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Case Report

Stimulus-induced reflex epileptic spasms in 5p− syndrome

Kentaro Shirai [a,b,*], Yoshiaki Saito [a], Atushi Yokoyama [c], Yoko Nishimura [a], Akiko Tamasaki [a], Yoshihiro Maegaki [a]

[a] Division of Child Neurology, Institute of Neurological Sciences, Faculty of Medicine, Tottori University, Yonago, Tottori, Japan
[b] Department of Pediatrics, Tsuchiura Kyodo Hospital, Tsuchiura, Ibaraki, Japan
[c] Department of Neurology, Shiga Medical Center for Children, Moriyama, Shiga, Japan

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Abstract

Here we describe two patients with 5p− syndrome who suffered from epilepsy characterised by stimulus-induced epileptic spasms manifesting as head nodding. In patient 1, a series of spasms were exclusively triggered by eating, and were associated with diffuse high-voltage slow waves on ictal EEG, particularly presenting as a positive slow potential at the left mid-temporal area. Clusters of sharp waves with negative polarity emerged in the same area during the inter-spasm periods during eating. In patient 2, spasms were provoked by either eating or micturition. Ictal EEG of clustered spasms after micturition showed positive slow or triphasic waves, which correlated with each spasm, over the bifrontal and vertex areas. These findings suggest that the focal cortical areas act as trigger regions in reflex epilepsies, and that a spasm-generator responsible for the execution of reflex spasms exists either in other cortical areas or in the subcortical structures.

Although epilepsy is an unusual complication of 5p− syndrome, this syndrome may have a propensity to develop reflex epilepsy, particularly epileptic spasms. However, identification of responsible genes and their roles in this phenotype requires further investigations.

Keywords: 5p− syndrome; Epileptic spasm; Reflex epilepsy; Eating epilepsy; Micturition-induced epilepsy

1. Introduction

5p− syndrome (cri-du-chat syndrome) results from the deletion of the short arm of chromosome 5, and is characterised by high-pitched cry during infancy, distinctive craniofacial dysmorphism, microcephaly and severe psychomotor retardation. Epilepsy is a rare complication; only few cases have been reported to date [1,2].

Here we report two patients with 5p− syndrome who developed epileptic spasms provoked by eating and micturition. Reflex spasm is a reflex seizure in which somatosensory stimulus from the trigeminal area or the extremities acts as specific trigger to provoke epileptic spasms [3,4]. Eating behaviour can elicit reflex spasms in rare occasions [5,6]. Herein we delineate seizure semiology and ictal EEG findings in two patients, and discuss the pathophysiology and significance of this epileptic phenomenon in 5p− syndrome.

* Corresponding author at: Department of Pediatrics, Tsuchiura Kyodo Hospital, 11-7 Manabe-shinmachi, Tsuchiura 300-0053, Ibaraki, Japan. Tel.: +81 298 23 3111; fax: +81 298 23 1160.
E-mail address: shiraken@hotmail.co.jp (K. Shirai).

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2. Case reports

2.1. Patient 1

The patient was born at 34 weeks of gestational age with a weight of 1378 g. He presented with characteristic high-pitched monochromatic cry soon after delivery, and karyotype analysis confirmed the total deletion of the short arm of chromosome 5. His psychomotor development was severely delayed since early childhood, and was equivalent to the level of a two-year-old in his twenties.

Repetitive head nodding movements were manifested during eating since the age of 12. His family did not recognise the seizures until the development of generalised convulsion with cyanosis at the age of 17. Interictal EEG during sleep showed frequent and widespread epileptiform discharges with a mid- and posterior temporal predominance. Although MRI revealed pontine and cerebellar hypoplasia, was otherwise unremarkable including the cortical structures. Seizures were refractory to medication with carbamazepine (CBZ), valproate (VPA) and nitrazepam, and appeared in clusters up to 30 times a day during breakfast or supper, and less frequently during lunch. Head nodding emerged 5 to 10 min after meal initiation, and was occasionally followed by motion arrest for several seconds. The patient could continue eating between noddings. A seizure-free period longer than six months was achieved with a regimen of VPA, zonisamide and clobazam (CLB), but seizures recurred after the reduction of CLB dosage was necessary due to an adverse effect. The patient never experienced unprovoked seizures. Video-EEG recording was conducted during breakfast at the age of 27, when monthly seizures occurred. After taking 38 spoonful of meal in 394 s, repetitive head nodding appeared. Nodding was occasionally accompanied by the flexion of the trunk and upper extremities. A total of 32 seizures were captured in 14 min, typically accompanied by a diamond-shaped surface EMG burst of the

