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Case Report

Lhermitte-Duclos disease: A rare case of cerebellar tumor with successful surgical treatment

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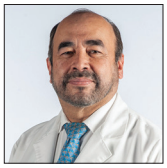

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ABSTRACT

Background: Lhermitte-Duclos disease (LDD) or dysplastic gangliocytoma of the posterior fossa is a slow-growing and extremely rare mass lesion that involves the Purkinje neurons and the granular layer of the cerebellum. It is characterized by specific neuroradiological features and secondary hydrocephalus. However, documentation of surgical experience is scarce.

Case Description: A 54-year-old man with LDD manifesting as progressive headache is presented with vertigo and cerebellar ataxia. Magnetic resonance imaging demonstrated a right cerebellar mass lesion with the characteristic "tiger-striped appearance." We decided to perform partial resection with reduction of tumor volume improving symptomatology as a result of the mass effect in the posterior fossa.

Conclusion: Surgical resection is a good alternative for the management of LDD, especially when neurological compromise exists due to mass effect.

Keywords: Cerebellum neoplasia, Dysplastic gangliocytoma, Hamartoma, Lhermitte-Duclos disease, Posterior fossa

INTRODUCTION

Lhermitte-Duclos disease (LDD), also known as dysplastic cerebellar gangliocytoma, is a rare and benign tumor of the cerebellum.[8,19] It is a rare condition, with only 230 cases reported in the literature with the characteristics of our patient, most common between the third and sixth decade of life. It was first described by Lhermitte and Duclos in 1920, and since then, only a few hundred cases have been reported worldwide.[9,12] LDD is characterized by enlarged and disorganized cerebellar folia, which is composed of hypertrophic and dysplastic ganglion cells.[6] The disease can present with a wide range of neurological symptoms, including headache, dizziness, ataxia, and visual disturbances, and diagnosis is typically made through neuroimaging and histopathological examination.[14] The diagnosis of LDD is based on radiological and histological findings, with magnetic resonance imaging (MRI) being the most useful diagnostic tool.[3,5,11] Treatment options for LDD are limited and surgical resection is often the primary treatment modality in symptomatic patients. However, the optimal surgical approach remains controversial,

as complete resection is often difficult to achieve and may be associated with significant morbidity.[9] In this case report, we present the case of a 54-year-old male with LDD who presented with a range of neurological symptoms. Through this case, we aim to highlight the importance of surgery as a method of resolving the patient's clinical condition in the treatment of LDD.

CASE DESCRIPTION

A 54-year-old man with a history of systemic arterial hypertension was admitted to the hospital because of a sudden onset of the severe persistent occipital headache of 2 months of evolution accompanied by vertigo and cerebellar ataxia. The neurological examination revealed the presence of hypotonia of the right hemibody, limitation to infraduction and adduction of the right eye, and multidirectional nystagmus in both eyes. Signs and symptoms compatible with increased intracranial pressure were also found, such as headache, nausea, and vomiting; no clinical data were found such as macroglossia, polydactyly, gigantism, or hydromyelia. Initial brain MRI showed a right cerebellar hemisphere sharply defined mass of 7.6 cm × 7.6 cm × 8.9 cm, with a prominent striated foliar pattern, causing obstructive hydrocephalus. The T1 appeared a hypointense, and T2 was a hyperintense-weighted image with parallel linear striations with a characteristic "tiger-striped appearance" visible on T2-weighted imaging [Figures 1 and 2]. It was considered a distortion of the fourth ventricle along with a compression of the bridge and medulla oblongata is also observed. From the classical findings on neuroimaging, a diagnosis of LDD was made. Surgery was planned in a Concorde position. A right lateral sub-occipital craniotomy was performed to expose the tentorial surface, incising the midline to the edge of the foramen magnum. The dural opening was performed in a Y-shape and the cisterna magna was subsequently drained. On surgical exposure, the right cerebellar hemisphere was pale to grayish in color with an enlarged and widened folium with appearance of venous ingurgitation. The lesion was diffuse and moderately vascular with poorly defined margins under magnification using a Kinevo 900 surgical microscope (Carl Zeiss, Germany). The resection was completed using bipolar forceps and conventional suction. During the surgical procedure, we perform volume reduction with preservation of the integrity of cerebellar nuclei as much as possible, thus keeping the patient's functionality and resolving symptomatology. An intraoperative frozen section analysis in open biopsy of lesion was performed, reporting chronic inflammatory tissue. The surgical specimen measuring 4.5 cm × 2.5 cm × 1.3 cm was sent to the neuropathology area for analysis, and 24 h later, a histopathological report

