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A case of perioral myoclonia with absences and its evolution in adulthood?

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ABSTRACT – The rare syndrome of perioral myoclonia with absences (POMA) is described as a specific type of idiopathic generalized epilepsy in which absence seizures are accompanied by prominent perioral myoclonus as a consistent symptom. We present a 52-year-old man who was referred to our department due to treatment-resistant epilepsy. Typical seizures were described as rhythmic twitching of the lips which started at six years old, and his first convulsive seizure occurred at around 20 years old. Based on video-EEG recordings, we present two distinct EEG patterns accompanied by slight differences in clinical manifestations, which appear to be atypical of POMA. Firstly, consciousness was preserved during seizures, with no manifestation of absences. Secondly, regarding the EEG features, in some of the seizures, the perioral motor symptoms were tonic rather than myoclonic. The defining features of POMA are discussed in relation to this case.

Key words: perioral myoclonia with absences, idiopathic (genetic) generalised epilepsy, clinical features, electrographic features

The rare syndrome of perioral myoclonia with absences (POMA) was first described by Panayiotopoulos *et al*. as a different type of idiopathic generalized epilepsy in which absence seizures are accompanied by prominent perioral myoclonus as a consistent symptom (Panayiotopoulos *et al*., 1994). POMA is not listed in the 2010 ILAE classification of epileptic syndromes. Here, we present a patient with perioral motor manifestations with distinct EEG and clinical features based on video-EEG recording, without apparent loss of consciousness, and discuss these features with regards to POMA. The patient is currently resistant to treatment and the disease course appears to be progressive.

Case study

A 52-year-old (date of birth: 20.08.1964) male farmer was admitted to our department due to treatment-resistant epilepsy. Typical seizures started at around the beginning of the first school year (at six years), and were described as

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rhythmic twitching of the lips without any loss of consciousness. The patient had no treatment until he experienced his first convulsive seizure in the army at around 20 years old. Initially, sodium valproate (VPA) treatment was introduced and titrated up to 1,500 mg/day without clear benefit. Later, levetiracetam (LEV) was added to treatment and titrated up to 3,000 mg/day without a prominent change in seizure frequency. The seizure frequency increased gradually and at the time of admittance, he was experiencing clusters of perioral twitching episodes almost every day, which evolved into bilateral convulsive seizures, one to three times a month. In recent years, as an additional feature, the patient started to show a loss of consciousness during some of these perioral twitching attacks.

Although he is not sure, he may have had a febrile seizure during childhood. He was born to first-degree consanguineous parents. His grandmother may have had epilepsy. He has six siblings, two of whom are twins. One of the twins has epilepsy and the other had a single seizure. His daughter had a febrile seizure at the age of six. His past history was otherwise normal. Neurological examination and cranial MR imaging were normal.

The patient underwent video-EEG monitoring twice under antiepileptic treatment. During each EEG recording, the patient developed a cluster of seizures triggered by hyperventilation. Background activity was normal. Interictal EEG findings were brief (lasting for less than a second), fronto-centrally dominant, and generalized, with high-amplitude spike/polyspike-slow-wave paroxysms (*figure 1A, B*). Intermittent photic stimulation had no effect on epileptiform activity.

Seizure clusters lasted for a total of 13 minutes and 33 seizures were observed. EEG was examined using the standard test according to Beniczky *et al*. (2013). The patient was aware of his attacks. During seizure clusters, we observed two distinct EEG patterns

