

## Lhermitte-duclos Disease: A Case Report.

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### Abstract:

Lhermitte-Duclos disease (LDD) is a benign neoplasm of posterior fossa, involving cerebellum. It is also known as dysplastic cerebellar gangliocytoma. It is not a true neoplasm but a hamartoma. It can be either isolated finding or associated with Cowden disease (multiple hamartoma syndrome). Lhermitte-Duclos disease typically presents in young adults.

**Key Words:** Lhermitte-duclos disease, Hamartoma, Dysplastic Cerebellar Gangliocytoma

### Introduction:

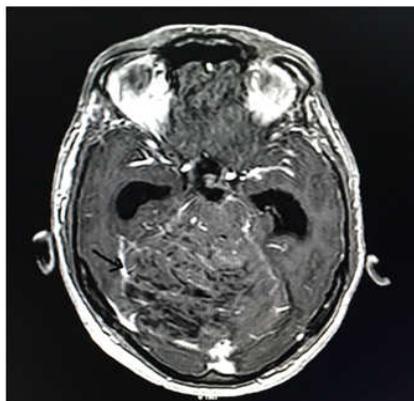
Lhermitte-Duclos disease (LDD) is a benign neoplasm of cerebellum. Lhermitte-Duclos disease typically presents in young adults (mean age of presentation is 20-40 years with peak in 35 years). It is a rare tumour which occurs in association with Cowden's disease. It can also occur sporadically.

### Case History:

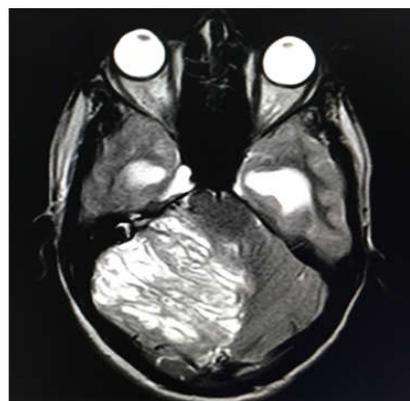
A twenty year old male patient was presented with complains of continuous headache since 4 months, diplopia since 2 months, inability to maintain balance and vomiting after meals. On examination, there was medial deviation of right eye. Patient was advised MRI brain with contrast.



**Image 1: T1 Post contrast showing Prominent venous channels**



**Image 2: T2 Weighted images showing Tiger stripe appearance**



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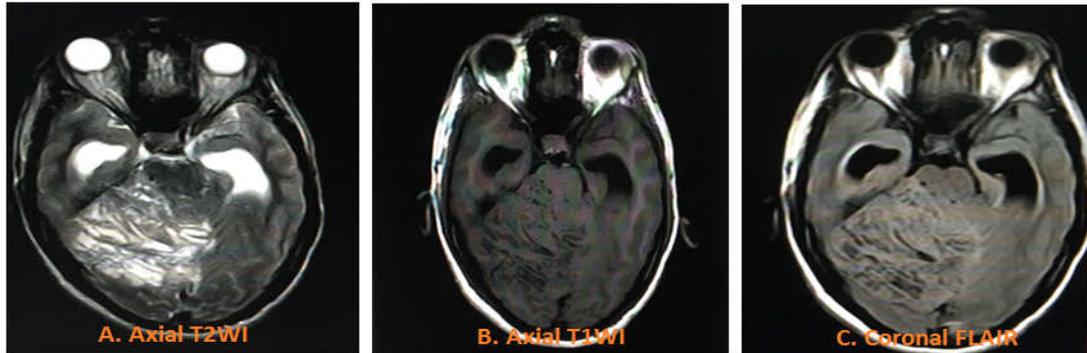
MRI findings were suggestive of altered signal intensity lesion involving right cerebellar hemisphere,

right cerebellar peduncles and vermis. On T1WI, the lesion was iso-hypointense with linear hypo-intense bands. On T2WI, the lesion shows alternating hyper and hypo-intense bands in the enlarged cerebellar folia. The lesion shows diffusion restriction on DWI (diffusion weighted images). On post contrast study, there is prominence of adjacent venous channels.

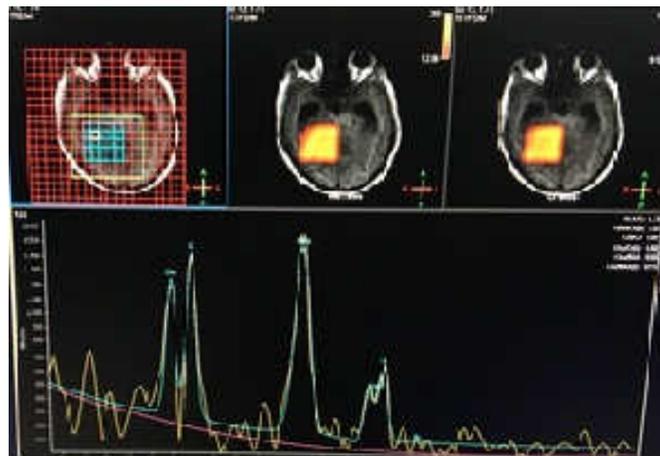
There was mass effect in the form of obliteration of basal cisterns and hydrocephalus with herniation of cerebellar tonsils.

MR Spectroscopy reveals lactate doublet peak.