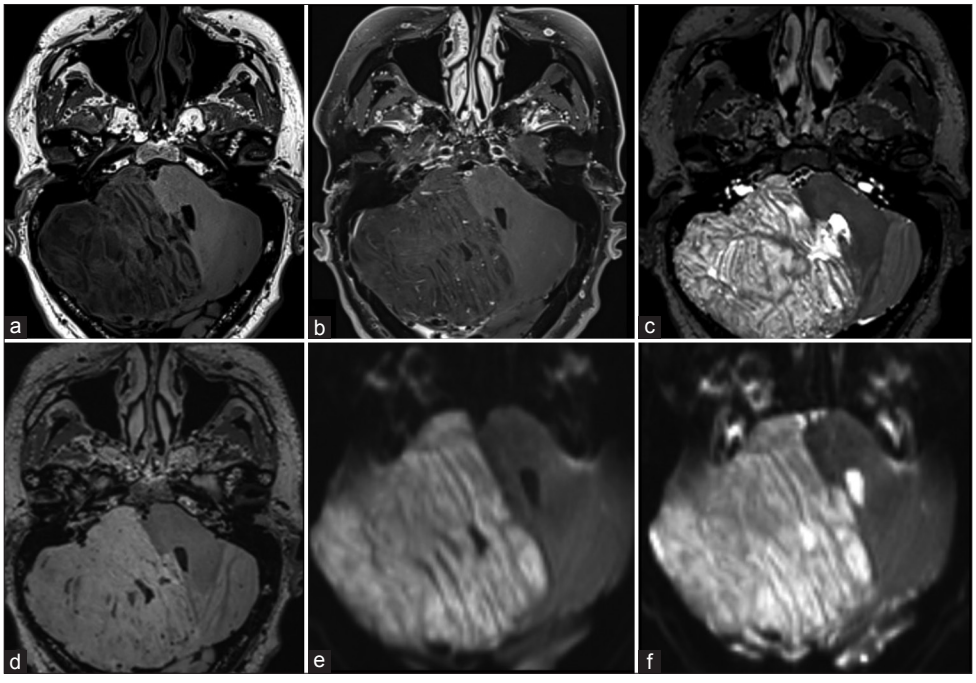

Figure 1: Preoperative axial magnetic resonance imaging. It shows a predominantly right hemispheric lesion, a characteristic laminar appearance is observed due to abnormal thickening of the cerebellar folia ("tiger-stripe appearance"). In T1-weighted (a) the lesion shows hypointensity, in the contrasted T1-weighted (b) no contrast-enhancement, no edema in T2-weighted (c) and fluid attenuated inversion recovery-weighted (d), no diffusion restriction, and no calcifications or hemorrhage in the susceptibility (e and f).

was obtained describing the presence of a solid neoplasm expanding the cerebellar folia at the expense of proliferating dysplastic ganglion cells with large nuclei of granular chromatin with the presence of positive glial fibrillary acidic protein in reactive astrocytes and positive neuronal specific enolase in dysplastic neurons [Figure 3]. These neurons were highlighted by NeuN immunohistochemistry consistent with a dysplastic cerebellar gangliocytoma, also called LDD, as the World Health Organization (WHO) Grade I according to the 2016 WHO Classification of Tumors of the Central Nervous System.[10] A genetic diagnosis was not obtained. However, it is not part of a diagnostic criterion for the disease; suspicion was mainly based on histopathology and imaging for topographic and definitive diagnosis.[14,16] He was discharged on the 5th postoperative day with significant improvement in his symptomatology. At 1-month follow-up, the ataxia had disappeared, and the headache was minimal.

DISCUSSION

The incidence is extremely rare and not reported. The disease should be suspected in a young adult in the third or fourth decade of life who presents with clinical signs and symptoms of a progressive posterior fossa mass.[14] The disease has also been reported in association with Cowden syndrome, an autosomal dominant disorder characterized by multiple hamartomas and an increased risk of certain cancers.[16,18] While the etiology of LDD remains unclear, studies have suggested that it may be associated with germline mutations in the PTEN gene, which is involved in the regulation of cell growth and division; however, it is not part of a diagnostic criterion for the diagnosis of LDD.[11,16,17] In the particular case of our patient, the presence of another hamartoma in some other location was ruled out. When the lesion is not diagnosed and treatment is delayed, the lesion may grow, compress the brain, cause a mass effect, and elevated intracranial pressure, warranting surgical removal.[3] The usual clinical presentation of these patients is with gradual deterioration with cerebellar deficits, such as diplopia, ataxia, and gait disorders, as well as hydrocephalus due to obstruction of the passage of cerebrospinal fluid in the fourth ventricle.[2,13,15] Growth of the cerebellar cortex gives rise to a thickened folia, which loses its architecture resulting in asymmetric expansion of the cerebellar hemisphere.[17] Although, for disease confirmation, a typical "tiger-striped appearance" of hyperintensity on T2-weighted images and corresponding the abnormal thickening of the cerebellar folia affected by the tumor on MRI is required.[1,4,14] Nonspecific histologic findings include dysplastic ganglion cells, microscopic expansion of the granular cell layer with abundant cytoplasm, large nuclei with prominent nucleoli and granular chromatin, and absence of the Purkinje cell layer.[3,17,19,20] Therefore, imaging is of great significance for

