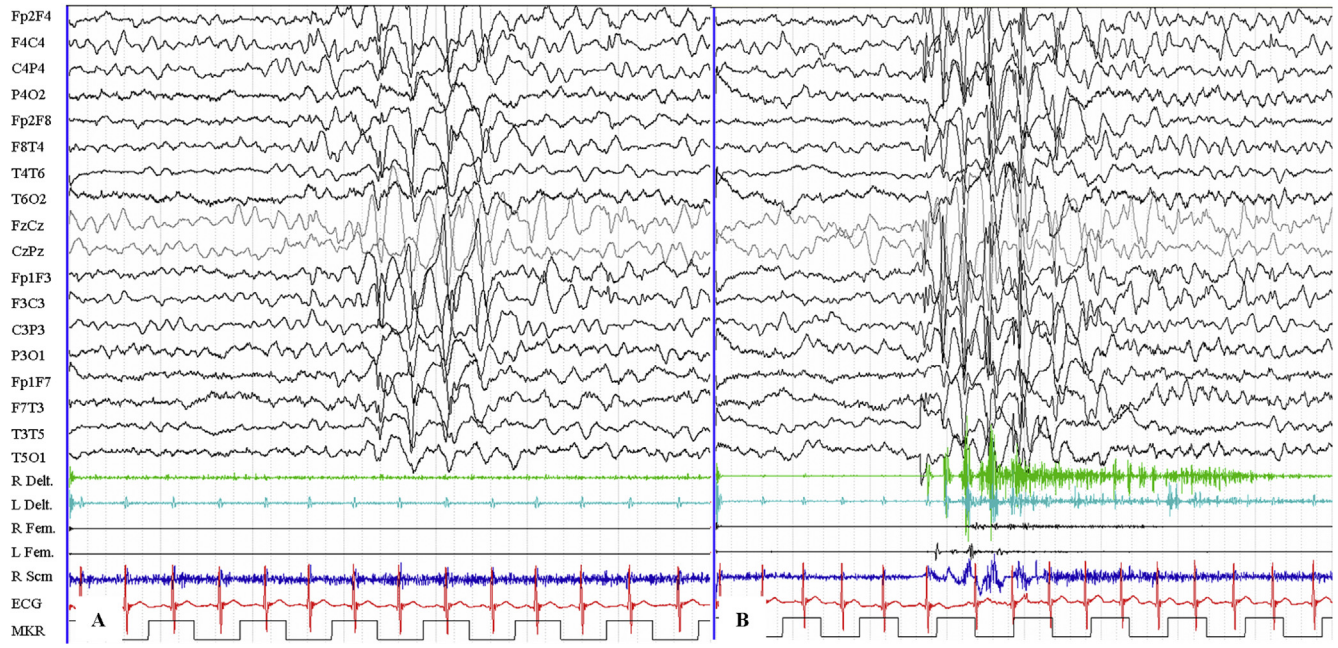

Figure 1. Spontaneous generalized spike-wave and polyspike-wave discharges with anterior predominance both in **A,** drowsiness and **B,** sleep. **B,** During sleep, the generalized discharges were accompanied by a burst of rhythmic myoclonias recorded on the 2 deltoid muscles, the right sternocleidomastoid muscle and, with a minor intensity, on the 2 femoral quadriceps muscles (bandpass filter, 1.6-30 Hz; notch, 50 Hz; sensitivity, 150 μV/cm). *ECG*, electrocardiogram; *L Delt.*, left deltoid; *L Fem.*, left femoral; *R Delt.*, right deltoid; *R Fem.*, right femoral; *R Scm*, sternocleidomastoid muscle.

and spontaneous seizures disappeared 6 months after their onset. At the last follow-up visit, performed 1 year after the onset of epilepsy, the EEG recording, in wakefulness and sleep, confirmed the absence of seizures and showed normal background activity, even with tapping of the mouth, nose, glabella, and vertex of the head. Psychomotor development was still completely normal for age.

Discussion

MEI is an idiopathic, generalized epilepsy, with a male predominance, characterized by spontaneous myoclonic seizures presenting during the first 3 years of life in neurologically and developmentally normal children.[6] A variant with reflex seizures was first described in 1995.[3] There is a debate as to whether or not MEI and RMEI are two separate conditions. RMEI seems to have an earlier onset, a better response to antiepileptic drugs, and a better cognitive outcome.[9] However, RMEI is not currently recognized as a distinct entity.[4,6,10,11] In our patient, no family history for febrile seizures or epilepsy was reported; however, a complex genetic inheritance of RMEI has been suggested, owing to the presence of febrile convulsions in some patients and to a family history of febrile convulsions or idiopathic generalized epilepsies in a significant percentage of cases.[7,9] Even though the precise pathogenic mechanism underlying this condition are unclear, an age-dependent hyperexcitability of the motor cortex may be involved.[5,6,9] Although different types of provocative stimuli are reported, including thermal, proprioceptive, visual, and photic triggers in various combination, the acoustic and tactile stimuli seem to be the most provocative.[3,10,12-15] As confirmed in our case, photosensitivity is occasionally evident on EEG, but is not a specific feature of this type of epilepsy[6,9] and sometimes does not trigger seizures. Expected or frequently repeated stimuli are not able to provoke seizures.[5,9] Although seizures with falls are possible, as seen in our patient and in other reports,[9] they are uncommon.[6,10] We further confirm that spontaneous seizures in RMEI (32% of the subjects) usually occurs after the onset of the reflex seizures and were evident only during drowsiness and sleep.[7,9]

In the present case, even though there were spontaneous seizures, epilepsy resolved without antiepileptic treatment in 6 months. This finding suggests that, because RMEI is a self-limited condition with a brief duration of seizures and fast remission of epilepsy, it is possible, at least for patients suffering from reflex-only seizures, to avoid antiepileptic therapy. Furthermore, another option would be to postpone therapy in case seizures persist for >6 months or if they become more frequent, longer, or spontaneous.[3,9,13] The association of normal psychomotor development and well-organized EEG background activity in a child with only reflex myoclonic seizures or both reflex and spontaneous ones would suggest a "reflex variant" of MEI. This is a rare and probably underestimated condition,