Magnetic Resonance Imaging Diagnosis of Focal Cerebellar Cortical Dysplasia

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ABSTRACT

Focal cerebellar cortical dysplasia is an extremely rare disease. The diagnosis could be easily missed on routine computed tomography (CT) and magnetic resonance imaging (MRI) images because the lesion is isodensity on CT images, and isointense to normal gray and white matter on MRI images. Focal distortion of the cerebellar gray-white matter can be found if MRI images are surveyed carefully. Here we report an adult case of focal cerebellar cortical dysplasia and review the literatures.

Cerebellar malformations received less attention as compared with cerebral anomalies. Only lately had their classifications been proposed by Sandeep Patel and A James Barkovich with an imaging-based classification system in 2002 [1]. They divided cerebellar malformations initially into those with hypoplasia and dysplasia, then divided into focal and diffuse malformations, and finally according to other features.

MRI provides good image quality for evaluating posterior cranial fossa anatomy and pathologic change. Therefore, cerebellar malformations could be diagnosed well by MRI study. However, because only subtle disorganized gray-white matter architecture can be detected in cases of focal cerebellar cortical dysplasia, the diagnosis can be easily missed. It is mandatory for radiologists to be familiar with its presentation and to inspect the images carefully to make correct diagnosis.

CASE REPORT

A 49 years old female patient was referred from the department of rehabilitation to our neurological out-patient-department for help. Her chief complaint was right hand tremor for many years. She had history of head injury and left humeral neck fracture during motor vehicle accident 8 years ago. Her left hand became disabled since humeral fracture then. She has become wheel chair bound since another accident resulted in right femoral fracture few months ago. Her prenatal, birth and family histories were non-contributory. Physical examination revealed muscle power R 4/0 L 3/3 with cogwheel rigidity. She presented with mild dysarthria, but not desmetria. Resting tremor and diminished deep tendon reflex (UL ++/-; LL +++/+++ ) were noted. Her sensory was impaired on the left side. The symptom/sign of diminished muscle power, diminished deep tendon reflex, and sensory impairment could be traced to several years ago in her clinical history. Upon the impression of secondary Parkinsonism, non-contrast axial brain CT scan was arranged and performed on Philips MX 8000 CT scanner with 5mm collimation.

Brain CT images reveal ill-defined isodense lesion at tonsil of right cerebellar hemisphere with compression of the 4th ventricle (Fig. 1). There is no hemorrhage, calcification,
Focal cerebellar cortical dysplasia

or other abnormal density in the brain. Bilateral temporal horns are slightly dilated. Bilateral cerebral hemispheres are unremarkable. Because of cerebellar tumor was suspected by CT images, MRI study was recommended by radiologist for further evaluation and was performed few days later.

MRI study was performed on Philips Achieva 1.5T SE with axial T1W_SE, T2W_TSE, T2W_FLAIR, DWI, T2W_FFE, sagittal T2W_TSE, coronal T2W_FLAIR, and post-contrast (Dotarem, 0.2 ml/kg i.v. Guerbet S.A.) enhanced T1W_FFE pulse sequences with reconstructed images in three orthogonal planes. Folia malformation with deformed gray-white matter at posterior lobe of right cerebellar hemisphere was noted (Fig. 2). The lesion appeared isointense to white matter on T2W, FLAIR and DWI images without abnormal hyperintensity nor contrast enhancement. Focal cerebellar cortical dysplasia was diagnosed. Tumor was excluded because the lesion showed findings of folial thickening, deformed gray-white matter junction, absent contrast enhancement. She received medical treatment for her symptoms. Her symptom/sign did not change during 4 months follow-up period.

**DISCUSSION**

Focal cerebellar cortical dysplasia is an extremely rare disease with etiology still not clearly understood. In the retrospective review study by Sandeep Patel and A James Barkovich [1], 70 patients with diagnosis of some form of cerebellar malformation were summated during 15 years. Among the cases, only two were focal cerebellar cortical dysplasia. They also proposed an imaging-based classification system of cerebellar malformations by MRI finding because limited understanding of cerebellar embryology, limited histologic studies of these disorders, and lack of a practical classification of cerebellar malformations [1]. The cerebellar malformations were initially divided into those with hypoplasia or dysplasia, then into focal or diffuse malformations. And finally, the cerebellar lesions were separated by other features, such as brain stem and cerebral involvement. In the category of cerebellar focal dysplasia, two subdivisions were defined: 1. Isolated vermian dysplasia; which includes rhombencephalosynapsis and molar tooth malformations (associated with brain stem dysplasia); 2.

