American Journal of Multidisciplinary Research & Development (AJMRD)
Volume 2, Issue 7 (July- 2020), PP 19-22
ISSN: 2360-821X
www.ajmrd.com

Research Paper **Open Access**

Lhermitte Duclos disease - A case report

Ramya C[1], Renuka IV[2], Durga Prasad B[3], Pratyusha N[4]
[1,23,4]*Department of Pathology, NRI Medical college, India*
**Corresponding Author:Renuka IV*

ABSTRACT:- Lhermitte Duclos disease is a rare cerebellar disease with very few cases published in the literature. There is no gender predilection and can occur at any age group. We present a rare case of unilateral LD disease in a young female with a lesion in left cerebellum. Preoperative diagnosis was made by MRI, which is confirmed on histology. A 32yr old female presented with headache and blurring of vision since two months. MRI revealed tiger stripped appearance of a lesion in cerebellum measuring 4.5cms× 3.5cm, which on histopathology showed Lhermitte Duclos disease. Most of the cases can be diagnosed by MRI, but certain malformations and medulloblastoma can be differential diagnoses on imaging. The prognosis is good with very few cases of recurrence. Lhermitte Duclos disease is a rare cerebellar disease usually associated with Cowden syndrome. The patient was screened for other malignancies.

Keywords:- Cerebellum, Dysplastic cerebellar gangliocytoma, Lhermitte Duclos disease

I. INTRODUCTION

Lhermitte Duclos (LD) disease is a rare disease described by French physicians Lhermitte and Duclos in 1920. [1] There are various synonyms like dysplastic cerebellar gangliocytoma, cerebellar granule cell hypertrophy, diffuse hypertrophy of cerebellar cortex, gangliomatosis of the cerebellum. [2] Very few cases have been published from different countries all over the world. Most patients become symptomatic in the third to fourth decades of life, although the disease may be present in any age group. [3] They are usually confined to one hemisphere but bilateral cases have also been reported. [4] Clinically, patients may be asymptomatic, or they may present with symptoms of ataxia, headache, cranial nerve palsy, paroxysm of vertigo, psychic deterioration and, in severe cases, signs and symptoms of intracranial hypertension secondary to hydrocephalus. [5, 6]

Magnetic resonance imaging (MRI) is the appropriate technique allowing preoperative diagnosis with the characteristic striated pattern of exaggerated folia appearance on T2-weighted images.[7] MRI with the newer imaging capabilities can demonstrate the pathology and pathophysiology in Lhermitte-Duclos disease better.[7] Neurological deterioration or increase in the size of the cerebellar mass necessitates surgery and removal of the mass. PTEN mutations have been identified in adult patients with LD disease but not in children. [8]

II. CASE HISTORY:

A 32year old female presented with headache & blurring of vision since 2 months. Headache was in left temporoparietal region, more in the mornings. On neurological examination, left cerebellar sign was positive. Fundus examination revealed bilateral papilledema. Complete Blood Picture and renal profile were normal. Chest X-Ray, Electrocardiogram (ECG), Echocardiography were within normal limits. MRI revealed a mixed intensity (T1 hypo, T2 heterogenously hyperintense intra axial mass lesion in the left cerebellum and vermis measuring 4.5cmx 3.5cms in size (Fig 1a & 1b). The lesion was causing mass effect over adjacent stuctures causing supratentorial ventricular system dilatation with cerebrospinal fluid seepage.

