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Case report

Longitudinal MR findings in a patient with hemimegalencephaly associated with tuberous sclerosis

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Abstract

A patient with an unusual association between tuberous sclerosis complex and hemimegalencephaly is reported. At 2 days of age, CT scan disclosed right hemimegalencephaly with periventricular faint calcification. At 1 month of age, magnetic resonance imaging showed right hemimegalencephaly with T1 and T2 shortening. The right frontal and parietal lobes were compatible with transmantle dysplasia. At 6 months of age, the right hemisphere had decreased in size. Atrophic changes continued to progress until 3 years of age. Periventricular calcification in the right hemisphere became marked with age. The patient developed frequent partial motor seizures from 6 weeks of age and infantile spasms from 5 months of age. Electroencephalogram revealed low amplitude of background activities in the right frontal area. The cause of this atrophic change in the hemimegalencephalic hemisphere, which could be a hamartomatous lesion, remains unknown.

Keywords: Hemimegalencephaly; Tuberous sclerosis complex; Longitudinal study; Atrophic change; MRI

1. Introduction

Hemimegalencephaly is a rare developmental brain malformation characterized by enlargement of only one cerebral hemisphere [1]. Hemimegalencephaly is divided into two subtypes: an isolated form and a syndromic form. The syndromic form is associated mainly with neurocutaneous syndromes, including epidermal nevus syndrome, hypomelanosis of Ito, Proteus syndrome, Klippel–Trenaunay–Weber syndrome, and tuberous sclerosis complex (TSC). The combination of hemimegalencephaly and TSC is extremely rare and only a few such cases appear in the literature [2–5]. TSC patients have extensive hamartomatous lesions in the central nervous system [6,7] and other systemic organs (heart, kidney, retina, etc.). It is known that cardiac hamartomatous lesions can decrease in size naturally over time, but this same phenomenon is not reported for hamartoma(s) in the central nervous system in TSC patients. In patients with hemimegalencephaly, reports of the large hemisphere becoming smaller with age are exceptional [5,8]. This case report details such a patient.

2. Case presentation

The patient was the first child of non-consanguineous healthy parents. There were no other cases of TSC in the family. He was born at full term after cesarean section due to cephalopelvic disproportion without asphyxia. His head circumference was 38.3 cm (+3.4 SD) at birth. Hypomelanotic macules of almost 1 cm in diameter were observed on his back. An echocardiogram revealed a hyperechoic, oval-shaped mass in the interventricular septum. This lesion was compatible with cardiac rhabdomyoma. In addition, retinal hamartomata were confirmed. According to the diagnostic criteria [9], the patient was diagnosed as having TSC. At 2 days of age he underwent computed tomography (CT) due to a large head, and this indicated the presence of a large right hemisphere, especially in the frontal lobe.

