

Magnetic Resonance Imaging of Lhermitte-Duclos Disease a Case Report

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Abstract: *The Lhermitte-Duclos disease, or dysplastic gangliocytoma, is a benign hamartomatous condition involving the cerebellum of unknown etiology^[1,2]. We report a 30-year-old female patient who presented to us with history of headache and gait ataxia and was observed to have Lhermitte-Duclos disease and a brief review of the condition with image description.*

1. Introduction

Lhermitte-Duclos disease is also called dysplastic cerebellar gangliocytoma, granular cell hypertrophy, granulomolecular hypertrophy of the cerebellum, diffuse hypertrophy of the cerebellar cortex, Purkinjeoma, cerebellar hamartoma, diffuse ganglioneuroma^[3], gangliomatosis of the cerebellum, neurocystic blastoma, and hamartoblastoma, is a rare disease of unknown etiology^[4], when associated with PTEN mutations as in Cowden's syndrome its origin is strongly suggestive of a hamartomatous disorder^[5,6].

Lhermitte-Duclos disease is always infratentorial, usually involving the cerebellar hemisphere or the vermis. Large lesions involve both. The brainstem is a rare site. Dysplastic cerebellar gangliocytomas often become very large, displacing the fourth ventricle and causing obstructive hydrocephalus. The vast majority are unilateral, although a few cases of LDD with bilateral lesions of the cerebellar hemispheres have been reported where mostly they have crossed over to the opposite side.

The gross appearance is a tumor-like mass that expands and replaces the normal cerebellar architecture. On cut section, the cerebellar folia are markedly widened and have a grossly gyriform appearance. Although it is probably a hamartoma and not a true neoplasm, it is designated as WHO grade I. The prevalence of LDD is unknown. The incidence of Cowden syndrome with PTEN mutation is estimated at 1 in 250,000. The average age at diagnosis is 34 years. There is no gender predilection. Patients may be asymptomatic or present with symptoms of increased intracranial pressure such as headache, nausea, and vomiting.

Cranial nerve palsies, gait disturbance, and visual abnormalities are also common. It enlarges very slowly over many years. No cases of metastatic spread or CSF dissemination have been reported. Treatments options include shunting or surgical debulking for symptomatic patients with hydrocephalus. Because dysplastic cerebellar gangliocytoma is not encapsulated and blends gradually into normal cerebellar tissues, complete resection is difficult and the complication rate is high, but recurrence is very rare.

2. Case Report

A 30-year-old woman presented with chronic headache on and off, localized to the occipital region, she also had blurring of vision and gait disturbances. She had 4 to 5 episodes of vomiting for the last 6 days. Her vitals were normal and CNS examination revealed cerebellar ataxia with no other focal neurological deficit. Magnetic resonance (MR) imaging of the brain was performed with the following findings as discussed below.

3. Discussion

This patient's history of headache, blurred vision, and vomiting was suggestive of raised intracranial pressure. Raised intracranial pressure and the long-standing progressive nature of this patient's symptoms argued against acute infarction or cerebellar encephalitis. It should be noted that the lesion was not confined to any specific vascular territory; thus, an infarct was ruled out. Lack of enhancement on the contrast-enhanced images helped us rule out leptomeningeal metastases and inflammatory diseases, such as cerebellar encephalitis.

There is considerable controversy over the etiology of this disease: It may have a hamartomatous, neoplastic, or congenital malformative origin. Eighty percent of patients with Cowden syndrome have germ line mutations in the *PTEN* gene at locus *10q23.2*, which has been identified as the major susceptibility gene for Cowden syndrome^[10,11]. Most patients with

Lhermitte-Duclos disease appear to have a germ line loss of one *PTEN* allele and go on to lose the remaining *PTEN* allele at some point, thereby allowing abnormal growth of the granule cells^[11]. However, recurrence though very rare is known to occur after resection^[7]. This finding may be indicative of a neoplastic origin.

