



IMAGING IN A CASE OF LHERMITTE DUCLOS DISEASE – DYSPLASTIC CEREBELLAR GANGLIOCYTOMA.

Radiodiagnosis

Dr. Arun George*	Junior Resident. K.S Hegde Medical Academy. Department of Radiodiagnosis K.S. Hegde Medical Academy NITTE (Deemed to be University) Deralakatte, Mangalore Karnataka, India*Corresponding Author
Dr. Rajesh Venunath	Assistant Professor. K.S Hegde Medical Academy. Department of Radiodiagnosis K.S. Hegde Medical Academy NITTE (Deemed to be University) Deralakatte, Mangalore Karnataka, India
Dr. K.V. Tejaswi	Senior Resident. K.S Hegde Medical Academy. Department of Radiodiagnosis K.S. Hegde Medical Academy NITTE (Deemed to be University) Deralakatte, Mangalore Karnataka, India
Dr. Lathika Shetty	Professor. K.S Hegde Medical Academy. Department of Radiodiagnosis K.S. Hegde Medical Academy NITTE (Deemed to be University) Deralakatte, Mangalore Karnataka, India

ABSTRACT

Lhermitte-Duclos disease (LDD) is a rare cerebellar lesion of uncertain origin. It is linked to an autosomal- dominant phakomatosis known as Cowden's disease in 40% of patients. The MRI features of LDD are almost unique and can be considered diagnostic. (1) We report on a case of 48 year old female patient who presented to us with history of seizures and bilateral lower limb weakness with the typical MRI features of the above disease. We also discuss the pathology and genetics of this rare disease.

KEYWORDS

Dysplastic cerebellar gangliocytoma, Cerebellum MRI, LDD

I. INTRODUCTION

Lhermitte-Duclos disease (LDD) or dysplastic gangliocytoma is a rare slow-growing benign lesion of the cerebellum. Its precise aetiology is uncertain. It is postulated that this disease may be a hamartoma. Recent published data suggest that LDD is a hamartomatous lesion related to a phakomatosis, rather than a neoplasm. (2)

LDD becomes usually clinically apparent in the third and fourth decade and with no obvious gender bias. The clinical manifestations are usually related to posterior fossa mass-effect and resultant secondary obstructive hydrocephalus. The most frequent complaints are headaches and ataxia due to intracranial hypertension. Cerebellar signs and symptoms are present in 40 - 50%. One-third of patients have associated cranial nerve palsies. (2)

CASE HISTORY:

A 48yr old female presented to the Neurology Department of our institution with history of recurrent seizures and bilateral lower limb weakness. Her vitals were stable and neurological examination reveals no obvious cranial nerve palsies or sensory or motor deficits. No obvious signs of cerebellar dysfunction were present in our patient. EEG was taken which shows few non specific occipital spikes. She was initially sent of CT and later MRI evaluation as a part of the seizure evaluation protocol.

II. IMAGING FINDINGS

Computed Tomography (CT):

Axial contrast enhanced CT section were obtained in GE 16 slice multidetector CT scanner. The imaging reveals an ill defined non enhancing hypo dense area of 20-25HU with loss of grey white matter differentiation in the left cerebellar hemisphere with mass effect on the adjacent fourth ventricle and mild supratentorial hydrocephalus. Imaging findings were consistent with diagnosis of cerebellar infarct however the clinical findings were not supportive of infarct and the patient was referred for MRI Brain with contrast for further evaluation.

Magnetic Resonance Imaging (MRI):

MRI brain with gadolinium was performed in a GE 1.5Tesla scanner. The study reveals a large ill defined heterogenous lobulated lesion appearing isointense on T1WI and hyperintense on T2/FLAIR sequences in the left superior cerebellar hemisphere with preserved internal striations causing partial compression of adjacent fourth

ventricle with mild supratentorial hydrocephalus with periventricular ooze. The lesion shows no significant peri lesional edema or diffusion restriction with no significant post contrast enhancement. Imaging features were consistent with Lhermitte Duclos disease or Dysplastic Cerebellar Gangliocytoma.

The patient underwent an excisional biopsy of the lesion and the microscopy reveals diffuse enlargement of molecular and internal granular layer of the cerebellum. Internal granular layer is filled with large dysplastic ganglion cells, interspersed with neuronal cells and ectatic blood vessels. The findings were consistent with Dysplastic Cerebellar Gangliocytoma Grade I.