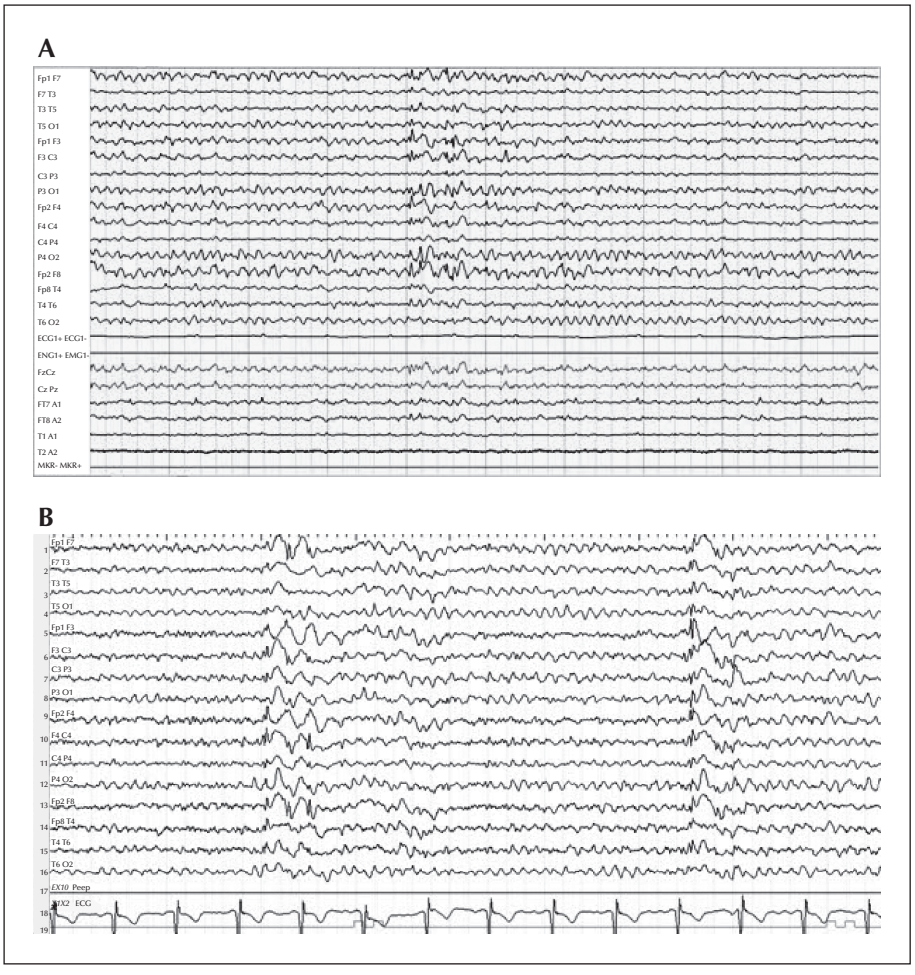

Figure 1. (A, B) Interictal EEG of the patient showing fronto-centrally dominant, generalized, high-amplitude spike-and-slow-wave discharge.