The incidence of dysplastic cerebellar gangliocytoma is equal in males and females and is seen most frequently in young adults around 30 to 40 years of age. Clinically, patients may be asymptomatic, or they may present with symptoms and signs of increased intracranial pressure. Obstructive hydrocephalus causes cranial nerve palsies, cerebellar symptoms, and sudden neurologic deterioration; these are the usual presenting symptoms^[13]. This disease is commonly associated with other congenital malformations, such as megalencephaly, polydactyly, multiple hemangiomas, and skull abnormalities^[6]. Usually, patients have long-standing symptoms that have been present for years, indicating the slowly progressive nature of this disease. Histopathology of such lesions show disruption of the normal cell layers with dysplastic hypertrophied ganglion cells and expansion of the granule layer and commonly with increased myelination in the molecular layer causing it to widen. There is loss of Purkinje cells and white matter but maintains its cortical foliar architecture..

4. Imaging

Skull x ray may show thinning of the skull in the occipital region due to a space-occupying lesion in the posterior fossa manifesting.

CT shows a space occupying lesion which is hypo attenuating on non enhanced computed tomographic images; however, it may be isoattenuated^[13], merging imperceptibly with the adjacent normal cerebellum^[14]. In such cases, the only diagnostic clue may be the mass effect, which manifests as compression of the fourth ventricle, effacement of the cerebellopontine angle cistern, and hydrocephalus. Necrosis and hemorrhage are absent. Calcifications within the lesion may be seen. There are no large feeder vessels or neovascular collaterals so no appreciable enhancement is seen on contrast-enhanced CT images. However, CT remains of limited value because of the beam-hardening artifacts that are caused by the petrous temporal bone and obscure the details.

MR imaging is the modality of choice for all posterior fossa abnormalities. The image findings are typical on MR showing a cerebellar mass with a trigroid- striated, corduroy, or tiger-stripe design folial pattern that consists of alternating bands on both T1- and T2-weighted images. The bands are hyper- and isointense relative to gray matter on T2-weighted images iso and hypo intense on T1-

weighted images^[15]. Most dysplastic gangliocytomas do not enhance; however, enhancement has been reported^[16] and is probably due to the presence of anomalous veins, showing linear enhancement of veins around thickened folia^[17]. Mass effect is common and causes compression of the fourth ventricle and occlusive hydrocephalus^[13].

In our case almost all the characteristic findings of Lhermitte duclos was seen. There was a space occupying which turned out to be an expansile mass in the right cerebellar hemisphere causing mild midline shift. The lesion was causing obstructive hydrocephalus as expected as it was seated near the fourth ventricle. It had a text book trigroid pattern, causing disarrangement of folial pattern. Both T1- and T2-weighted images showed alternate bands of iso and hyper intensity. On T1-weighted images the lesion shows predominantly iso and few hypo intense bands in the right cerebellar hemisphere. The bands are isointense relative to gray matter on T2-weighted image with intervening hyperintensities. The lesion did not show any significant enhancement but the presence of few veins resulted in linear enhancements.