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**Figure 1**

Non-contrast brain CT reveals suspicious isodense ill-defined mass lesion (white arrow) at tonsil of right cerebellar hemisphere with compression of 4th ventricle.
Focal cerebellar cortical dysplasia

**Figure 2.** MRI axial T1W a. T2W b. coronal FLAIR c. sagittal T2W d. images reveal folia malformation with deformed gray-white matter at posterior lobe of right cerebellar hemisphere.
Focal cerebellar cortical dysplasia

Isolated hemispheric dysplasia; which includes Lhermitte-Duclos-Cowden syndrome (gangliocytoma), and focal cerebellar cortical dysplasia/heterotopia. Rhombencephalosynapsis is defined as absence of cerebellar vermis with midline fusion of the two cerebellar hemispheres. Molar tooth malformations is defined as vermian hypoplasia/agenesis, fourth ventricle “bat wing” or triangular configuration and communicated with the cisterna magna, deep interpeduncular fossa, thin isthmic region, and thickened superior cerebellar peduncles that were perpendicular to the pons. Lhermitte-Duclos-Cowden syndrome is defined as sharply margined, non-enhancing focal cerebellar mass with T1 and T2 prolongation, and associated with some other anomalies and visceral hamartomas. Focal cerebellar cortical dysplasia/heterotopia is defined as focal area or disorganized architecture. The case we report here is focal cerebellar cortical dysplasia which is a mild form among cerebellar malformations.

Cerebellar cortical dysplasia had been reported in association with chromosomal abnormalities of the trisomy type, congenital muscular dystrophies and related syndromes, in intrauterine infections, in gamma-radiation, in ethanol exposition [2]. However, the pathogenesis of cerebellar cortical dysplasia is still unclear. Histopathological study reported finding of scrambled folia together with apparent fusing of apposed molecular layers and surrounding small cavities with meningeal vessels, and superficially obliterated folia pattern [2].

The abnormalities of the cerebellar cortex and folia pattern can be well depicted on MRI images. The MRI findings of cerebellar cortical dysplasia included defective, large or vertical abnormal fissures, irregular gray matter-white matter junction, lack of normal arborization of the white matter, and heterotopia within cerebellar hemisphere, all can lead to disorganized folia [3]. The nodulus, flocculus, and tonsils are common sites of cortical dysplasia, but any portion of the cortex may be affected [4]. Intracortical cystlike structures may also be noted. It may represent a more severe injury occurring during development [4]. In our case, the posterior lobe of right cerebellar hemisphere was involved. The lesion is present with folial thickening, deformed gray-white matter junction and absent contrast enhancement.

Cerebellar cortical dysplasia may be associated with other abnormality, such as vermian malformation, cerebral cortical dysplasia, dysplasia of corpus callosum, heterotopia, Dandy-Walker, Chiari II and III, and hypoplasia of brain stem [3]. The clinical presentations include seizure, apraxia, ataxia, tremor, developmental delay, hypotonia, facial deformities, and abnormalities in eyes [3]. There was no other associated anomaly of supratentorium brain or brain stem on the MRI images of our case. However, she had history of head injury, so the symptoms of tremor and cogwheel rigidity may not be completely attributable to her focal dysplastic cerebellar cortex.

In conclusion, cerebellar cortical dysplasia belongs to a spectrum of developmental anomalies of cerebellum with unknown pathogenesis. Focal cerebellar cortical dysplasia is a mild form with lesser affected part and relatively mild symptom/sign. To make exact diagnosis of focal cerebellar cortical dysplasia requires MRI study. Its findings include folial thickening, deformed gray-white matter junction, and absent contrast enhancement.

REFERENCES