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Recurrent Lhermitte-Duclos Disease: Report of Two Cases and Association with Cowden's Disease

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Summary: Two cases of Lhermitte-Duclos disease (LDD), one associated with Cowden's disease, are reported. Both demonstrated recurrence long after initial surgical resection. There is a propensity for occurrence in the left cerebellar hemisphere. On MR there is hypointensity on T1-weighted images and moderately high signal on T2-weighted images, with parallel linear striations on the surface of the lesion felt to represent dysplastic cerebellar folia. LDD is felt to be a low-grade neoplasm and potential component of Cowden's phakomatosis.

Index terms: Cerebellum, neoplasms; Cerebellum, abnormalities and anomalies; Hamartoma; Phakomatosis

Lhermitte-Duclos disease (LDD) is a rare disorder of the cerebellum, characterized by the presence of hypertrophic ganglion cells in the granular and molecular layers with excessive axonal myelination (1). Fewer than 65 cases have appeared in the medical literature since this entity was first described in 1920 (1–20). Considerable controversy remains concerning whether this lesion represents a hamartomatous malformation, a developmental anomaly, a manifestation of a phakomatosis, or a low-grade neoplasm.

Case Reports

Case 1

The first patient, a 55-year-old woman, was referred to us for cranial magnetic resonance (MR) imaging because of headaches, dizziness, and ringing in her ears. Her past medical history was significant in that 12 years previously, she had presented to our institution with hearing loss and cerebellar signs. A posterior fossa mass was discovered by computed tomography (CT) and verified with pneumoen-

cephalography. Subtotal surgical resection was carried out, and histologic examination of the specimen revealed classic changes of LDD. The pathologic and CT findings in this case were subsequently reported in the neurosurgery literature in 1980 (6).

Since that time, the patient did well neurologically, but developed a multitude of other clinical problems. Two years following her neurosurgical procedure she was noted to have multiple cutaneous lesions, including fibroepithelial polyps, capillary hemangiomas, and papular trichilemmomas of her face, neck, and axillae. A clinical diagnosis of Cowden's disease was then made, supported by the fact that an aunt had also been previously diagnosed with this disorder. Subsequently, our patient developed multiple intraabdominal tumors, including three hepatic cavernous hemangiomas and a benign serous cystadenoma of the ovary, all of which were removed surgically.

A cranial MR scan was obtained in 1989, prompted by a gradual onset of new neurologic complaints, including headaches, vertigo, and tinnitus. The study was performed at 1.5 T, both with and without paramagnetic contrast material. A large recurrent left cerebellar mass was present that was of moderately high signal on T2-weighted images and slightly hypointense to brain on T1-weighted images (Fig. 1). A cluster of enlarged draining veins was noted in association with this lesion. Elsewhere in the brain, three additional venous angiomas were observed. Because of her rather mild neurologic symptoms, this patient has not yet undergone a repeat craniotomy for the above abnormalities.

Case 2

This 37-year-old white man came to medical attention in 1984 because of severe headaches while bending over. Clinical examination revealed papilledema, but no evidence of visual or cerebellar disturbance. CT demonstrated ob-

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Fig. 1. Case 1; a 55-year-old woman with recurrent LDD and Cowden's disease.

A, Axial T2-weighted (2300/60/1) image through cerebellum shows high-signal mass in left cerebellar hemisphere with associated vessels (*arrows*). High signal paralleling vascular flow voids (*arrowheads*) is misregistration artifact and signifies centrifugal flow.

B, Parasagittal SE 600/20/2 (T1-weighted) image through left cerebellar hemisphere. Note hypointense mass with a few linear striations (*arrows*).