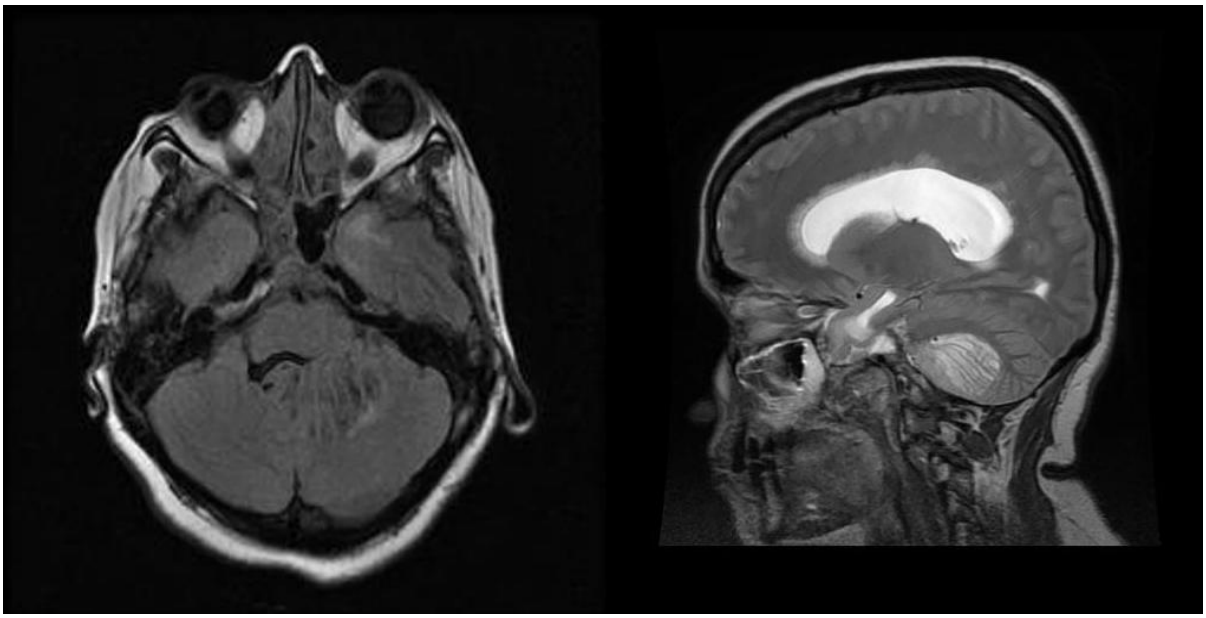

Figure 1a: MRI - Axial T2 weighted image showing lesion with striated appearance in left cerebellum
Figure 1b: MRI - Sagittal T2 image showing expansion and striated appearance of lesion in cerebellum

Left retromastoid suboccipital craniotomy with tumour excision was done and sent for histopathological study. We received multiple grey white tissue bits of total size 2 × 1.5cm. There was relative preservation of cerebellar folia on gross appearance. Entire tissue was submitted for study. On Microscopy, Normal cerebellar tissue was also included in the material along with the lesion (Fig 2a &2b). There was an abnormal increase of dysplastic ganglion cells (Fig 3a, 4a) and myelinated axons of nerve fibers (Fig 3b). There were no areas of necrosis or increased mitotic activity. Immunohistochemistry (IHC) was positive for synaptophysin in the ganglion cells (Fig 4b). Post-operative computed tomography (CT) scan showed minimal subarachnoid haemorrhage along the interhemispheric fissure with minimal hydrocephalus. There is decrease in the size of ventricles compared to preoperative scan. The patient was screened for malignancies and had no manifestations of Cowden syndrome.

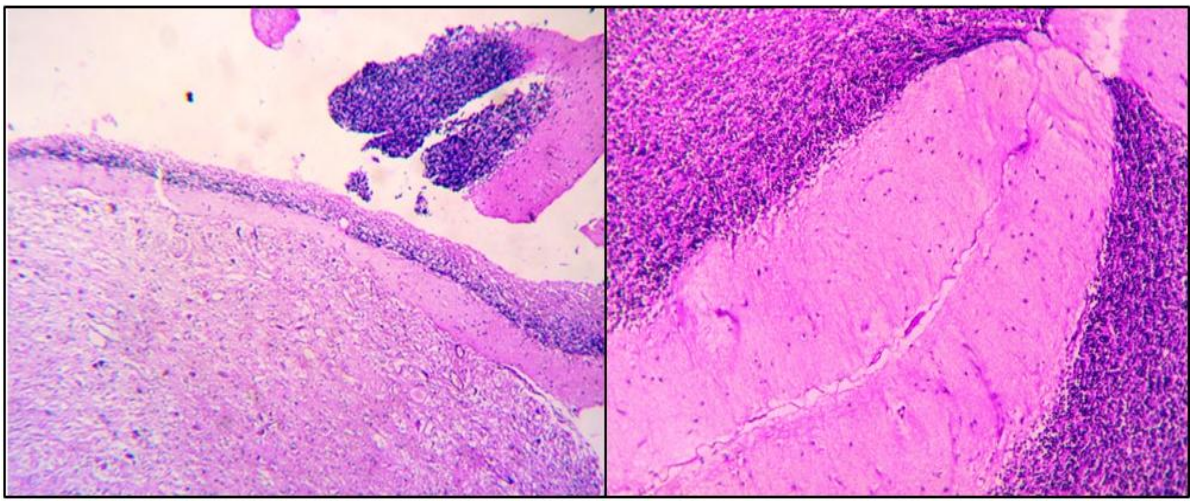

Figure 2a: Lesion with adjacent normal cerebellar tissue 40X, H&E
Figure 2b: Normal cerebellar tissue 100x, H & E