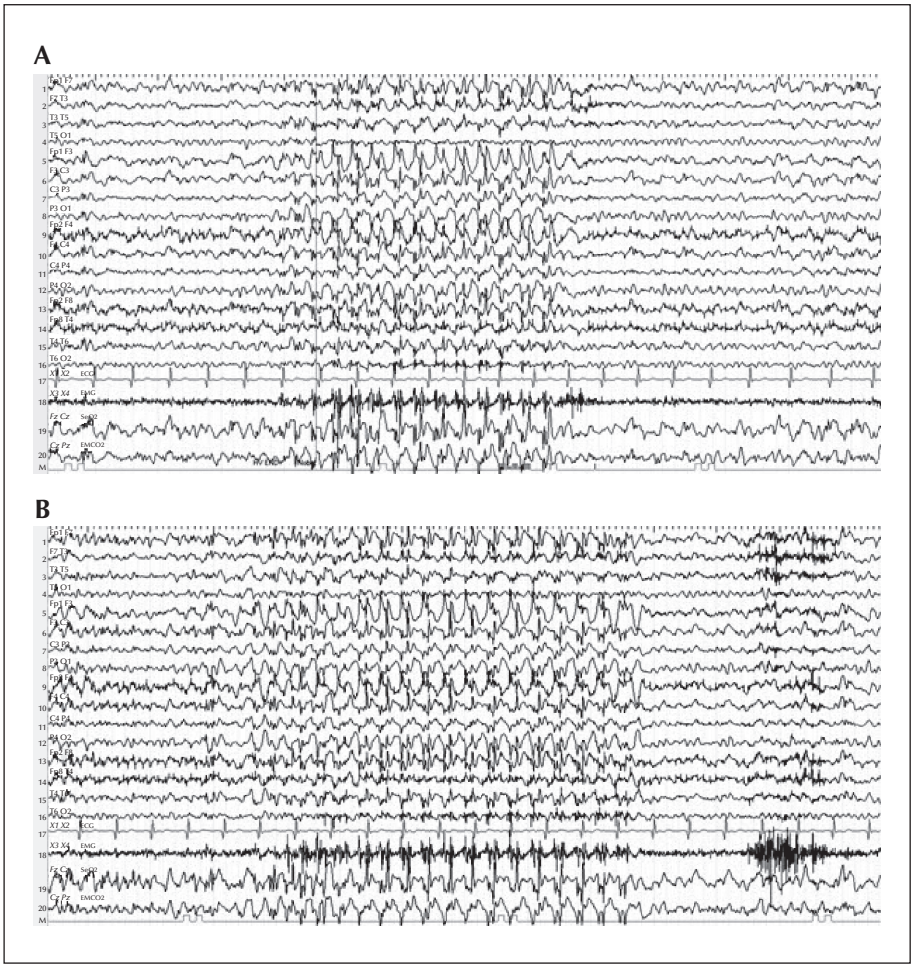

Figure 2. (A, B) Pattern 1 (P1): EEG during perioral myoclonus, showing fronto-centrally prominent, generalized, 3-4-Hz spikes/multiple spikes-and-slow-wave discharges.

accompanied by slight differences in clinical manifestations. Pattern 1 (P1) manifested as fronto-centrally prominent generalized 3-4-Hz spikes/multiple spikes-and-slow-wave paroxysms, lasting for 2-7 seconds (*figure 2A, B*). Pattern 2 (P2) manifested as trains of generalized, 10-12-Hz sharp-wave activity, lasting for 1-3 seconds (*figure 3*). The occurrence and order of these two patterns were variable, for example: P1 followed by P2 and P1 again; P2 alone; P2 then P1; or P1 then P2. The clinical presentations that occurred concomitantly with each pattern were perioral rhythmic contractions resulting in twitching consistently for P1 (perioral myoclonic seizure), and a fixed posture in the form of "*chapeau de gendarme*" (which we referred to as a perioral tonic seizure) for P2. For both P1 and P2, the patient exhibited a slight head deviation towards the right side (15 degrees). Eyes were always open or half-open during seizures. During the seizures, the patient was examined and was shown to be unresponsive; he stopped counting but restarted immediately upon cessation of the ictal pattern without any mistakes. He remembered each item that was given to him during a seizure and answered questions immediately after the seizure. Following IV administration of 10-mg diazepam, the discharges on the EEG disappeared approximately five minutes later.

The patient stated that despite the use of three antiepileptic drugs (VPA at 1,500 mg/day, LEV at 3,000 mg/day, and lamotrigine at 200 mg/day), there was no benefit at all. He was administered topiramate, at 50 mg bid, together with VPA and lamotrigine. At his last visit, he had been almost seizure-free for the previous month.

Discussion

The onset of seizures in this patient occurred in childhood, presenting with perioral motor symptoms and generalized tonic-clonic seizures which did not

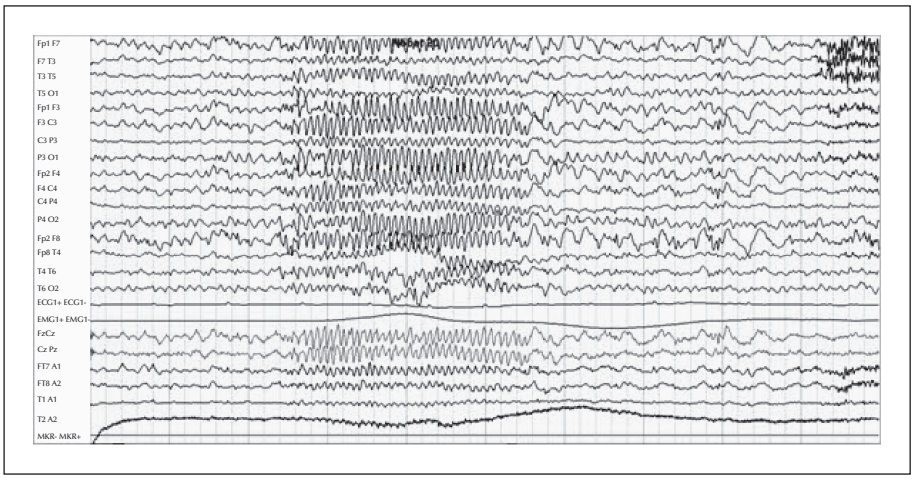

Figure 3. Pattern 2 (P2): EEG during a perioral "*chapeau de gendarme*" seizure, showing trains of generalized, 10-12-Hz sharp-wave activity.

respond to any given antiepileptic drugs, until recently. The EEG features and perioral myoclonia indicated a diagnosis of POMA. However, certain features, atypical of POMA, make this case interesting and worthy of discussion.

