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A rare case of dysplastic cerebellar gangliocytoma - Lhermitte Duclos Disease

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ABSTRACT

Lhermitte–Duclos disease (LDD), also known as dysplastic cerebellar gangliocytoma, is an uncommon hamartomatous lesion found in the cerebellar cortex. First described by French physicians Lhermitte and Duclos in 1920, this condition involves enlarged cerebellar folia containing abnormal ganglion cells. Clinical manifestations typically include headaches, visual disturbances, and cerebellar dysfunction. While imaging techniques like computed tomography (CT) scans may indicate the presence of mass lesions, magnetic resonance imaging (MRI) is crucial for accurate delineation of the lesion's boundaries and characterization of tissue involvement. Surgical intervention, typically in the form of respective surgery, remains the cornerstone of treatment for LDD. Despite advancements in imaging technologies and surgical techniques, the management of LDD remains fraught with complexities. The prognosis can be variable, and in some cases, despite appropriate therapeutic interventions guided by MRI-assisted surgical planning and confirmation through CT screening, the outcome may be unfavourable, as observed in this instance with the demise of the 50-year-old male patient. Further research and a deeper understanding of the pathophysiology underlying LDD are imperative to enhance diagnostic accuracy, refine treatment modalities, and potentially improve patient outcomes. This case underscores the challenges in managing this rare neurological condition and highlights the need for continued exploration into more effective therapeutic avenues.

Keywords: Lhermitte–Duclos disease, dysplastic cerebellar gangliocytoma, tiger striped appearance, MRI, CT

INTRODUCTION

Lhermitte-Duclos disease (LDD) or Dysplastic cerebellar gangliocytoma, is a rare hamartoma classified as a grade I mixed neuro-glial tumor by the WHO (World Health Organization). This condition arises due to abnormal cerebellar development leading to unilateral hemispheric expansion. Due to its rarity and varied presentation, diagnosis can often be overlooked [1]. Clinical manifestations usually arise from the posterior fossa mass effect, causing symptoms such as increased intracranial pressure and cerebellar dysfunction. As per existing literature, the duration of symptoms associated with this condition spans several months to more than 10 years [2]. Symptoms indicating increased intracranial pressure, such as headaches, nausea, vomiting, papilledema, alterations in mental function, and loss of consciousness, tend to manifest in the later stages of the disease due to the progressive impact of the enlarging tumor mass [3]. Imaging modalities play a pivotal role in diagnosis, with CT scans revealing hypodense, partially calcified, non-enhancing lesions [4]. However, MRI is a superior diagnostic tool, displaying a non-enhancing cerebellar lesion. On T1-weighted images, it appears hypointense, while on T2-weighted images, it exhibits hyperintensity with a characteristic 'tiger-striped' appearance, aiding in differentiation from other posterior fossa neoplasms [5,6]. Advanced MRI techniques such as diffusion-weighted imaging (DWI) and magnetic resonance spectroscopy (MRS) have also been employed to further elucidate the pathological features of LDD, providing additional information regarding tissue cellularity and metabolic activity, aiding in its differential diagnosis from other cerebellar lesions [7]. Therefore, this case report aims to underscore the diagnostic challenges and complexities in managing Lhermitte-Duclos disease, emphasizing the critical role of advanced imaging techniques in facilitating accurate diagnosis and guiding neurosurgical interventions, while also highlighting the ongoing ne-

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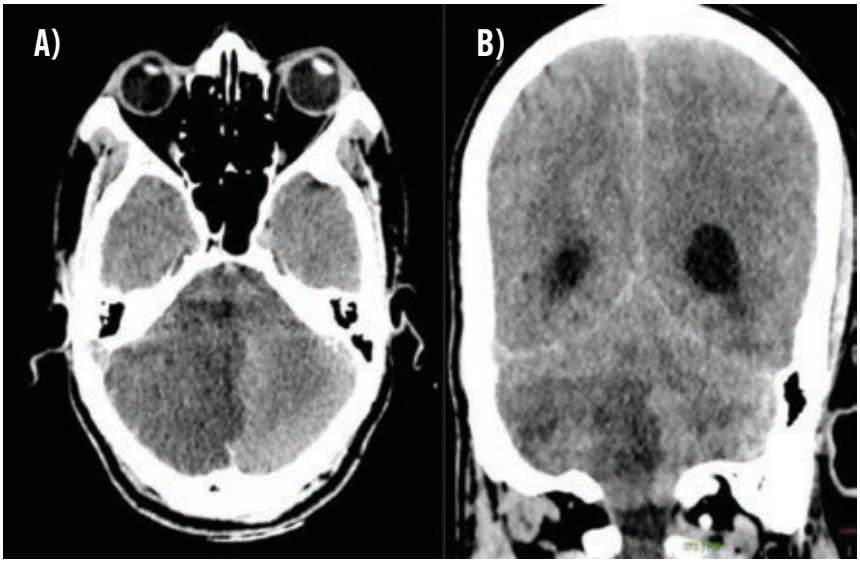