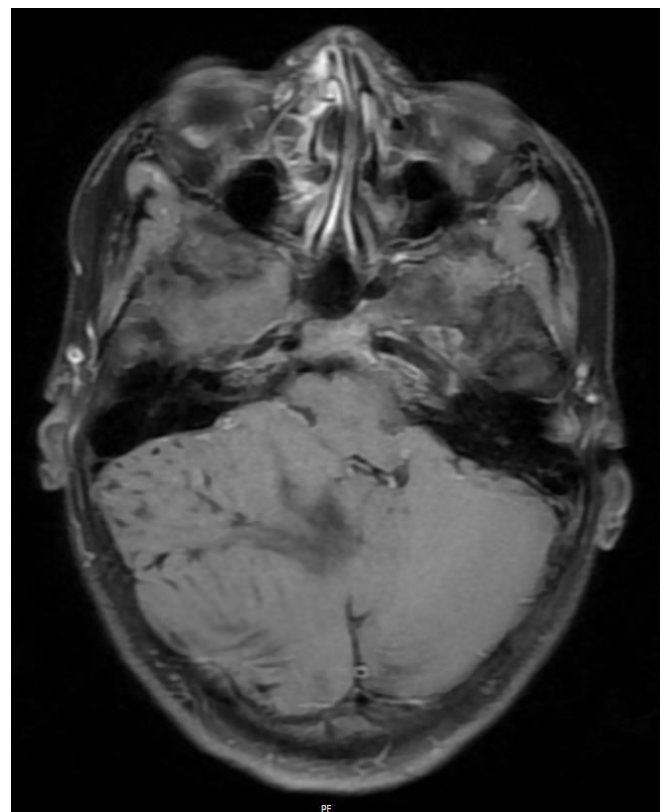


Figure 1. An expansile cerebellar mass with linear hypo to isointense bands on T1WI

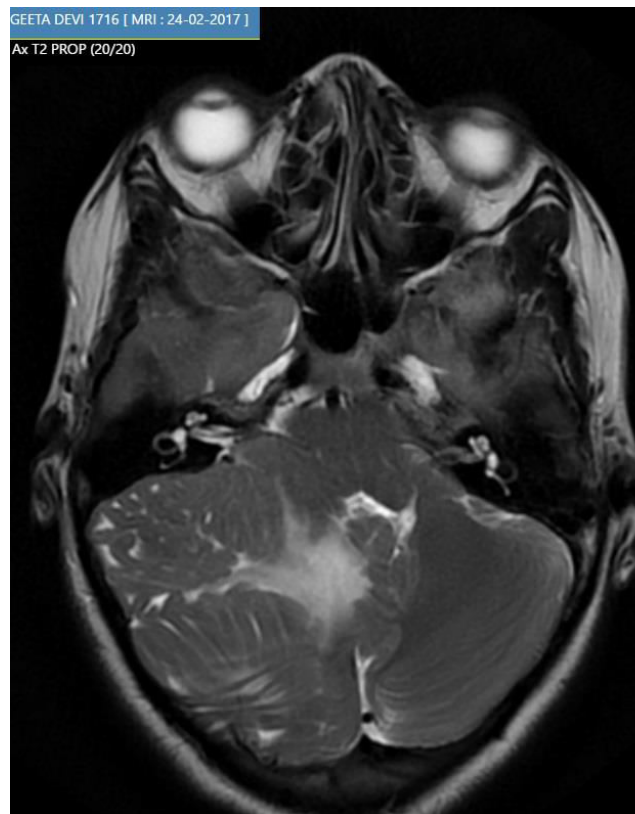


Figure 2. Linear hyper to isointense bands on T2WI axial sections

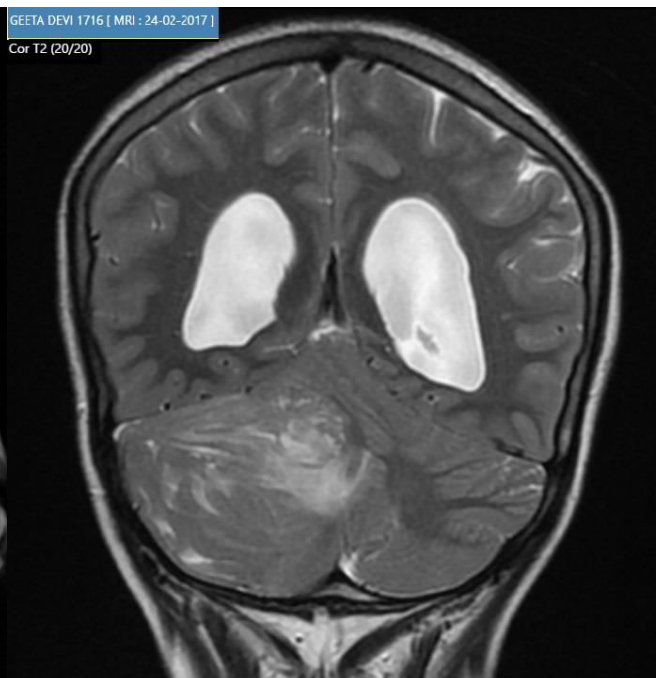


Figure 4. Similar linear hyper to isointense bands on T2WI coronal sections showing the midline shift

