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Case Report

Late-onset epileptic spasms in a patient with 22q13.3 deletion syndrome

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Abstract

Patients with 22q13.3 deletion syndrome present with diverse neurological problems such as global developmental delays, hypotonia, delayed or absent speech, autistic behavior, and epilepsy. Seizures occur in up to one-third of patients with 22q13.3 deletion syndrome; however, only a few reports have provided details regarding the seizure manifestations. The present report describes a patient with 22q13.3 deletion syndrome who presented with late-onset epileptic spasms (ES) and electroencephalography features like Lennox–Gastaut syndrome. An array comparative genomic hybridization analysis revealed that a chromosomal deletion of this patient included *SHANK3*. To the best of our knowledge, this is the first confirmed case of late-onset ES occur in patients with 22q13.3 deletion syndrome with a SHANK3 deletion.

Keywords: 22q13.3 deletion; Epileptic spasms; *SHANK3*; Lennox–Gastaut syndrome; Topiramate

1. Introduction

The 22q13.3 deletion syndrome, also known as Phelan–McDermid syndrome, is a genetic disorder characterized by global developmental delay, hypotonia, delayed or absent speech, autistic behavior, minor dysmorphic features, and epilepsy [1,2]. Although the clinical phenotype is variable, the neurological problems, including epilepsy, are complicated. The deletion size is correlated with some of the clinical features, including the severity of the developmental delay and ability to speak and walk, although it is not correlated with epilepsy [3].

Seizures occur in up to one third of 22q13.3 deletion syndrome patients, although there are only a few reports giving detailed seizure manifestations [2]. We experienced a 22q13.3 deletion patient presenting with late-onset epileptic spasms and whose electroencephalography (EEG) features were similar to Lennox–Gastaut syndrome (LGS). Here, we describe the seizure and EEG findings of this patient and review the literature.

2. Case report

A 5-year-old boy was monitored using video-EEG because he presented with daily head nodding. He was

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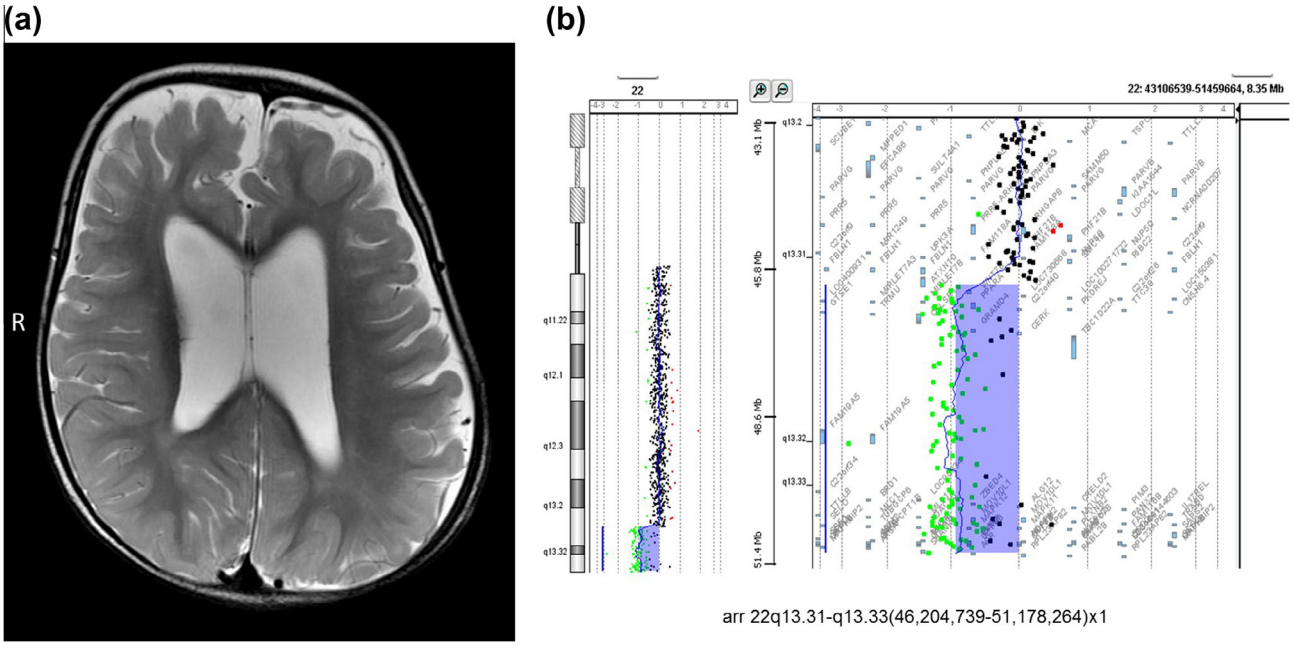

Fig. 1. (a) Head MRI at the age of 1 year showed corpus callosum thinning, reduced white matter, and enlarged lateral ventricles. (b) The array-CGH revealed the 22q13.3 deletion: arr 22q13.31–q13.33 (46,204,739–51,178,264) × 1. This region includes *SHANK3* gene.