FIGURE 1. A) CT brain axial and B) CT brain coronal showing a right posterior cranial fossa lesion that is hypodense

cessity for further research to enhance therapeutic approaches and improve patient outcomes in this rare neurological condition.

CASE PRESENTATION

A 50-year-old man, previously in good health with no notable medical background, presented to the emergency department exhibiting a two-day history of persistent headache diffuse type of dull aching pain associated with blurring of vision and vomiting. He had no complaints of fever, neck pain, weakness of his limbs or seizure episodes. His relatives also gave a history of decreased responsiveness since morning.

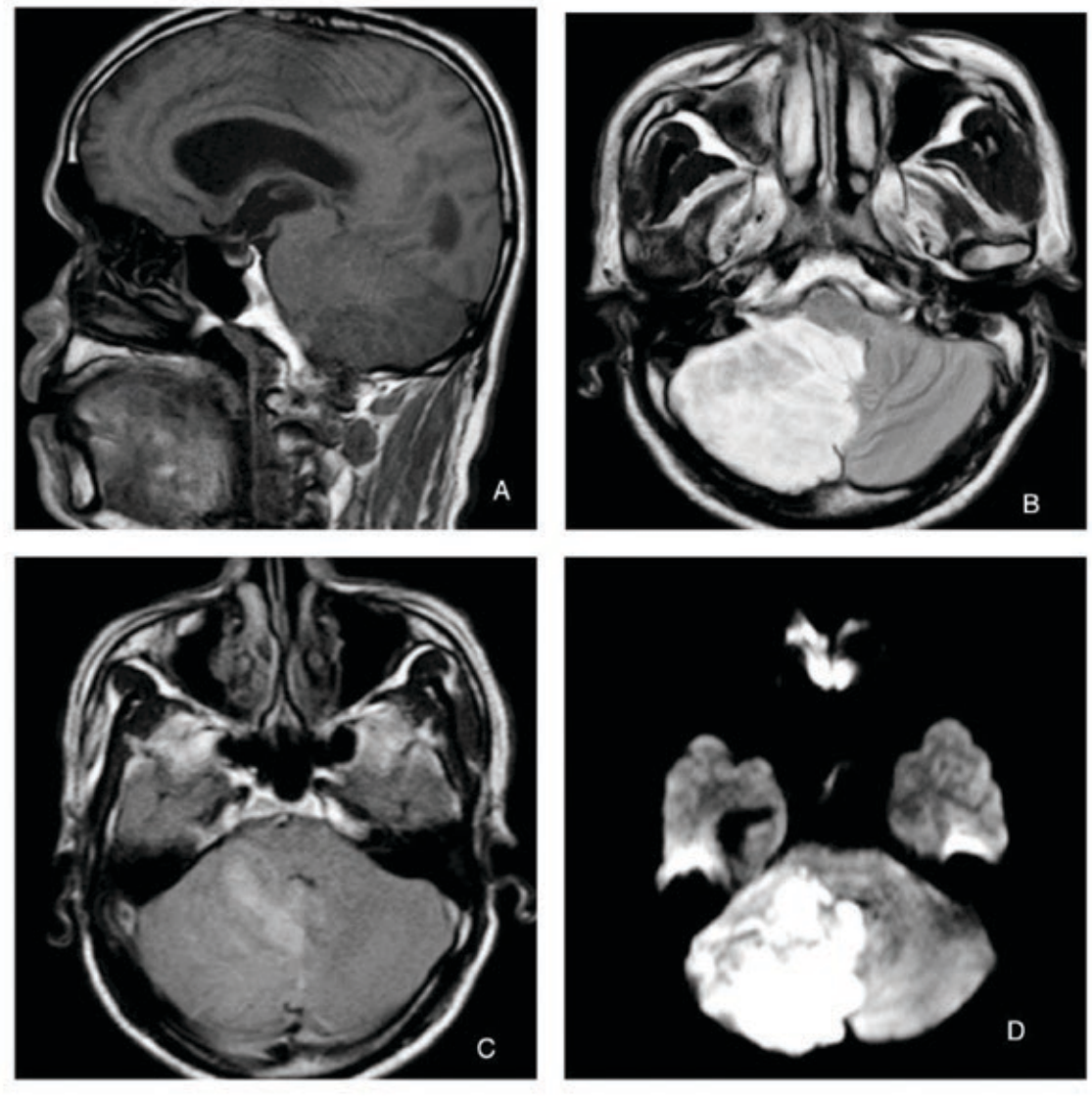

FIGURE 2. MRI brain a) T1 weighted sagittal b) T2 weighted axial image c) FLAIR axial d) diffusion-weighted showing a well-defined lesion in right cerebellum with tiger stripe appearance hypointense on T1 sequence and hyperintense on T2 sequence and bright on DWI

