CASE REPORT

Nagoya J. Med. Sci. **83**. 361–365, 2021
doi:10.18999/nagjms.83.2.361

A case of paroxysmal kinesigenic dyskinesia suspected to be reflex epilepsy

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ABSTRACT

An 11-year-old male patient developed weakness or right arm elevation after sudden movement at the age of eight. Reflex epilepsy was initially suspected; however, magnetic resonance imaging and electroencephalography (EEG) revealed no abnormality. Video-EEG monitoring was performed, but no change was noted during attacks of weakness. He was diagnosed with paroxysmal kinesigenic dyskinesia (PKD) and carbamazepine has stopped his attacks. PKD is a rare neurological disorder characterized by brief attacks of involuntary movement triggered by sudden voluntary movements, which may be confused with reflex epilepsy. PKD should be considered as a differential diagnosis of reflex epilepsy.

Keywords: Paroxysmal kinesigenic dyskinesia; reflex epilepsy; video-EEG monitoring

Abbreviations:
BFIE: benign familial infantile epilepsy
EEG: electroencephalography
MRI: magnetic resonance imaging
PKC: paroxysmal kinesigenic choreoathetosis
PKD: paroxysmal kinesigenic dyskinesia
PRRT2: proline-rich transmembrane protein 2

INTRODUCTION

Paroxysmal kinesigenic dyskinesia (PKD) is a disease characterized by involuntary movement at the initial start of movement. It frequently develops in infancy to puberty.[1] Involuntary movement can include choreoathetosis, dystonia, or ballismus; however, when choreoathetosis is primarily observed, the condition is called paroxysmal kinesigenic choreoathetosis (PKC). This disease is rare, with a reported incidence of 1 in 100,000 to 150,000 individuals. As PKD resembles reflex epilepsy, it is important to consider PKD as a disease to be differentiated. Although the differentiation method has not been established, long-term video-electroencephalogram (EEG)

Received: August 24, 2020; accepted: September 29, 2020

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may be useful to exclude reflex epilepsy.

In this study, we report a patient with PKD in whom differentiation from reflex epilepsy was required and long-term video-EEG monitoring was useful for diagnosis.

CASE REPORT

An 11-year-old boy with a complaint of involuntary movement of the right upper limb was referred to our department. His medical history was not contributory as there was no history of infantile convulsion. His elder brother had a similar symptom, although its severity was mild.

At the age of 8 years, he began to play baseball, but weakness was sometimes noted after running or quick movement. Simultaneously, elevation of the right upper limb was sometimes observed. Attacks often occurred upon standing up suddenly or touching cold water. The frequency of attacks gradually increased and similar attacks were observed every day. The boy was brought to our department.

There were no abnormal neurological findings on the initial consultation. Cephalic magnetic resonance imaging (MRI) revealed no organic abnormality (Fig. 1).