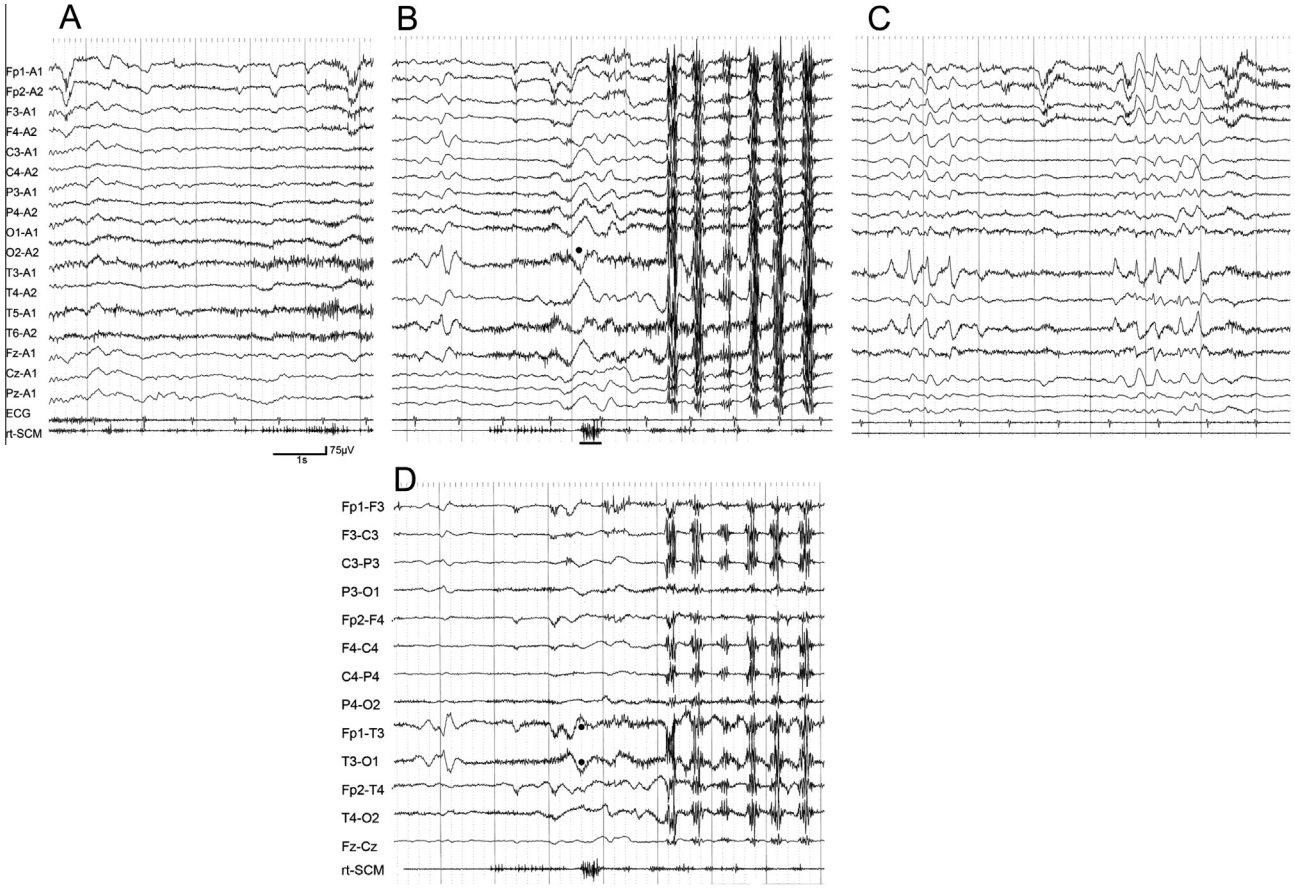

Fig. 1. Ictal and interictal EEG of patient 1 during eating. (A) Before the initiation of eating, no epileptiform discharges are observed. (B) Ictal EEG of a spasm provoked with the spoon touching the lips. Simultaneously with the head nodding, a diamond-shaped EMG burst (bar) is observed in the sternocleidomastoid muscle, concomitant with the emergence of a positive potential at the T3 electrode (dot). Note the presence of wide triphasic sharp waves with a T3 predominance on the left side of the tracing. (C) Trains of sharp waves with a T3 predominance on the interictal EEG, which emerged after the repetitive provocation of epileptic spasms. ECG: electrocardiography, SCM: surface EMG on the sternocleidomastoid muscle. (D) The same record as (B) shown in a bipolar montage. The positive slow potential in T3 appears as the "inverse" phase reversal (dots).