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MR of Cerebellar Cortical Dysplasia

Philippe Demaerel, Lieven Lagae, Paul Casaer, and Albert L. Baert

Summary: MR imaging findings are described in four patients with cerebellar cortical dysplasia. Typically, cerebellar disorganized folia were seen as an irregular bumpy gray-white matter interface. In addition, cystlike cortical abnormalities were observed in two patients and associated supratentorial developmental abnormalities were seen in three patients. To our knowledge, cerebellar cortical dysplasia without supratentorial abnormalities, as seen in one patient, has not been reported before. We suggest that cerebellar cortical dysplasia represents a spectrum of abnormalities ranging from mild to extensive in severity.

The MR appearance of cerebellar cortical dysplasia in association with lissencephaly type II has been reported in patients with Fukuyama congenital muscular dystrophy (FCMD) and related syndromes. The present report describes the imaging appearance of cerebellar cortical dysplasia in four patients without clinical or laboratory evidence of muscular dystrophy.

Case Reports

We reviewed the clinical and MR imaging findings of four patients with cerebellar cortical dysplasia. At the time of imaging, the patients ranged in age from 5 to 21 years. All except one patient had psychomotor retardation. The two female patients were sisters. Serum creatine kinase levels were within normal limits in three patients and were not obtained in the fourth patient. There was no evidence of congenital muscular dystrophy, and no patient had a history of intrauterine infection. The clinical findings are summarized in the Table. Axial T2-weighted double-echo short-tau turbo inversion recovery (5100/145/14, 85 [TR/TI/TE]) images were obtained in the four patients. In one patient, sagittal and coronal T1-weighted spin-echo (600/14) images were obtained, and in two patients, 3-D magnetization-prepared rapid gradient echo T1-weighted (9.7/4) images were obtained.

The MR imaging findings are summarized in the Table. The cerebellar abnormalities consisted of developmental derangement of the cortical architecture, resulting in disorganized folia and a bumpy gray-white matter interface (Figs 1 and 2). Intracortical cystlike structures were seen in two patients, but they were more conspicuous in one than the other (Fig 1). The abnormalities were located in the midportion and dorsal surface of both cerebellar hemispheres. The cerebellar developmental abnormalities were best seen on the T2-weighted images. The vermis and brain stem appeared normal.

Supratentorial developmental abnormalities were seen in three of the four patients. Excessive folding of the cortical ribbon that gives the appearance of thickened gyri with shallow sulci was found in two patients, and was located in both the frontal and parietal lobes and to a lesser extent in the occipital lobe (Fig 1). In the third patient, the abnormal cortical architecture was seen in the left frontal, parietal, and temporal lobes. Two of these three patients had a dilated supratentorial ventricular system and one had white matter hyperintensities in the subcortical area. The fourth patient had no associated supratentorial abnormalities.

Discussion

Our understanding of congenital brain malformations has improved considerably since the advent of MR imaging. Subtle developmental abnormalities of the cerebral and cerebellar cortical architecture have been reported (1, 2). Cerebellar cortical dysplasia has been seen in patients with FCMD, and has also been observed in association with intrauterine infection (3–6). FCMD is a common form of muscular dystrophy in Japan, and consists of muscular hypotonia with prenatal onset, severe developmental delay, and epilepsy. Cerebellar MR findings in FCMD include a disorderly alignment of the cerebellar folia, particularly in the midportion and dorsal surface of the hemisphere, and intraparenchymal cysts near these disorganized folia. It has been suggested that these cysts might form from subarachnoid spaces that were engulfed by the fusion of disorganized folia (4). The nomenclature on focal dysplasia of the cerebellar cortical architecture is confusing. Cortical dysplasia is the generally accepted term for this condition. Polymicrogyria is occasionally used, although it is well known that there are no gyri and sulci in the cerebellum (7, 8). Heterotaxia, as originally described by Brun (9), is a general term used to describe an abnormal arrangement of organs or parts of the body in relation to one another, but it has never gained wide acceptance.

In one study, a minor degree of cerebellar cortical dysplasia was found histologically in up to 85% of 147 normal infants (10). These authors distinguished four types of minor anomalies, but acknowledged that their frequency was not representative of that of major malformations found in the cerebellum later in life. Indeed, some of these anomalies undergo involution after the ninth month of life. The cerebellar

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From the Departments of Radiology (P.D., A.L.B.) and Pediatrics (L.L., P.C.), University Hospitals, Leuven, Belgium.

Address reprint requests to Philippe Demaerel, MD, PhD, Department of Radiology, University Hospitals, Herestraat 49, B-3000 Leuven, Belgium.

Clinical and MR findings in four patients with cerebellar cortical dysplasia

Case	Age, y/Sex	Clinical Findings	Cerebellar Abnormalities	Supratentorial Abnormalities
1	8/M	Right hemiparesis, poor language development, normal intelligence	Bilateral hemispheric cortical dysplasia	Polymicrogyria in left frontal, parietal, and temporal lobes
2	15 /F	Psychomotor retardation, epilepsy	Bilateral hemispheric cortical dysplasia with cystlike inclusions	Polymicrogyria in left and right frontal, parietal, and occipital lobes, dilated ventricular system
3	21 /F	Psychomotor retardation, epilepsy	Bilateral hemispheric cortical dysplasia with subtle cystlike inclusions	Polymicrogyria in left and right frontal and parietal lobes, dilated ventricular system, bilateral subcortical white matter hyperintensities
4	5/M	Psychomotor retardation, poor language development	Bilateral hemispheric cortical dysplasia	None