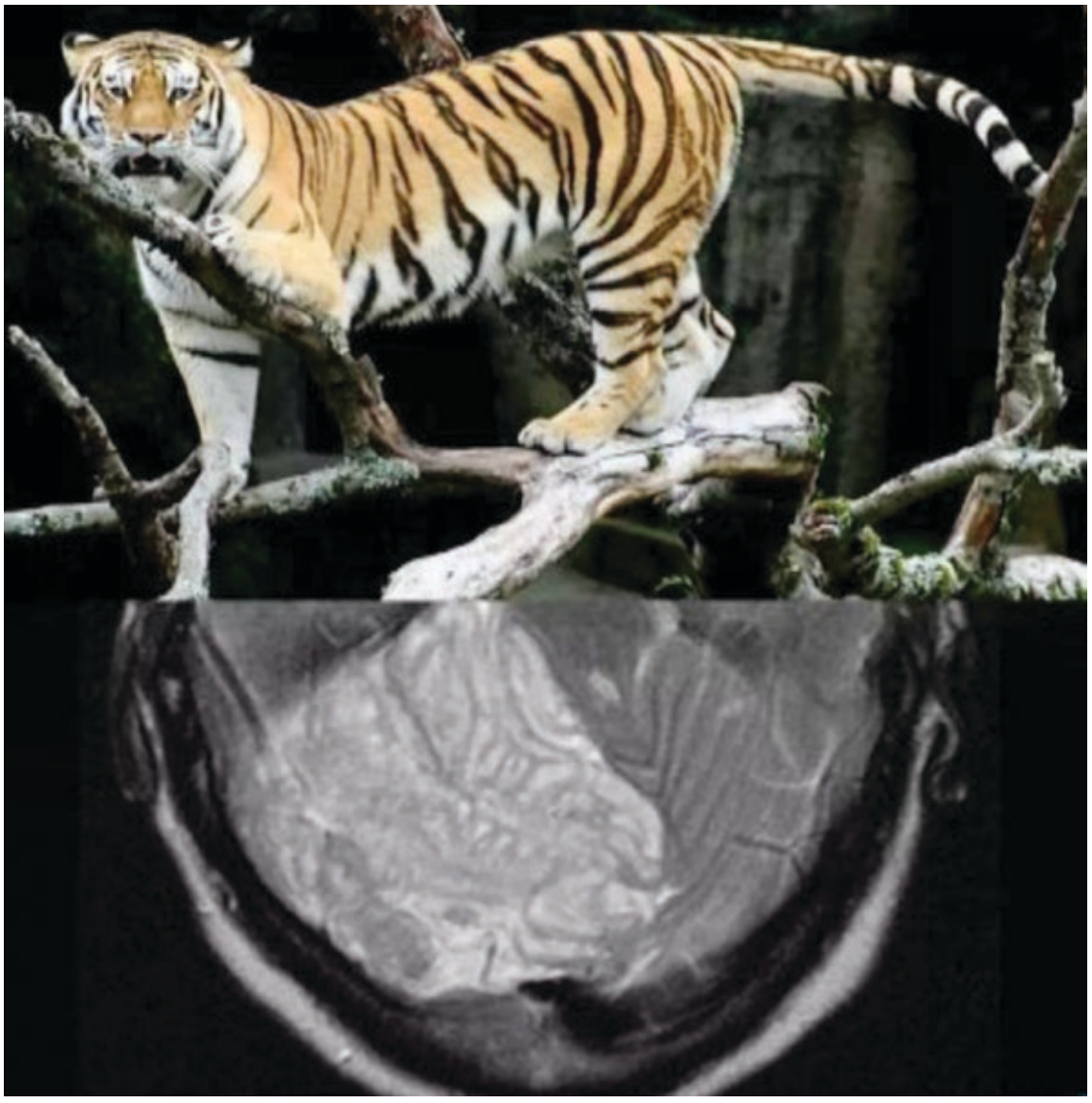

FIGURE 3. Tigroid /striated appearance

On examination, his Glasgow Coma Scale (GCS) was assessed at 10/15 (E4V1M5), and bilateral pupils were 2mm reacting to light. On Neurological examination, power was 4/5 with normal tone on bilateral upper and lower limbs with normal deep tendon reflexes and bilateral plantar flexor. A cranial nerve examination was done, and no abnormality was found. No signs of meningitis were elicited. Fundus examination revealed bilateral papilledema. Considering the low GCS and symptoms suggestive of increased intracranial pressure we went ahead with brain imaging. In the imaging studies CT scan of the brain revealed a hypodense lesion in the right posterior cranial fossa, confirming the presence of a significant mass (Figure 1).

Further characterization of the lesion was conducted through MRI imaging displaying distinct features on T1-weighted sagittal, T2-weighted axial, diffusion-weighted, and FLAIR axial sequences revealing ill-defined areas of T2/FLAIR hyperintensities predominantly affecting the right hemi-cerebellum (Figure 2).

The imaging exhibited widened cerebellar folia with a striated or tigroid appearance, along with significant mass effect causing effacement of various cerebellar and brain cisterns, mild ventricular dilation, and mild right ascending transtentorial herniation suggestive of Lhermitte Duclos Disease. The images showcased a well-defined lesion within the right cerebellum exhibiting the classic 'tiger-striped' appearance on T2-weighted imaging. All laboratory parameters were found to be within limits (Figure 3).

Neurology and Neurosurgical consultation recommended commencing prophylactic antiepileptic and anti-edema measures and planning a repeat CT scan after 24 hours. Despite conservative management in the ICU, the patient's neurological status rapidly declined, necessitating intubation and subsequent consideration for surgical intervention. Unfortunately, the patient went into asystole and was treated with appropriate resuscitation measures

but despite rigorous resuscitation attempts, he ultimately succumbed to his illness.

DISCUSSION