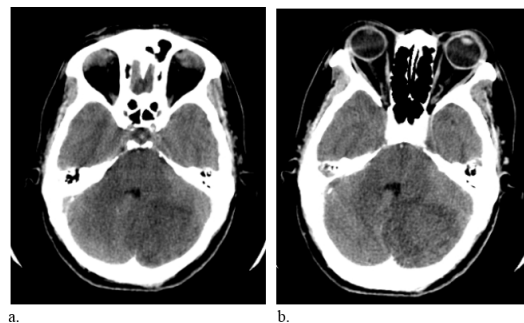


Figure 1. (a) and (b) demonstrates an ill defined non enhancing hypo dense area with loss of grey white matter differentiation in left cerebellar hemisphere with mass effect on fourth ventricle.

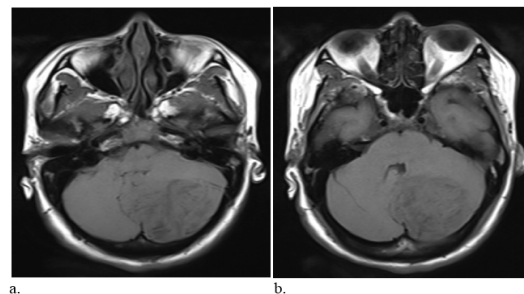


Figure.2 Axial T1WI (a) and (b) shows a large ill defined

heterogeneous lesion appearing isointense.

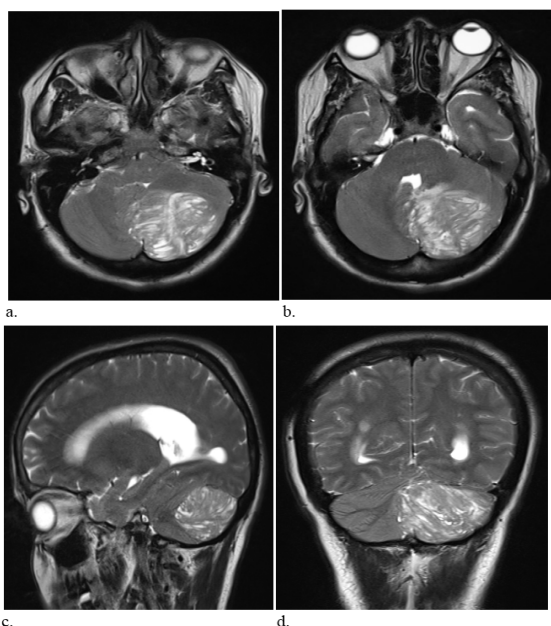


Figure 3. Axial (a and b), coronal (c) and sagittal (d) T2WI shows a lobulated hyperintense lesion with preserved internal striation in left cerebellar hemisphere.

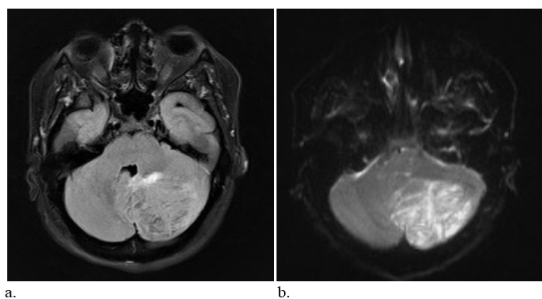


Figure 4. Axial (a) FLAIR sequence shows hyperintense lesion in left cerebellar hemisphere with preserved internal striations. (b) shows DWI with no diffusion restriction.

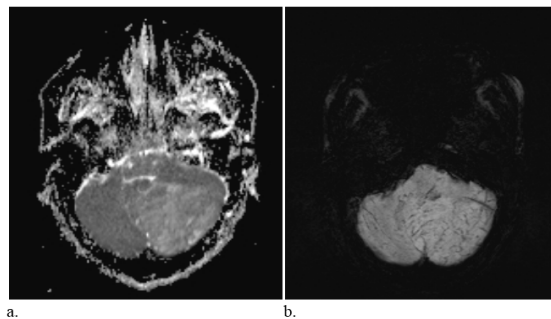


Figure 5. (a) ADC shows no matched hypointensity. (b) SWI shows no hypointense flow voids.

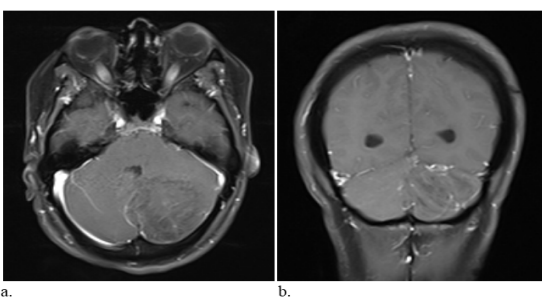


Figure 6. Axial (a) and coronal (b) Post contrast images show no enhancement.