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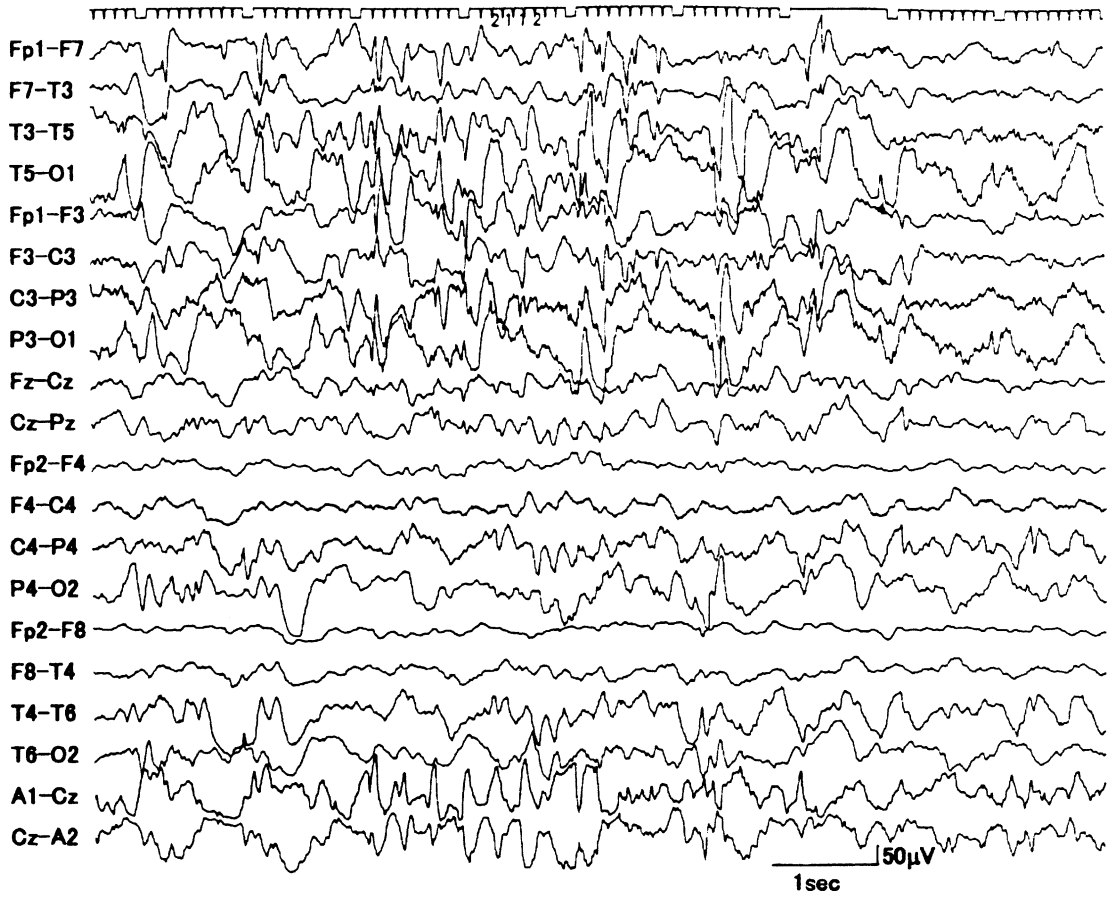

Fig. 1. EEG findings at 3 years of age. Bipolar EEG shows spike activities in the left hemisphere and low background activities in the right hemisphere, especially in the frontal area.

At 6 weeks of age, frequent partial motor seizures began to occur in his left upper extremity. At 3 months of age, the seizure predominancy changed to the right side. Carbamazepine therapy was initiated, but the seizures were not controlled completely.

By 5 months of age, brief tonic seizures with head nodding and asymmetrical extension of the extremities occurred frequently (5–10 times/day). The electroencephalogram (EEG) revealed hypsarrhythmia with left predominant spikes. He was diagnosed as infantile spasms and admitted to hospital for treatment. His head circumference was 47.5 cm (+3.0 SD). Hypomelanotic macules were noted on his cheek, back, and legs, and a shagreen patch was seen on his right cheek. Ocular fundoscopy revealed bilateral retinal phakomas. The patient could smile and pursue visually, but could not yet control his head. He showed hypotonia and a paucity of voluntary movement of the left extremities (left hemiparesis). The deep tendon reflexes were slightly exaggerated on the left side. Laboratory assessments revealed no abnormalities. Because of the epileptic foci in both hemispheres, neurosurgical therapy was not performed. Instead, antiepileptic drugs were administered, and the tonic seizures ceased after administration of valproate, clonazepam, and zonisamide. Three months after the onset of therapy, the hypsarrhythmia had disappeared on EEG and focal spikes were seen over the left parietal area.

Subsequently, partial motor seizures in the right upper extremity reappeared. EEG indicated that spike activities were seen in the left hemisphere and the background activities (BGA) were lower in the right hemisphere, especially in the frontal lobe, than the other side (Fig. 1). The patient has severe psychomotor retardation at 5 years old, in that he has no meaningful words, and he is still unable to sit or stand up, though the movement of his left extremities gradually improved and there are almost no laterality concerning the extremity movement.

3. Imaging findings

CT scans were performed at 2 days, 6 months, and 14 months of age. Magnetic resonance imaging (MRI) was done at 1 month, 6 months, 18 months, and 3 years 2 months of age.