The presented case exemplifies the diagnostic complexities and therapeutic challenges posed by Lhermitte-Duclos disease (LDD). LDD poses diagnostic challenges due to its neoplastic and hamartomatous features, often identified in adults in their 3rd or 4th decades. There's a strong correlation with Cowden disease, indicating a higher likelihood of both benign and malignant neoplasms [8]. In this case, since the patient had a sudden downfall and died within 48 hours of hospitalization hence further genetic testing could not be done. The primary cause is a germline mutation of phosphatase and tensin homolog on chromosome 10q236. Miller et al. suggested that many individuals with LDD exhibit a loss of the PTEN allele in their germline, leading to subsequent loss of the remaining PTEN allele, consequently facilitating abnormal growth of granule cells [9]. Histopathologically, the condition involves dysplastic ganglion cells replacing the granular cell layer and Purkinje cells, resulting in global cerebellar folia thickening [5].

According to Buhl et al., individuals affected by LDD may exhibit a range of clinical presentations. Some patients might not show any symptoms, while others may experience manifestations such as ataxia, headaches, cranial nerve impairment, bouts of vertigo, cognitive decline, and in severe instances, indications of increased intracranial pressure due to hydrocephalus [10]. Typically, these patients display prolonged symptoms persisting for years, suggesting the gradual and progressive nature of the disease. In line with this characterisation, our current case report details a patient who exhibited symptoms, including headache, blurred vision, vomiting, and altered consciousness.

Imaging plays an important role in the diagnostic process. The study by Zhang et al. (2022) suggested that the integration of magnetic resonance spectroscopy along with susceptibility-weighted imaging, in conjunction with conventional MRI, represents an enhanced approach for diagnosing Lhermitte-Duclos diseases (LDDs), highlighting the "tiger-striped" pattern's specificity in differentiating LDD from these neoplasms [11].

CONCLUSION

Lhermitte-Duclos disease, an exceedingly rare benign cerebellar lesion, presents challenges in clinical and pathological diagnosis. It is crucial for appropriate management and prognosis to distinguish it from malignancies and other cerebellar malformations. The reported case underscores the challenges in navigating the diagnosis and management of LDD. Continued research and innovative diagnostic strategies are imperative to improve the understanding of this rare neurological condition and optimize therapeutic approaches for better patient outcomes.

Conflicts of interest: none declared
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