This patient presented with two distinct clinical and electrographic features. For EEG P1, the corresponding clinical seizure was typical of perioral myoclonia, as described in the literature. Seizures described in POMA may present as one of three forms: rhythmic contractions of the perioral muscles, rhythmic protrusion of the lips, or rhythmic jaw jerking (Panayiotopoulos, 2005). D'Orsi *et al*. studied a patient during perioral myoclonic seizures and showed that spikes corresponded to myoclonic activity of the orbicularis oris muscle (D'Orsi *et al*., 2011). Slow waves following spike activity seem to represent the inhibitory phase, leading to the typical rhythmic appearance. Our case presented with rhythmic contractions of the perioral muscles during P1, similar to earlier POMA case descriptions. On the other hand, P2 and the corresponding seizure manifestation was a tonic, fixed posture associated with the orbicularis oris muscle. The EEG correlate of this type of seizure was also different, showing trains of sharp activity but no slow waves (tonic pattern). For some of the recorded seizures, the patient developed only P2 and a corresponding "*chapeau de gendarme*" type of perioral movement. We believed this type of seizure to be the result of age or drugs. However, this remains speculative since his earlier EEG and video recordings were not available to us.

Although the patient denied any loss of consciousness during seizures until recently, he may have had phantom absences accompanied by perioral motor seizures. During the video-EEG monitoring, no loss of consciousness was observed, and the existence of absence attacks in the early phase of the disease course is debatable.

A consensus on the features of POMA is still under discussion, and the response to treatment varies (Loiseau, 1992). Additional muscle involvement, such as neck muscles, during typical seizures has been reported (Yang *et al*., 2009). The syndrome was originally defined as an absence syndrome, however, it has also been proposed to be classified as a myoclonic syndrome (Clemens, 1997).

In this case, the uncertainty of absences initially raised a question. However, given the possibility of phantom absences, we may still classify the patient as POMA. The second issue was the tonic/sustained nature of some motor manifestations which led us to consider his seizures as perioral motor seizures, instead of perioral myoclonic seizures. Based on these two distinct semiologies, the condition of this patient might be considered as a unique, as yet, unclassified condition. Alternatively, one might consider the patient's condition as POMA, if the spectrum of clinal presentations associated with POMA is expanded to include the features presented by the patient. Further detailed analysis of seizures in POMA patients may clarify this question. □

Supplementary data.

Summary didactic slides are available on the www.epilepticdisorders.com website.

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None of the authors have any conflict of interest to declare.

References

Beniczky S, Aurlien H, Brogger JC, *et al*. Standardized computer-based organized reporting of EEG: SCORE. *Epilepsia* 2013; 54: 1112-4.

Clemens B. Perioral myoclonia with absences? A case report with EEG and voltage mapping analysis. *Brain Dev* 1997; 19: 353-8.

D'Orsi G, Demaio V, Trivisano M, *et al*. Ictal video-polygraphic features of perioral myoclonia with absences. *Epilepsy Behav* 2011; 21: 314-7.

Loiseau P. Childhood absence epilepsy. In: *Epileptic Syndromes in İnfancy, Childhood and Adolescence*. Roger J, Bureau M, Dravet C, Dreifuss FE, Perret A, Wolf P. London: John Libbey, 1992: 135-50.

Panayiotopoulos CP. Syndromes of idiopathic generalized epilepsies not recognized by the International League Against Epilepsy. *Epilepsia* 2005; 46: 57-66.

Panayiotopoulos CP, Ferrie CD, Giannakodimos SE, *et al*. Perioral myoclonia with absence: a new syndrome. In: *Epileptic Seizures and Syndrome*. Wolf P. London: John Libbey & Company Ltd, 1994: 143-53.

Yang Z, Liu X, Qin J, *et al*. Neck myoclonia with absence seizures: report of 3 cases. *J Child Neurol* 2009; 24: 1026-9.

TEST YOURSELF

(1) What are the features of typical perioral myoclonia with absences (POMA)?

(2) What features make this case atypical of POMA?

(3) Describe the ictal EEG patterns of this case with regards to the clinical manifestations?

(4) Discuss the localizing value of the "*chapeau de gendarme*" sign with regards to seizures.

Note: Reading the manuscript provides an answer to all questions. Correct answers may be accessed on the website, www.epilepticdisorders.com, under the section "The EpiCentre".