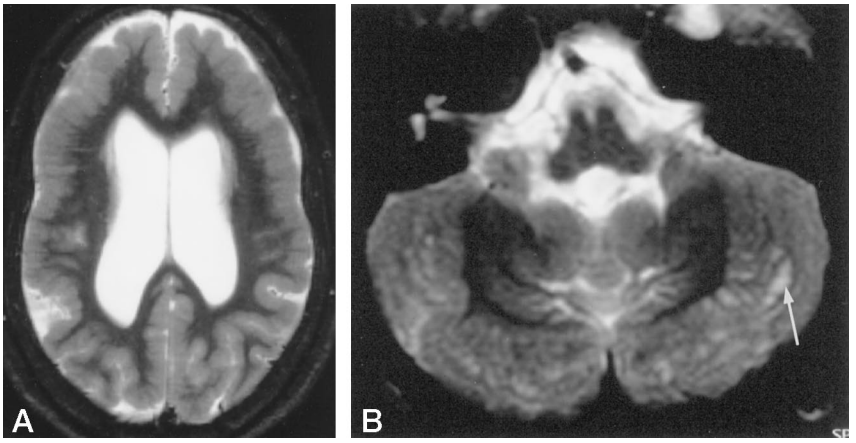

FIG 1. Axial T2-weighted MR images in a 15-year-old girl. The frontal and parietal lobes show thickening of the cortex with shallow sulci (*A*). At the level of the middle portion of the cerebellum, the folia appear diffusely disorganized with a bumpy gray-white matter interface (*B*). Note the presence of intracortical areas of high signal (*arrow, B*).

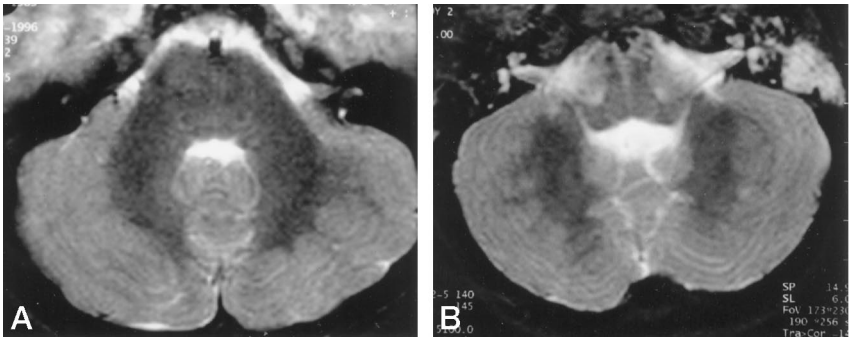

FIG 2. Axial T2-weighted MR images of the middle portion of the cerebellum in an 8-year-old boy (*A*) and in a 5-year-old boy (*B*) show the disorganized folia and the bumpy gray-white matter interface.

cortex achieves its adult histologic composition only by the end of the first postnatal year.

Generally, two different types of cerebellar maldevelopment have been distinguished pathologically: heterotopias, resulting from a disturbance in the migration of neuroblasts from the rhombic lips, and cortical dysplasias, caused by disturbances of cortical layering (11). Histologically, the features of cerebellar cortical dysplasia have been found to be consistent with the fusion of molecular layers, a granular layer deficit, and Purkinje cells scattered at the boundaries of these layers (11, 12). The nodulus, flocculus, and tonsils are common sites of cortical dysplasia, but any portion of the cortex may be affected. The presence of intracortical cystlike abnormalities may represent a more severe injury or an injury occurring at a different time during development. We prefer "cystlike abnormalities," because the lesions are different from the well-defined intraparenchymal cysts described in FCMD (4, 5). It cannot be excluded that these longitudinal cystlike abnormalities represent remnants or sequelae of the migration of neurons that form the external granular layer of the cerebellar cortex at 11 to 13 weeks of gestation. Remnants of daughter cells, which migrate inward to form part of the outer molecular layer and inner granular layer at 16 weeks, may also be responsible for these abnormalities.

In FCMD and related syndromes, such as Walker-Warburg syndrome, the cerebellar abnormalities are always associated with supratentorial type II lissencephaly or cobblestone lissencephaly (4, 5, 13). Histologically, lissencephaly type II combines appearances of agyria and polymicrogyria, and usually this term is used in the above-mentioned syndromes only.

We considered the supratentorial abnormalities in our patients to be more consistent with polymicrogyria or cortical dysplasia. Our patients did not meet the criteria for a cobblestone lissencephaly syndrome, because two necessary diagnostic criteria, brain stem and cerebellar hypoplasia, were absent (14). None of our three patients with supratentorial polymicrogyria had FCMD. In the fourth patient, the only MR finding was cerebellar cortical dysplasia, which, to our knowledge, has not been described before. Pathologically, cerebellar cortical dysplasia can be found as an incidental lesion in an otherwise normal cerebellum. The most extensive dysplasias are seen in FCMD and related syndromes. These observations together with our findings suggest that the cerebellar abnormalities may represent a spectrum of morphologic changes ranging from minimal to extensive. From the clinical point of view, two observations are of note: first, none of our patients had cerebellar signs, and it is reasonable to believe that the symptoms caused by the supratentorial findings overshadow the cerebellar signs; second, poor language development was seen in two of the patients, which may be related to the cerebellar cortical dysplasia, since it is accepted that development of language depends on a normal cerebellar function and morphology. The pathogenesis remains incompletely understood, and it is likely that, similarly to cerebral polymicrogyria, cerebellar cortical dysplasia does not represent a single entity but may instead result from different causes, both genetic and acquired (15, 16).

Conclusion

Cerebellar cortical dysplasia is an uncommon disorder that, to our knowledge, has not been described as a solitary finding and that until now has only been associated with FCMD and related syndromes. If the MR images are not reviewed with care, the abnormalities can be overlooked, particularly if there are no other anomalies.

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