Hemimegaloencephaly with Periventricular Heterotopia

—Case Report—

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Abstract

A 7-year-old girl was admitted with an unusual anomaly of hemimegaloencephaly associated with periventricular heterotopia manifesting as intractable seizures and mental retardation. Magnetic resonance (MR) imaging showed left hemispheric hypertrophy and left ventricular dilatation. Proton density-weighted MR imaging revealed a periventricular lesion isointense with gray matter. MR imaging is an effective method for diagnosing hemimegaloencephaly and heterotopia.

Key words: hemimegaloencephaly, heterotopia, intractable seizure, magnetic resonance imaging

Introduction

Hemimegaloencephaly is a rare congenital brain malformation in which all or part of one hemisphere is enlarged with ipsilateral ventricular dilatation in the absence of somatic hemihypertrophy. It is usually associated with abnormal differentiation of neurons and/or astrocytes, migration anomalies, and abnormal cortical organization. Histologically, there are areas of polymicrogyria and pachygyria, heterotopic neurons and foci of calcification in the subcortical white matter, an increased number of glial cells, and an increase of neuronal size. Hemimegaloencephaly include developmental delay, hemiparesis, hemianopia, and intractable seizures. Hemimegaloencephaly may be due to an abnormality in the first 4 months of fetal life, resulting in localized heteroploidy. Hemimegaloencephaly may be associated with neurocutaneous abnormalities, such as linear nevus sebaceous syndrome, of which it may be a neurological variant, hypomelanosis of Ito, Klippel-Trenaunay-Weber syndrome, or neurofibromatosis.

Heterotopia is a neuronal migration anomaly associated with both hemimegaloencephaly and intractable seizures. The lesion may be located anywhere from the subependymal region to the cortex. Generally, heterotopia is divided into the nodular form and the laminar form, but another type called "band heterotopia" is characterized by thick bands of heterotopia in the white matter of the bilateral hemispheres.

Here, we describe an unusual case of hemimegaloencephaly with periventricular heterotopia and discuss the use of magnetic resonance (MR) imaging for diagnosis.

Case Report

A 7-year-old girl was admitted to our hospital with a history of intractable seizures. She had been delivered after a normal pregnancy, but her head circumference was slightly greater at birth (38 cm). Family history was noncontributory. Her psychomotor development was gradually delayed, her neck became fixed at 3 months, and she started to walk at 16 months. Seizures occurred three times in the same month when she was aged 2 years. Computed tomography (CT) showed a slightly enlarged left hemisphere with ventricular dilatation and a slightly high-density area along the left lateral ventricle compared to the right (Fig. 1). However, no medication was given because no further seizures appeared.
Seizures occurred again when she was 7 years old and became intractable, manifesting as eye deviation to the right, clonic jerking of the right eyelid, flexion of the right upper extremity, and head rotation to the right with disturbance of consciousness. The seizures were refractory to anticonvulsant therapy (phenytoin, carbamazepine, valproic acid, and zonisamide), and occurred several times a month.

On admission, her head circumference was 52.5 cm (normal 48-54 cm), but psychomotor development was delayed (intelligence quotient 70). Neurological examination showed mild right hemiparesis. The interictal electroencephalogram (EEG) showed spikes and sharp waves in the left hemisphere, and spikes occasionally spread to the right hemisphere. T1-weighted MR imaging (1.5 T) showed hemispheric enlargement, ventricular dilatation, and a periventricular laminar area isointense with the gray matter, all on the left (Fig. 2 left). T2-weighted MR imaging also showed the left periventricular area isointense with gray matter, and pachyscal appearance in the left lower occipital lobe.
tricular area was the same intensity as that of gray matter (Fig. 2 center), as did proton density-weighted imaging (Fig. 2 right). Coronal views clearly showed hemispheric asymmetry, particularly in the occipitoparietal area. Hemispheric enlargement with lateral ventricular dilatation had caused midline shift and cerebellar compression. The ipsilateral hemisphere and cerebellum were otherwise apparently normal (Fig. 3). CT showed the same findings as previously. Cine-MR imaging demonstrated normal flow through the foramen of Monro. Based on these findings, hemimegalencephaly with periventricular heterotopia was diagnosed.

Despite administration of several anticonvulsant agents, her seizures are still not adequately controlled.

Discussion

Our patient with hemimegalencephaly demonstrated thick heterotopia along the dilated lateral ventricle, which did not correspond exactly to the reported classification. Therefore, this case was thought to be a rare type of periventricular heterotopia.

The marked abnormality of the affected hemisphere explains the high incidence of neurological abnormality in hemimegalencephaly. Seizures predominate, often starting in the neonatal period, and may become refractory to standard therapy. Several forms of developmental delay are common, and contralateral hemiparesis and hemianopsia may occur. EEG may demonstrate spike and wave complexes, burst and suppression, or hypsarrhythmia. Diagnosis of hemimegalencephaly by CT or pneumoencephalography is difficult, but MR imaging can provide greater detail. In our case, MR imaging clearly showed the hemispheric enlargement and ventricular dilatation, especially on the T1-weighted image. The proton density-weighted image is particularly useful for the diagnosis of hemimegalencephaly, gyral malformation, mental retardation, seizures, and facial hemihypertrophy. Diagnosis of hemimegalencephaly by MR imaging in five children.

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The seizures associated with hemimegalencephaly are intractable, so the prognosis is generally poor. In particular, the occurrence of seizures before age 1 month is associated with a poor outcome. However, hemispherectomy is effective for controlling seizures in patients with intractable seizures and severe neurological deficits. Ventriculo-peritoneal shunt or corpus callosotomy have not been effective for seizure control. Our patient was not treated surgically, as the neurological deficits and psychomotor delay were mild.

References


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