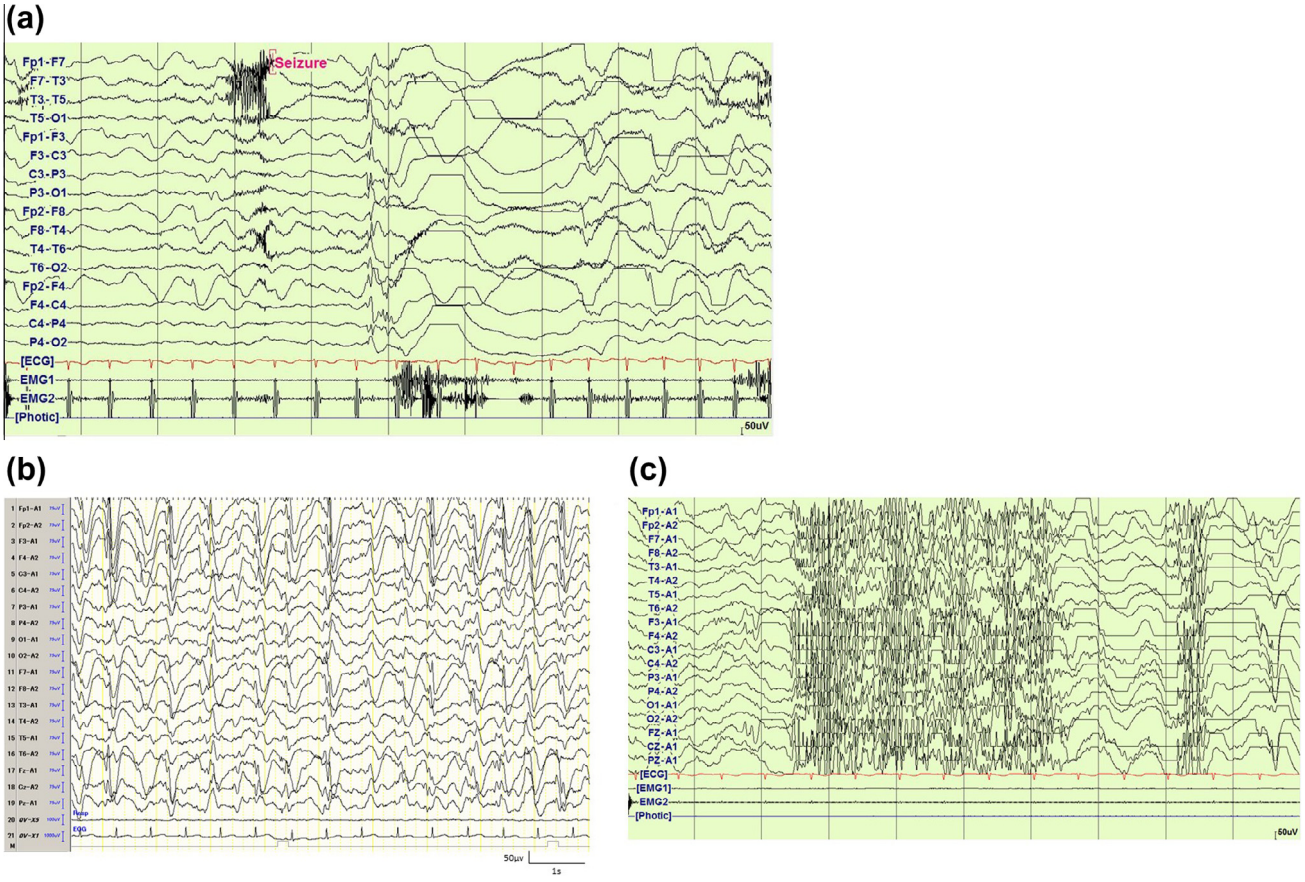

Fig. 2. (a) The ictal EEG showed preceding diffuse spikes followed by a diffuse high-voltage slow wave mixed with low-voltage fast muscle activity on the EEG corresponding to the "diamond-shaped" pattern on EMG. Subsequently, a diffuse electrodecremental pattern was observed on the EEG. EMG1 and EMG2 were recorded from the right and left deltoid muscles, respectively. (b) and (c) Diffuse slow spike–wave discharges (b) and the interictal EEG findings show paroxysmal diffuse fast activity (c).