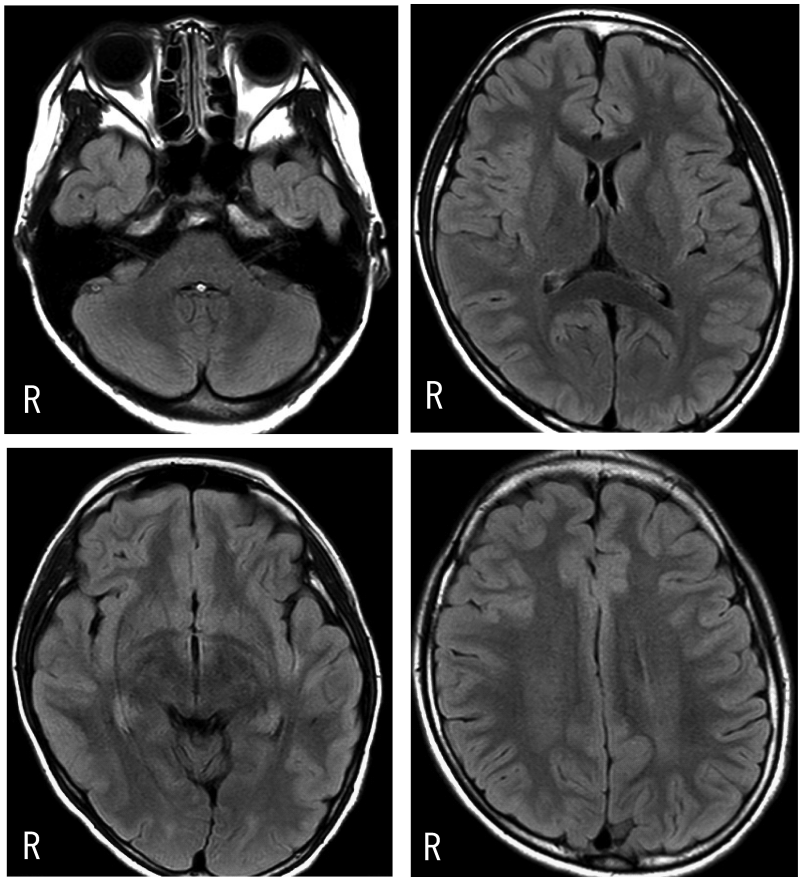

Fig. 1 Cephalic MRI

MRI showing no structural abnormality.

After admission to our department, long-term video-EEG monitoring was performed to differentiate from reflex epilepsy. There were no abnormal findings in the interval stage of attacks. During 3-day monitoring, weakness attacks were captured twice when playing a video game, but EEG at the time of attack demonstrated no abnormality (Fig. 2). Based on the EEG findings and symptoms, a diagnosis of PKD was made, excluding the possibility of epilepsy. Genetic tests on PPRT2 were not performed. The administration of carbamazepine at 150 mg/day was started, leading to the disappearance of attacks. During the 1-year follow-up, the course was favorable.