III. Histopathological findings:

The patient underwent a partial excision biopsy of the lesion and the histopathological evaluation revealed cerebellar tissue with diffuse enlargement of molecular and internal granular layer. Internal granular layer is filled with large dysplastic ganglion cells, interspersed with neuronal cells. Features suggestive of dysplastic cerebellar gangliocytoma-Grade I.

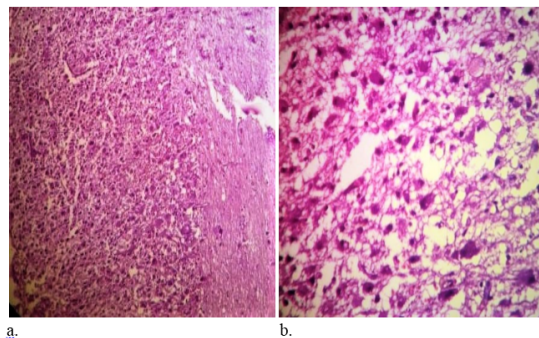


Figure 7. Slides (a) and (b) were air dried and fixed in alcohol and stained by Giemsa and Papinicolaou stain respectively. Sections showed diffuse enlargement of molecular and internal granular layer. The internal granular layer is filled with large dysplastic ganglion cells and interspersed with neuronal cells and ecstatic blood vessels are seen.

IV. DISCUSSION

The definitive diagnosis of LDD is histopathological, but MR imaging can preoperatively characterise the disease sufficiently well. The abnormality seen in LDD is related to abnormal development of the cerebellar cortex. Usually, the normal cerebellar cortex consists of an inner granular layer, an outer molecular layer and an intervening Purkinje cell layer. In LDD there are abnormal ganglion cells in the granular layer, thickening and hypermyelination of the molecular layer and loss of the middle Purkinje cell bodies. (3)

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. (1)

With the help of computed tomography, we can identify a nodular focal lesion, usually unique, located in one of the cerebellar hemispheres, iso/low attenuation, which may rarely contain calcification foci inside, without evident enhancement by iodinated contrast media and can determine thinning of the cranial cap in correspondence. In our case we were able to distinguish the lesion from an infarct due to the fact that it is not restricted to any vascular territory and lack of contrast enhancement.

Magnetic resonance imaging is the imaging test of choice for the diagnosis of Lhermitte-Duclos disease, featuring slightly heterogeneous morphological change, evidenced by thickening of the cerebellar folia, promoting mass effect and exhibiting linear bands with hypointense on T1 and alternating inner layers hyperintense and external hypointense on T2, resulting, in the latter consideration, in a typical striated pattern, which sets the cerebellar tissue the appearance of "tiger stripes". (4)(5)(6)

Lack of MR contrast enhancement is emphasised as an important MR diagnostic criterion of the disease but contrast enhancement has been reported in few cases. This is postulated to be due to venous proliferation that may also explain the large draining veins that are

seen on angiography and as flow voids on MRI. (7)

MRI is excellent in defining the limits of the lesion to accomplish the most radical excision during surgery. Despite the benign nature, surgical excision is the treatment of choice, although some centres may prefer conservative management in asymptomatic cases. Wide excision is necessary due to the risk of recurrence. However this is difficult because of ill-defined macroscopic margins. Radiation therapy is ineffective. (2)

The differential diagnosis of cerebellar dysplastic gangliocytoma is diffuse astrocytoma and desmoplastic medulloblastoma. Diffuse astrocytoma differs by more homogeneous signal change, with foci of abnormal gadolinium uptake and standard magnetic resonance spectroscopy demonstrating elevation of myoinositol in low-grade and elevated choline and lactate in high-grade lesions. The desmoplastic medulloblastoma, adult hemispherical variant presents real restriction on passive diffusivity of water molecules, high choline levels, reducing the peak of NAA to proton spectroscopy and alteration of vascular paths within the lesion to magnetic susceptibility sequence SWI. (8)(9)

V. CONCLUSION

Lhermitte-Duclos disease is characterized by dysplasia of the cerebellum which results in a tumor like mass replacing the normal architecture. MRI features are highly diagnostic with a characteristic trigroid appearance. Histopathology is confirmatory. 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.

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