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Case Report

Rub epilepsy in an infant with Turner syndrome

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Abstract

We report a case of infantile refractory epilepsy associated with Turner syndrome (TS), showing very frequent, focal clonic seizures of the left upper extremity. Characteristically, in addition to spontaneous fits, her seizure was inducible by rubbing her left hand and forearm for a few seconds. Accordingly, she was diagnosed with a rare form of reflex epilepsy, "rub epilepsy". Neuroradiological investigation indicated the existence of cortical abnormalities, such as focal cortical dysplasia of the right parietal lobe. Patients with TS are reported to have neuroanatomical abnormalities, especially of the parietal lobe. Thus, our case may imply a causal relationship between potential cortical hyperexcitability of the parietal lobe and epilepsy in TS. This is the first reported infantile case of rub epilepsy, and more generally, reflex epilepsy associated with TS.

Keywords: Turner syndrome; Focal clonic seizures; Rub epilepsy; Focal cortical dysplasia

1. Introduction

Turner syndrome (TS) is a chromosomal disorder involving the loss of part or all of an X chromosome. Despite evidence showing cognitive and neuroanatomical abnormalities in TS [1], there are only a few reports of patients with TS who have epilepsy [2–4]. We report an infant with TS and refractory epilepsy, showing a rare form of reflex seizure named "rub epilepsy" [5,6].

2. Case report

The patient was the second child of healthy nonconsanguineous Japanese parents. She was born at term without complications. At 5 months of age, it was noted that she had slightly delayed motor development and had not yet achieved head control. At 8 months of age, an afebrile clonic seizure on the left side of her body occurred. She was diagnosed with epilepsy and treated with valproate. However, the frequency of seizures increased. During investigation of epilepsy, her karyotype was revealed as 45,X and she was diagnosed with TS. At 9 months of age, she showed right-handed predominance over the left. At 1.1 years of age, she was referred to our hospital for treatment of epilepsy.

Physical examination showed a decreased range of motion in bilateral elbow joints, broad fingertips, and epicanthal fold. Neurologically, she showed mild global developmental delay.

Her habitual seizure was a train of clonic movements of the left upper extremity, continuing for minutes with a maximum frequency of over 50 times daily. During the seizure, consciousness was maintained. We could not

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determine the existence of any aura preceding clonic seizures. Characteristically, soon after epilepsy-onset, her mother noticed that in addition to spontaneous fits, a similar seizure could be induced by rubbing her left hand and forearm for a few seconds. Rubbing other part of the body never induced her seizures. The magnitude of rub required to induce a seizure probably depends on epilepsy-control, as when her epilepsy was poorly controlled, even the gentle rubbing sensation of changing clothes could induce a seizure. Self-induced seizures were not observed, and single touch or startle did not induce seizures.

Interictal electroencephalography (EEG) showed no laterality in background activity and bursts of diffuse 3-Hz delta activity in both the awake and sleep states. During light sleep in the absence of any symptoms, trains of 3-Hz spike-and-wave complexes (SWC) were sometimes observed from the right centro-parietal electrode (Fig. 1A). At seizure onset, ictal EEG revealed no overt epileptic changes, but seconds later, a train of 3- to 4-Hz SWC on the right centro-parietal electrodes emerged synchronized with clonic movement of the left upper extremity (Fig. 1B). Brain magnetic resonance imaging (MRI) showed blurring of the boundary between gray and white matter in the right parietal lobe on a short-tau inversion recovery sequence (STIR) (Fig. 2A and B). Ictal [99mTc]-ethyl cysteinate dimer single photon emission computed tomography (ECD-SPECT) showed increased blood flow in this region compared with interictal SPECT (Fig. 2C and D), although this change was statistically insignificant by subtraction ictal SPECT co-registered to MRI (SISCOM). Sensory evoked potentials (SEP) induced by stimulation of the median nerve recorded at hospital admission were normal. The addition of topiramate achieved marked seizure decrease to a few times daily, and to duration of tens of seconds. Even under partial epilepsy control, rubbing her left hand and forearm