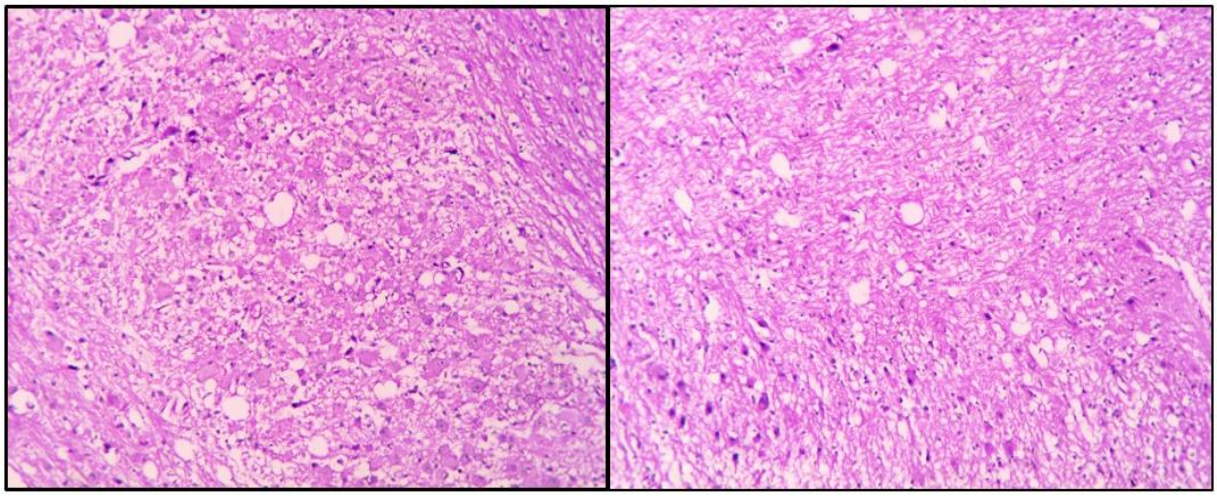

Figure 3a: Dysplastic ganglion cells 100X, H&E
Figure 3b: Myelinated axons 100X, H&E

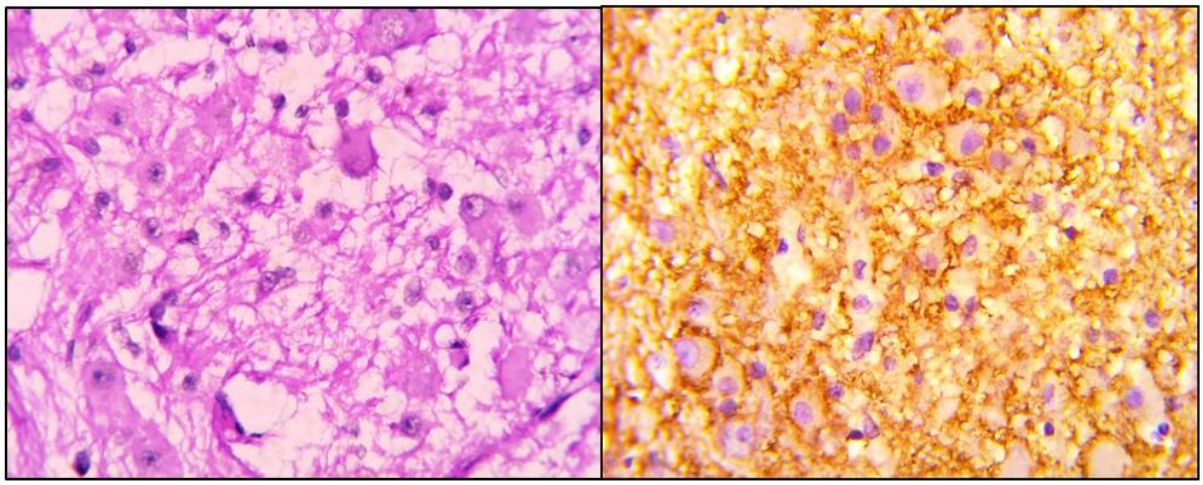

Figure 4a: Dysplastic ganglion cells 400X, H&E
Figure 4b: IHC -Synaptophysin 400X

III. DISCUSSION

Preoperative diagnosis was made by MRI in our case, but few other lesions can mimic LD disease on radiology. Hyuang et al. revealed characteristic tiger stripped appearance on MRI and histopathological examination revealed normal cerebellum with arachnoid vascular malformation and partial loss of Purkinje cells. [9] Nair et al. reported co existing LD disease with Dysembryoplastic Neuroepithelial tumour on histopathology with characteristic tiger stripping on MRI images.[10] Infiltrating medulloblastomas have also been reported to mimic LD on imaging.[11]