MR spectroscopy usually shows normal or reduced *N*-acetylaspartate–choline and *N*-acetylaspartate–creatine and normal Choline–creatine ratios compared with those of normal cerebellar tissue. Lactate doublet peak may also be present.^[18]

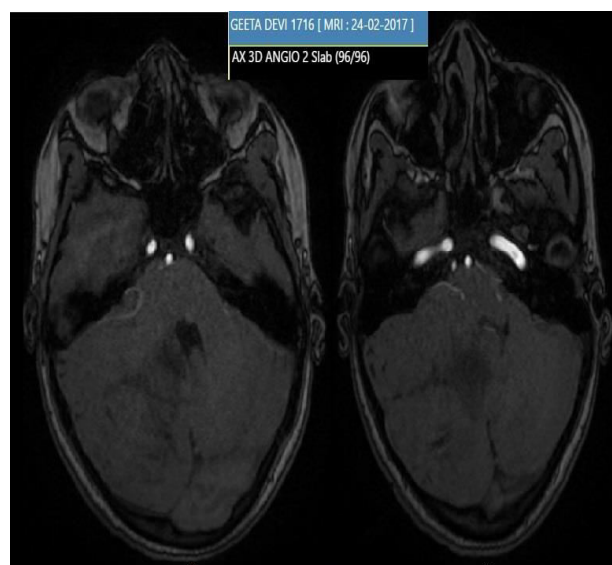


Figure 3. Post contrast images showing no enhancement but few enhancing veins can be seen

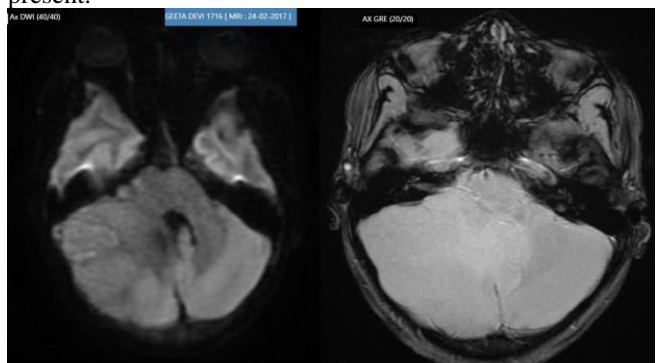


Figure 5. Diffusion imaging shows no restriction

Imaging findings are so characteristic that the diagnosis can usually be established without biopsy confirmation. Although a case of medulloblastoma mimicking dysplastic gangliocytoma at neurologic imaging has been reported in a pediatric patient, diagnostic confusion with medulloblastoma is unlikely in most patients because of differences in age group, medical history, and usual imaging features^[19]. MR is useful for preoperative planning, assessment of the lesion extent, mass effect, presence of hydrocephalus, and need to perform surgery.

Alternate diagnoses such as cerebellar astrocytoma, medulloblastoma, cerebellitis and tuberous sclerosis were ruled out and all of the above

could be considered as a differential to dysplastic cerebellar gangliocytoma.

5. Conclusion

Lhermitte-Duclos disease is characterized by dysplasia of the cerebellum which results in a tumor like mass replacing the normal architecture. Usual presentation is raised intracranial pressure. MRI imaging is characteristic and usually diagnostic. It may also reveal tonsillar herniation and syringomyelia. Surgical decompression is the treatment of choice and patient need to be kept on follow up as long term recurrence can occur. Immediate treatment is to relieve intracranial pressure by way of shunts; there is excellent prognosis due to minimal chances of recurrence after surgery^[20].

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7. References

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