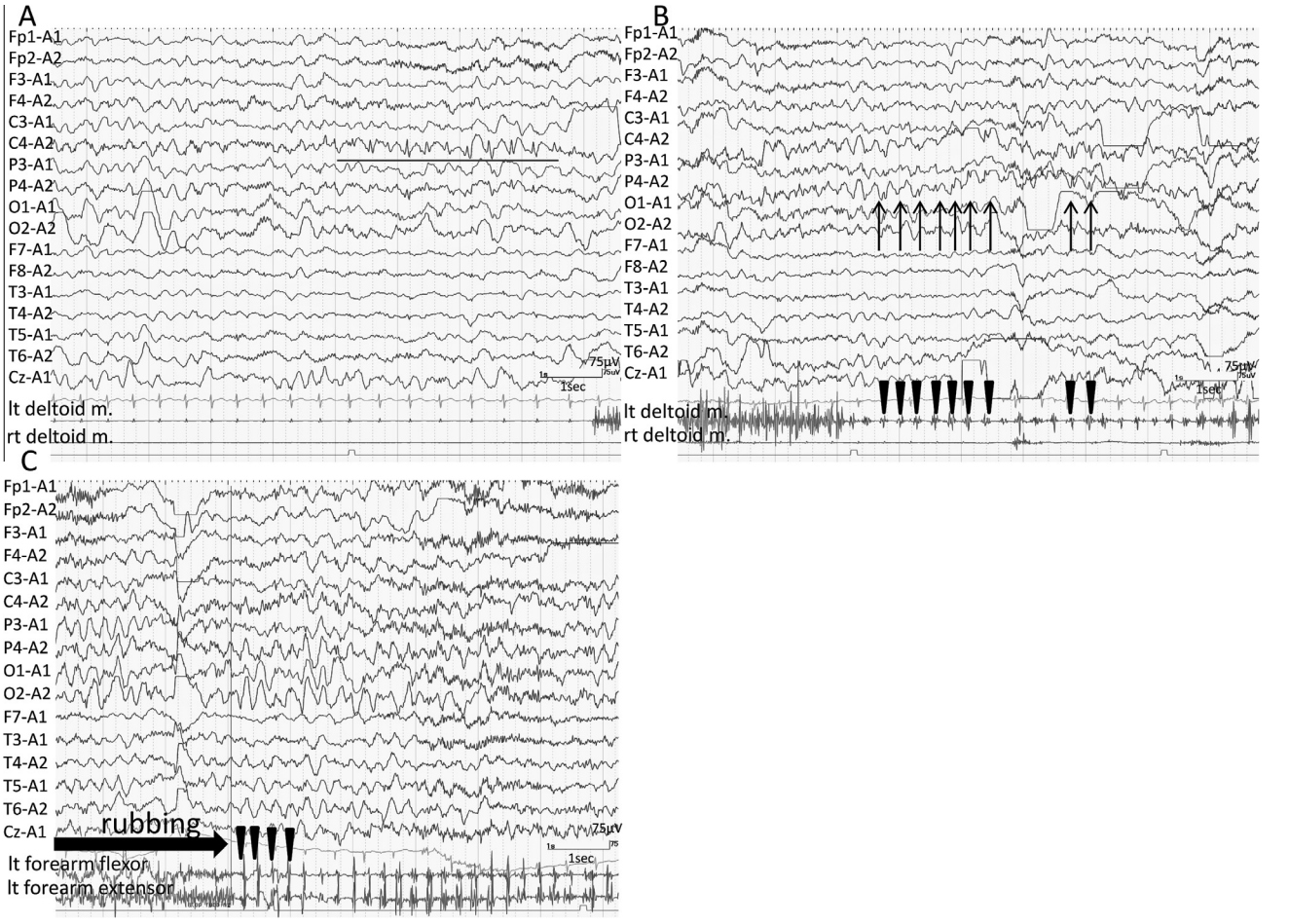

Fig. 1. Electroencephalographic findings. (A) and (B): recorded at 1.1 years of age. (C): recorded at 1.3 years of age after partially successful topiramate treatment. (A) During light sleep in the absence of any symptoms, trains of 3-Hz SWC were sometimes observed from the right centro-parietal electrode (under-bar). (B) Spontaneous seizure during the awake state. Seconds after clinical seizure onset, ictal EEG showed the emergence of 3- to 4-Hz SWC (arrows) synchronizing with clonic movements of the left upper extremity (arrowheads). (C) Induced seizure during the awake state. Rubbing her left forearm and hand for about 10 s induced her habitual seizure (arrowheads); however, possibly because of the short duration and small magnitude of the induced seizure, ictal-EEG changes did not emerge at this time.