Sheets of dysplastic ganglionic cells were seen in granular layer in our case. LD disease causes diffuse enlargement of molecular and internal granular layers of the cerebellum, which are filled by ganglionic cells of various sizes. [12] Markers of the neoplastic process, such as mitotic activity, necrosis and endothelial proliferation are characteristically absent as seen in our case. No case of malignant transformation has been reported. [13] It can recur locally but do not spread to other structures in the brain or outside central nervous system. [2]

There is no association with Cowden syndrome in our case. In a study of 211 patients with Cowden syndrome, 32% developed LD disease. [14] Because cerebellar lesions may develop before the appearance of other features of Cowden syndrome, patients with Lhermitte Duclos disease should be monitored for the development of additional tumours, including breast cancer in females. [2]

IV. CONCLUSION:

We are presenting a rare case of unilateral LD disease in young female with a lesion in left cerebellum. Preoperative diagnosis was made by MRI, which is confirmed on histology. The patient was screened for other malignancies even though there was no association with Cowden syndrome in our case.

Conflicts of interest : NIL
Acknowledgement : NIL
Sources of support : NIL

REFERENCES:

[1]. Lhermitte J, Duclos P. Sur un ganglioneurome diffus du cortex du cervelet. Bull Assoc Fr Etude Cancer 1920; 9:99 –107.
[2]. David NL, Hiroko O, Otmar D W, Webster K C. WHO classification of tumours of the central nervous system. Revised 4th edition IARC press Lyon 2016; 142-143.
[3]. Kumar R, Vaid VK, Kalra SK. Lhermitte-Duclos disease. Childs Nerv Syst. 2007 Jul; 23(7):729-32.
[4]. Bozbuga M, Gulec I, Suslu HT, Bayindir C. Bilateral Lhermitte-Duclos disease. Neurol India. 2010; 58(2):309-11.
[5]. Somagawa C, Ono T, Honda R, Baba H, Hiu T, Ushijima R et al. Frequent vomiting attacks in a patient with Lhermitte-Duclos disease: a rare pathophysiology of cerebellar lesions?. J Neurosurg Pediatr. 2017; 20(3):298-301
[6]. Otheman Y, Aalouane R, Aarab C, Rammouz I. A case report of Lhermitte-Duclos disease revealed by psychiatric disturbances. Ann Gen Psychiatry. 2017; 16:24.
[7]. Thomas B, Krishnamoorthy T, Radhakrishnan VV, Kesavadas C. Advanced MR imaging in Lhermitte-Duclos disease: moving closer to pathology and pathophysiology. Neuroradiology 2007; 49: 733-738.
[8]. Zhou XP, Marsh DJ, Morrison CD, Chaudhury AR, Maxwell M, Reinfenberger G, et al. Germ line inactivation of PTEN and dysregulation of the phosphoinositol-3-kinase/Akt pathway cause human Lhermitte Duclos disease in adults. Am J Hum Genet.2003; 73(5) 1191-8.
[9]. Huang S, Zhang G, Zhang J. Similar MR imaging characteristics but different pathological changes: a misdiagnosis for Lhermitte-Duclos disease and review of the literature. Int J Clin Exp Pathol. 2015; 8(6):7583-7.
[10]. Nair P, Pal L, Jaiswal AK, Behari S. Lhermitte-Duclos disease associated with dysembryoplastic neuroepithelial tumor differentiation with characteristic magnetic resonance appearance of "tiger striping". World Neurosurg. 2011; 75(5-6):699-703.
[11]. Mittal P, Gupta K, Saggar K, Kaur S. Adult medulloblastoma mimicking Lhermitte duclos disease: can diffusion weighted imaging help? Neurol india. 57(2): 203-5
[12]. Abel TW, Baker SJ, Fraser MM, Tihan T, Nelson JS, Yachnis AT, et al. Lhermitte Duclos disease: a report of 31 cases with immunohistochemical analysis of PTEN/AKT/mTOR pathway. J Neuropathol Exp Neurol. 64(4): 341-9.
[13]. Giorgianni A, Pellegrino C, De Benedictis A, Mercuri A, Baruzzi F, Minotto R, et al. Lhermitte-Duclos disease. A case report. Neuroradiol J. 2013; 26(6):655-60.
[14]. Rigert -Johnson DL, Gleeson FC, Roberts M, Tholen K, Youngborg L, Bullock M, et al. cancer and lhermitte duclos disase are common in cowden syndrome patients. Hered Cancer Clin Pract. 2010; 8(1): 6.