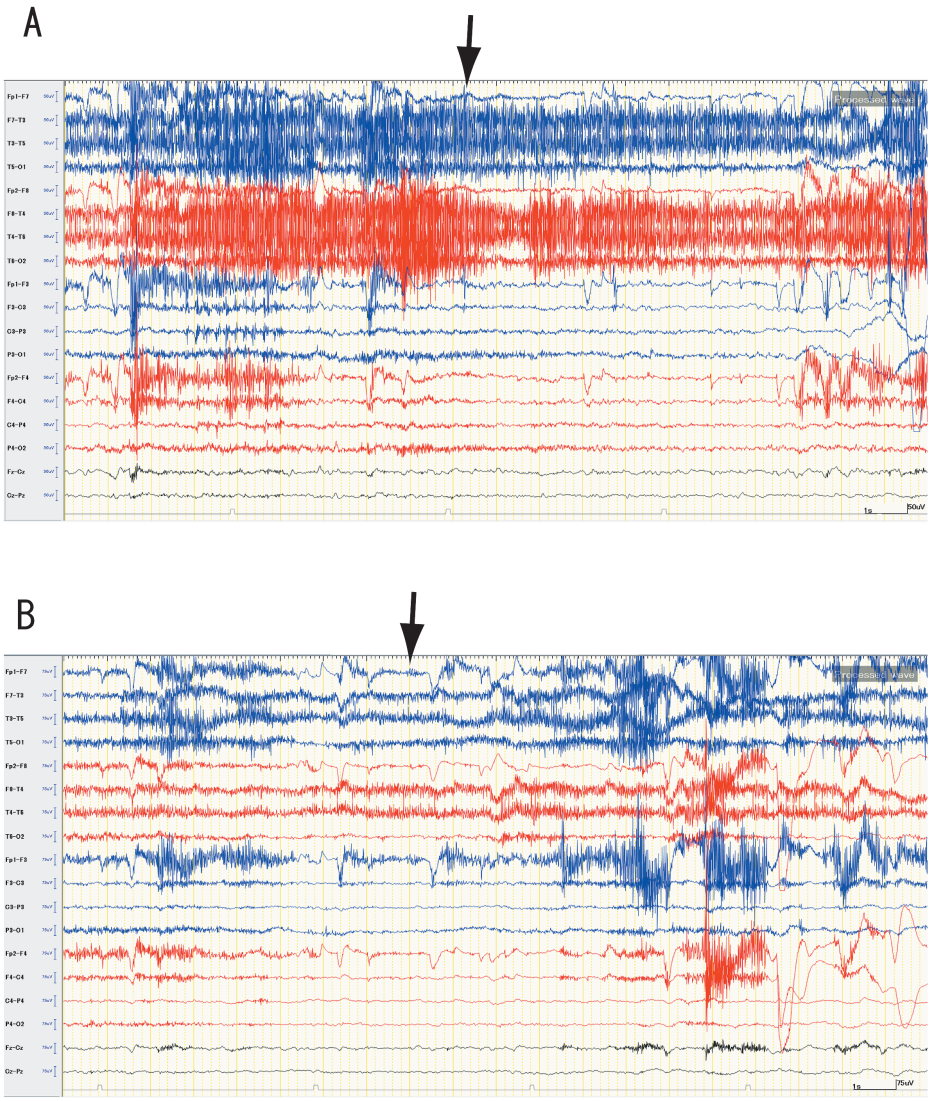

Fig. 2 EEGs at the time of attacks
Ictal EEGs showing no epileptic change at the moment of feeling weakness (arrows) in the first (A) and the second paroxysmal events (B).

DISCUSSION

Paroxysmal involuntary movement without symptoms in the interval stage of attacks is termed paroxysmal dyskinesia. Based on inducers, it is classified into 4 types: PKD, paroxysmal non-kinesigenic dyskinesia, paroxysmal exertion-induced dyskinesia, and paroxysmal hypnogenic dyskinesia.[2] Triggers for kinesigenic dyskinesia include systemic activities such as standing-up, walking, the start of running, turning over in bed, and jumping into a pool.[1] The diagnostic criteria for PKD are: 1) the sudden start of exercise induces involuntary movement (dystonia, ballism, or choreoathetosis); 2) an attack persists for ≤ 1 minute; 3) there is no loss of consciousness or pain; 4) neurological findings in the interval stage of attacks are normal; 5) a complete response to low-dose sodium channel blockers, such as carbamazepine, is achieved; and 6) the age at initial onset ranges from 1 to 20 years.[1,3] Our patient met all criteria; however, these criteria were insufficient for differentiation from partial epileptic seizures or tics. Beaumanoir et al reported that it was difficult to differentiate PKD from reflex epilepsy, and recommended exercise-induced reflex epilepsy be suspected when post-attack EEG demonstrates an abnormal finding, or in the case of a prolonged attack with loss of consciousness.[4] However, the two diseases concomitantly develop in some cases[5]; therefore, a diagnosis must be carefully made. Other diseases to be differentiated include psychiatric diseases, such as dissociative disorder and multiple sclerosis, and organic diseases such as head trauma. Furthermore, PKD is rare, leading to a delay in diagnosis in some cases. According to a single-institution study, the mean interval until a diagnosis of PKD was made was 4.8 years.[1]

The underlying mechanism of this pathophysiology has been previously discussed. Wang et al identified the proline-rich transmembrane protein 2 (PRRT2) gene as a causative gene for PKD.[6] Another study found that PRRT2 gene mutations accounted for 61.5 to 100% of patients with familial PKD and 12.5 to 50% of patients with sporadic PKD.[7] PKD (approximately 40%), benign familial infantile epilepsy (BFIE) (approximately 40%), and infantile convulsions with choreoathetosis (approximately 15%), in which BFIE is complicated by PKC, account for ≥ 95% of the PRRT2 gene-associated diseases. In addition, many symptoms, such as migraine, febrile convulsion, intellectual disability, or convulsive attacks, are concomitantly observed in a limited number of patients. Therefore, genetic tests are useful for the diagnosis of PKD, but mutations are not always present in all patients. Furthermore, some patients with PRRT2 gene mutations have epilepsy; therefore, the assessment of clinical symptoms using imaging procedures, such as MRI and EEG, are necessary for PKD diagnosis. In the present case, epilepsy and a tic were considered as disorders to be differentiated; therefore, long-term video-EEG monitoring involving EEG recording at the time of attack led to a diagnosis of PKD.

We report a patient with PKD in whom reflex epilepsy was initially suspected and video-EEG monitoring was useful for differentiation. PKD is rare; however, it is important to consider the possibility of PKD as a disease to be differentiated.

CONFLICTS OF INTEREST

There are no conflicts of interest.

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