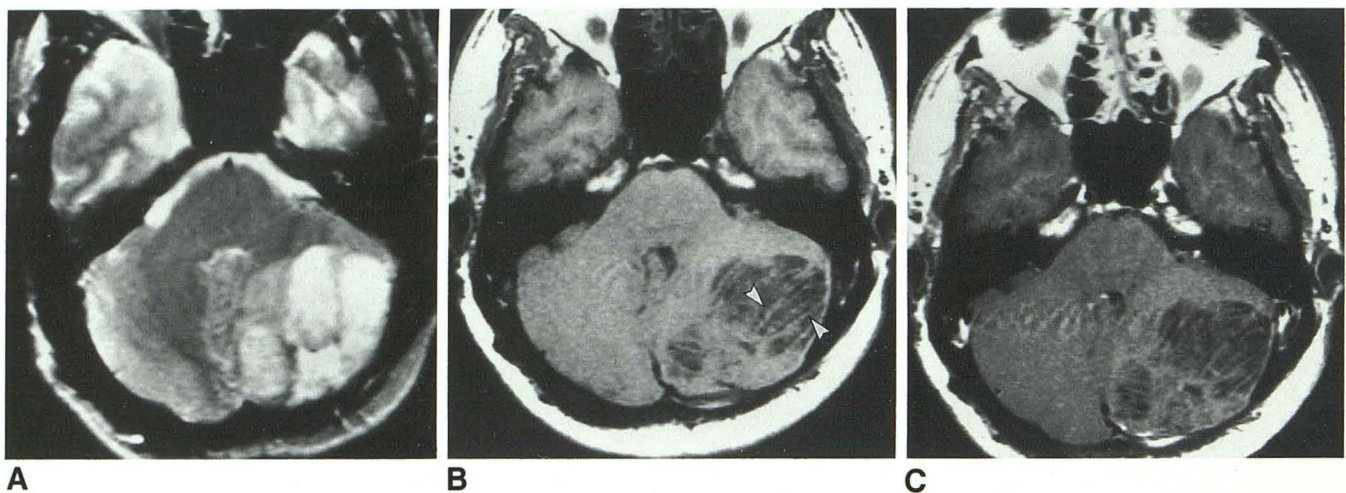
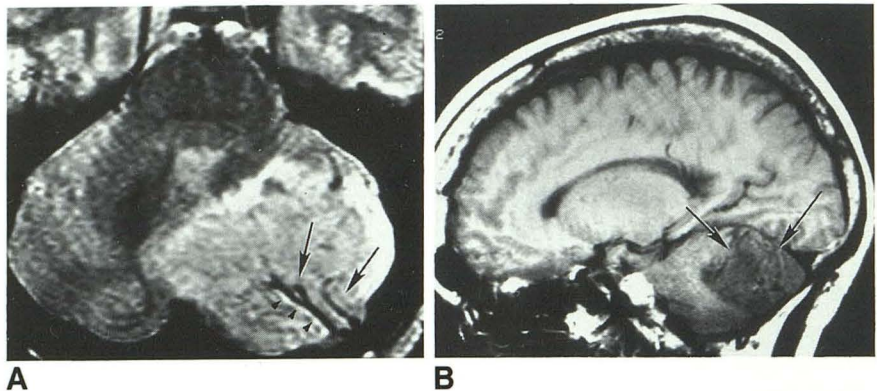


Fig. 2. Case 2; a 37-year-old man with recurrent LDD.

A, Axial T2-weighted (2500/80/1) image shows a high-signal complex lesion with relatively little mass effect considering its size.

B, Precontrast T1-weighted (600/20/2) image shows multiple linear striations consistent with thickened cerebellar folia (*arrowheads*).

C, Postcontrast T1-weighted image shows no enhancement of the lesion.

structive hydrocephalus caused by a nonenhancing, low-attenuation mass occupying approximately 75% of the left cerebellar hemisphere. A subtotal resection was performed with removal of most of the visible tumor, and histologic findings were pathognomic of LDD.

Postoperatively, the patient did very well for several years. Interim CT studies revealed a persistent region of low density in the posterior fossa thought to represent postoperative changes. By early 1990, the patient began to experience visual disturbances and ataxia and was referred for MR imaging. A large recurrent mass in the left cerebellar hemisphere was noted with effacement of part of the quadrigeminal plate cistern and upper fourth ventricle. T2-weighted images revealed a high-signal mass with multiple components (Fig. 2A). T1-weighted images demonstrated a low-signal mass with characteristic linear striations resembling thickened cerebellar folia (Fig. 2B). No enhancement of the mass was seen postcontrast. This patient, likewise, has not yet undergone a second surgery for this presumed recurrence of LDD.

Discussion

LDD is a rare disorder of the cerebellum known by a variety of names: dysplastic gangliocytoma of the cerebellum, Purkinjeoma, ganglioneuroma, granular or granulomolecular hypertrophy of the cerebellum, diffuse hypertrophy of the cerebellar cortex, gangliomatosis of the cerebellum, hamartoma of the cerebellum, myelinated neurocytoma, and gangliocytoma myelinicum diffusum (1–20). This multitude of names reflects the historic difficulty in its pathogenetic classification. Should LDD be considered to be a dysplasia, hamartoma, or neoplasm?