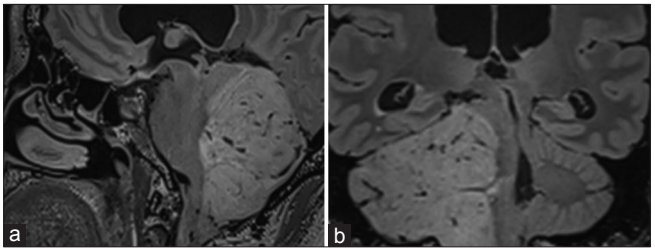

Figure 2: Magnetic resonance imaging sagittal and coronal projections. (a and b) fluid attenuated inversion recovery-weighted; infiltration is observed in the inferior, middle, and superior cerebellar peduncles, displacing the fourth ventricle and narrowing the anterior and lateral cisterns of the posterior fossa.

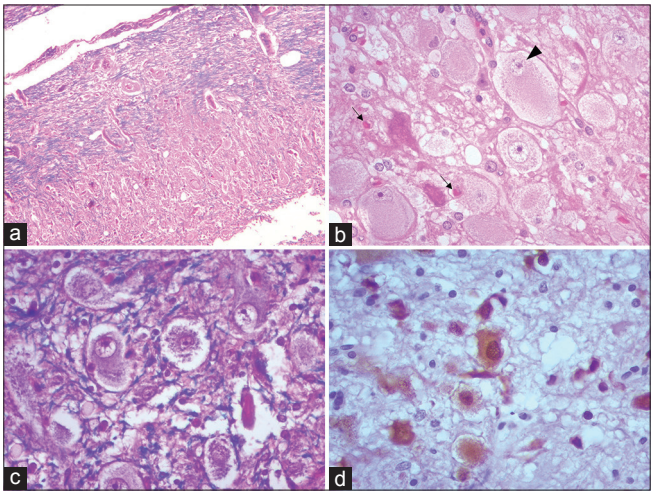

Figure 3: Photomicrographs of Lhermitte-Duclos disease showing the spectrum of changes. (a) Cerebellar cortex replaced by the presence of dysplastic cells characteristic of the disease, encompassing virtually all three histologic strata. H and E ×100 (b) Cluster of dysplastic cells with a large eosinophilic cytoplasm of globoid appearance, with a large nucleus, slightly recessed to the periphery. (Black arrowhead) Amphophilic nucleolus. (black arrows) Open neuropil, with edema and vacuolation. Alternates with apoptotic appearing neurons. H and E ×400 (c) Kluver-Barrera/H and E staining showing the direction in different directions of axons and loss of layering of neuronal somas. Note that the fibers are tortuous, serpiginous, and disorganized (blue). ×400 (d) Neuronal bodies of different sizes and morphology (brown). In addition, little gliosis or satellitosis was evident. IHC NeuN ×400.

confirming a disease diagnosis.[7] Treatment options for LDD are limited and the optimal management strategy remains controversial. Conservative management, observation, and permanent follow-up can be options in cases of small cerebellar lesions and clinically stable patients without neurological compromise because of the slow-growing pattern of this lesion. Surgery is the mainstay of treatment of cerebellar disorders and complete resection of the tumor is usually curative; however, natural history in some cases results in progressive growth of the tumor mass,[14,17] which

can be detected in early childhood, should be considered in the differential diagnosis of posterior fossa lesions, and can mimic low-grade glial tumors or infection diseases.[17]

CONCLUSION

This case represents a diagnostic challenge due to its low frequency. This case documents the surgical and functional success obtained by the patient, resolving his symptoms and his disease. Surgical resection is a good alternative for the management of LDD, especially when neurological compromise exists due to mass effect.

Declaration of patient consent

Patient's consent not required as patient's identity is not disclosed or compromised.

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Conflicts of interest

There are no conflicts of interest.

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