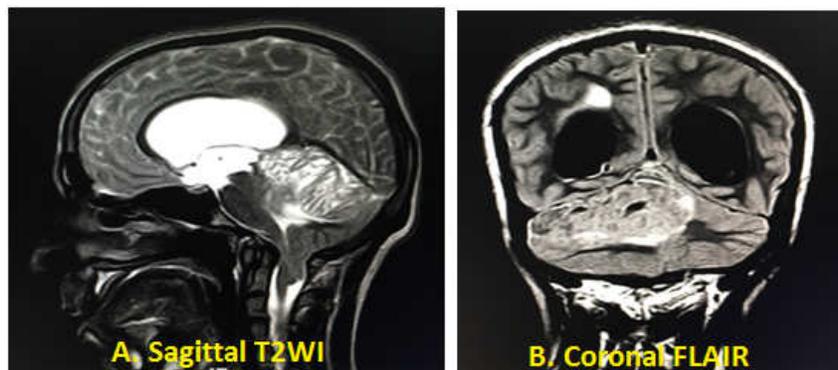
**Image 3: Altered signal intensity involving Right cerebellar hemisphere & Vermis.**



**Image 4: MR Spectroscopy shows Lactate doublet peak**



**Image 5 Hydrocephalus and Herniation of cerebellar tonsils (5.A)**



**Discussion:**

Lhermitte-Duclos disease (LDD) is a rare benign tumour of cerebellum. It is also known as dysplastic cerebellar gangliocytoma. It has probably hamartomatous origin, although the exact pathogenesis remains unknown. It is considered a WHO grade I tumour<sup>1</sup>

**Epidemiology:** Lhermitte-Duclos disease typically presents in young adults (mean age of presentation is 20-40 years with peak in 35 years). LDD is more often seen as sporadic case. However association with Cowden's disease is seen in 40 percent of cases<sup>1</sup>. Association with Cowden supports hamartomatous origin. When it is associated with Cowden disease then termed Lhermitte-Duclos-Cowden syndrome or COLDS syndrome<sup>1</sup>. Cowden syndrome has multiple hamartomas of skin and gastro intestinal tract, disorders of cortical formation. Breast, thyroid, endometrium and gastrointestinal cancers are the most prevalent other neoplasms<sup>2</sup>. When it occurs sporadically, it is known as Lhermitte-Duclos disease.

**Clinical presentation:** Presenting symptoms are not related to cerebellar dysfunction but are related to raised intracranial pressure, obstructive hydrocephalus, cranial nerve palsies.

**Pathology:** On Gross specimen, there are widened cerebellar folia. Microscopy reveals progressive hypertrophy of granular cell neurons with absence of Purkinje cells is characteristic.

**Markers:** Immunohistopathology shows positive staining for synaptophysin.

**Radiographic features:** The altered signal intensity is confined to one cerebellar hemisphere. The lesion may extend in to the vermis but only rarely involves the contralateral hemisphere<sup>3</sup>. The lesion is hypointense on T1WI and hyper intense on T2Weighted images with alternate stripes of inner hyperintense and outer hypointense layers giving a tigroid appearance<sup>3</sup>. The lesion causes mass effect in the form of compression of third or fourth ventricles with resultant hydrocephalus. Contrast enhancement is not seen in LDD<sup>4</sup>. The abnormally thickened folia were characterized by strong signal intensity on diffusion-weighted imaging with a high b factor, whereas ADC mapping showed no pathologic signal disturbances<sup>5</sup>.

**MR Spectroscopy:** The main role of MR spectroscopy (MRS) is to differentiate neoplastic lesions from non neoplastic etiology. MRS can also differentiate high grade from low grade glioma. Just like neoplastic lesions there are reduced NAA (N-acetyl aspartate) and increased lactate levels. Choline/Creatinine (Cho/Cr) levels were reduced in one study<sup>6</sup>. Double lactate peak is characteristically seen in LDD.<sup>1,6</sup> In our case there are both reduced NAA levels and reduced Cho/Cr ratio along with classical lactate doublet peak.

**Differentials:** Striated pattern on T2WI tigroid appearance is Aunt Minnie for the diagnosis and does not even need histopathological confirmation<sup>5</sup>.

However differential diagnosis would include posterior fossa masses-

- Medulloblastoma-desmoplastic variant: Age of presentation is in younger patients.
- Gangliocytoma- post contrast enhancement may show bizarre tigroid like appearance, but the Tigroid appearance of LDD is seen on T2WI.

- Cerebellar infarction- Symptoms are acute in onset, lesion limited to vascular territory
- Cerebellar cortical dysplasia- they lack progressive enlargement and mass effect.

**Conclusion:**

The above mentioned case is of a 20 year old male presenting with chronic complains. The age of the patient lies in the common age group of 20-40 years who most commonly present. The complains were long standing, ruling out any acute vascular insult. MRI study showed T2WI characteristic of the lesion. On post contrast study, there was no enhancement of the lesion; however vascular channels surrounding the lesion stood prominent. MRS showed classical lactate doublet peak which is characteristic of LDD. The patient was further screened for Cowden syndrome. Clinical examination did not reveal any cutaneous lesions. Ophthalmological examination did not reveal any hamartomatous lesions. Ultrasonography was normal. Hence association with Cowden syndrome could not be proved which is seen in 40 % cases. Therefore this was considered to be a sporadic case of Lhermitte-Duclos disease

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