The cerebellar lesion of LDD has a characteristic histologic appearance consisting of thickened cerebellar folia, markedly decreased central white matter, and loss of the normal cortical architecture (3–12). This latter finding is manifested by abnormal ganglion cells in the granular

layer, loss of Purkinje cells, and thickening and hypermyelination of the molecular layer. Patients usually present clinically in the 3rd or 4th decade of life with signs and symptoms of cerebellar dysfunction and/or hydrocephalus (headaches, ataxia, visual disturbance, nausea/vomiting). Numerous associated abnormalities have been described in patients with LDD (2, 3, 14). These include: megalencephaly (in approximately 50% of reported cases), heterotopias, microgyria, hydromyelia, polydactyly, peritheliomas, spongioblastomas, hemangiomas, partial gigantism, macroglossia, and leontiasis ossea. At least one patient has had clinical neurofibromatosis (3), familial cases have been documented (3), and a case has been described in a newborn (12).

The association of LDD and Cowden's disease has only recently been appreciated (20, 21). Cowden's disease, also called multiple hamartoma syndrome, is a rare hereditary condition characterized by multiple hamartomas, and neoplasms of ectodermal, mesodermal, and endodermal origin (22-24). Mucocutaneous abnormalities, as demonstrated in our first patient, are felt to be the most characteristic finding in this condition. Other reported manifestations and associations include macrocrania, gastrointestinal polyps, breast disease (carcinoma, fibrocystic disease), thyroid abnormalities (goiter, adenoma, carcinoma), supernumerary digits, ovarian cysts, hepatic lesions (hamartomas, hemangiomas), neuromas, neurofibromas, and meningiomas. The simultaneous occurrence of LDD and Cowden's disease has been reported previously in three patients (20, 21). In two of these three cases, as well as in one of our own, the diagnosis of Cowden's disease was not made until some time after biopsy of the cerebellar lesion. Because the manifestations of Cowden's disease are variable and few physicians are even aware of this entity, it remains unknown whether other reported patients with LDD may have also had undiagnosed Cowden's disease. Likewise, it is unknown how many patients initially diagnosed with LDD may have subsequently developed Cowden's disease.

Radiologic findings in LDD have been previously described both on CT and MR (6-20). The lesion has a marked propensity to involve the left cerebellar hemisphere compared to the right (10), a finding also borne out in our two cases. The usual CT appearance is a nonenhancing hypodense mass with occasional focal calcifications. The degree of mass effect is usually relatively slight, considering the size of the lesion; this

suggests a benign or slowly growing process. On MR imaging, the lesion is typically of moderately high signal on T2-weighted images and slightly hypointense to brain on T1-weighted images. The presence of parallel linear striations on the surface of the lesion, presumably representing thickened, dysplastic cerebellar folia, are nearly pathognomonic (19). This finding was observed in both our cases, although it is much better appreciated in case 2.

A new MR finding, which we believe may be supportive of the diagnosis of LDD, is the presence of prominent draining veins within or adjacent to the cerebellar mass. These were seen to good advantage in case 1, and similar vascular flow voids were also present (but not commented upon) in the recent case reported by Smith et al (19, fig 2D) (21). Oppenheimer, in one of his pathologic specimens of LDD, noted a "generalized proliferation of small blood vessels immediately below the pia but also in the underlying tissue in two places [amounting] to angiomatous nodules visible to the naked eye" (2). We thus feel that prominent draining veins may be an ancillary radiologic sign that supports the diagnosis of LDD.

A final feature of LDD illustrated by our two cases is its propensity for recurrence. Di Lorenzo et al (11) have drawn attention to the fact that the postsurgical follow-up periods for most patients with LDD reported in the literature have been extremely short, and we do not know the natural history of this disorder. Marano et al (14) reported the first documented recurrence of LDD in a child after surgical resection. Reeder et al (16) reported recurrence after gross removal in two other patients postoperatively at 4 and 10 years, respectively. We add two additional cases of recurrent LDD in this report. It should be noted that one of our cases was previously reported as a surgical success in 1980 (6). Based upon this experience and critical review of the existing literature, we feel LDD should be considered a low-grade neoplasm that may recur after subtotal resection. Furthermore, LDD should also be recognized as a central nervous system manifestation of Cowden's phakomatosis. Consideration of the characteristic radiologic features we have described, together with knowledge of its biological behavior and clinical associations, should allow a more confident diagnosis of LDD to be made preoperatively.

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