



Posterior Fossa Intra-Axial Lesion: There Are Pathologies Beyond Metastases

George Fotakopoulos¹ Alexandros G. Brotis¹ Konstantinos Paterakis¹ Eftychia Kapsalaki²
Kostas N. Fountas¹

¹Department of Neurosurgery, General University Hospital of Larissa, Larissa, Greece

²Department of Radiology, University of Thessaly, Medical School of Thessaly, Thessaly, Greece

Address for correspondence: George Fotakopoulos, MD, MSc, PhD, Department of Neurosurgery, General University Hospital of Larissa, Vounargo, Pyrgou, Ilias, 27100, Greece (e-mail: gfotakop@yahoo.gr).

AJNS 2022;17:680–682.

Abstract

Keywords

- ▶ Lhermitte–Duclos disease
- ▶ dysplastic ganglioglioma of the cerebellum
- ▶ magnetic resonance spectroscopy
- ▶ asymptomatic cases
- ▶ cerebellum

Cerebellar metastases represent the most common lesion requiring surgical treatment in the daily neurosurgical praxis. We report a rare case with Lhermitte–Duclos disease presenting to the neurosurgical outpatient department and describe our diagnostic and treatment challenges. A 57-year-old man presented with transient cerebellar manifestations, including walking difficulties, dizziness, and a persistent occipital headache. The magnetic resonance imaging demonstrated a high-intensity lesion in the right cerebellar hemisphere on T2-weighted images. We initially considered surgery to establish a histopathological diagnosis. However, the magnetic resonance spectroscopy was compatible with a dysplastic gangliocytoma of the cerebellum. Therefore, we preferred a more conservative approach. Posterior fossa contrast-enhancing lesions are much more than metastases. The role of advanced magnetic resonance in avoiding unnecessary surgical interventions is priceless.

Introduction

Intra-axial posterior fossa lesions constitute a common problem for the neurosurgeon. The differential diagnosis differs between adults and children significantly. Metastatic lesions to the cerebellum remain the most common diagnosis in adults.¹ Lungs, breasts in females, kidneys, and the gastrointestinal tract frequently harbor the primary lesion.²

Alternatively, several other neoplasms may be encountered less frequently as intraparenchymal lesions of the cerebellum. The differential diagnosis mainly includes hemangioblastomas, pilocytic astrocytomas, medulloblastomas, and ependymomas.³ Likewise, infections, such as cer-

ebellitis⁴ and cerebellar infarction,⁵ may also present as space-occupying lesions of the cerebellum.

Depending on the diagnosis of the primary lesion, the radiological appearance, its spread, and the patient's general performance, surgery provides the most effective treatment option. Herein, we present the rare case with Lhermitte–Duclos disease (LDD) and discuss our diagnostic approach and treatment challenges.

Case Report

A 57-year-old man presented at the outpatient department complaining about transient difficulty in walking, dizziness,

article published online
October 29, 2022

DOI <https://doi.org/10.1055/s-0042-1757433>.
ISSN 2248-9614.

© 2022. Asian Congress of Neurological Surgeons. All rights reserved.

This is an open access article published by Thieme under the terms of the Creative Commons Attribution-NonDerivative-NonCommercial-License, permitting copying and reproduction so long as the original work is given appropriate credit. Contents may not be used for commercial purposes, or adapted, remixed, transformed or built upon. (<https://creativecommons.org/licenses/by-nc-nd/4.0/>)

Thieme Medical and Scientific Publishers Pvt. Ltd., A-12, 2nd Floor, Sector 2, Noida-201301 UP, India

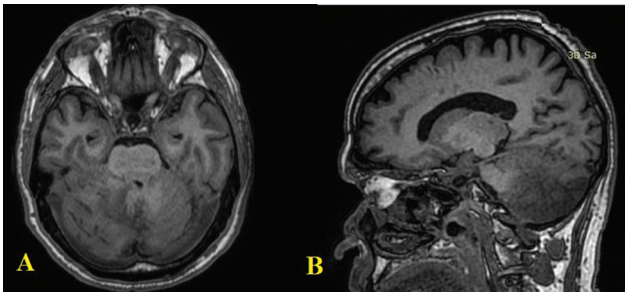


Fig. 1 The characteristic gyriform pattern with enlargement of cerebellar folia for Lhermitte-Duclos disease shown in T1-weighted (A) axial and (B) sagittal magnetic resonance imaging scans.

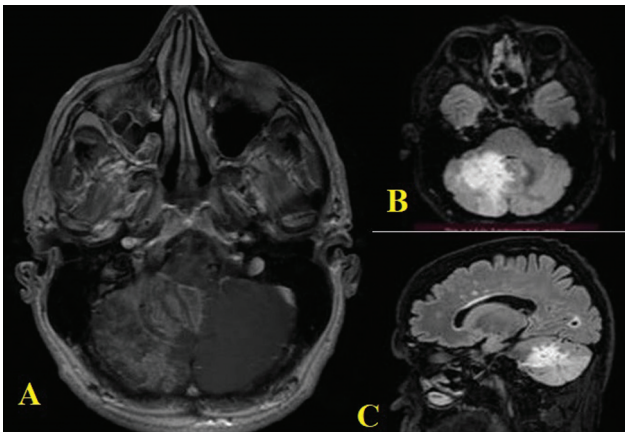


Fig. 2 (A) Magnetic resonance imaging scan hypointense on T1-weighted images; (B) axial and (C) sagittal hyperintense on T2 (figure) weighted images, with parallel linear striations.

and a persistent occipital headache. Physical examination identified cerebellar ataxia with positive cerebellar signs.

The T2-weighted magnetic resonance imagings (MRIs) demonstrated a high-intensity lesion at the right cerebellar hemisphere without obstructive hydrocephalus (► **Figs. 1, 2**). At the lesion's periphery, there were several indolent venous

dilations. We initially considered surgery to establish a histopathological diagnosis and decompress the posterior fossa neural structures.

Awaiting surgery, the patient's symptomatology improved with minimal doses of steroids. Based on the prominent striated folial pattern, an experienced neuroradiologist raised the suspicion for a dysplastic ganglioglioma of the cerebellum. Hence, the patient underwent a second MRI, including advanced imaging techniques. Cerebellar infarction and cerebellitis were excluded due to the absence of abnormalities in the MR perfusion and diffusion, respectively. Similarly, highly destructive lesions such as metastases were rejected since the MRI spectroscopy showed a marginally increased choline to creatinine ratio (► **Fig. 3**). Of note, the levels of N-acetyl aspartate were at the lower average values compared with the other hemisphere, and the lipid concentration was within normal values.

Finally, we preferred a more expectant approach since the patient's symptomatology improved. There were no clinical manifestations or positive family history for Cowden's disease. The chest and abdominal computed tomography findings were unremarkable. Therefore, the patient is scheduled for follow-up every 6 months at the outpatient neurosurgery department with regular MRIs.

Discussion

Dysplastic ganglioglioma of the cerebellum, also known as LDD, was initially described in 1920.⁶ It is characterized by enlarged cerebellar folia with abnormal ganglion cells in the granular layers of circumscribed cerebellar regions. To date, less than 300 cases have been described globally. It commonly affects males and females equally, particularly in the third and fourth decades of life.

In rare cases, there is familial predisposition.⁷ LDD is considered a member of the disease spectrum known as Cowden-Lhermitte-Duclos syndrome, associated with PTEN (phosphatase and TENsin homolog deleted on chromosome 10) germline mutations.^{8,9} The complete syndrome includes

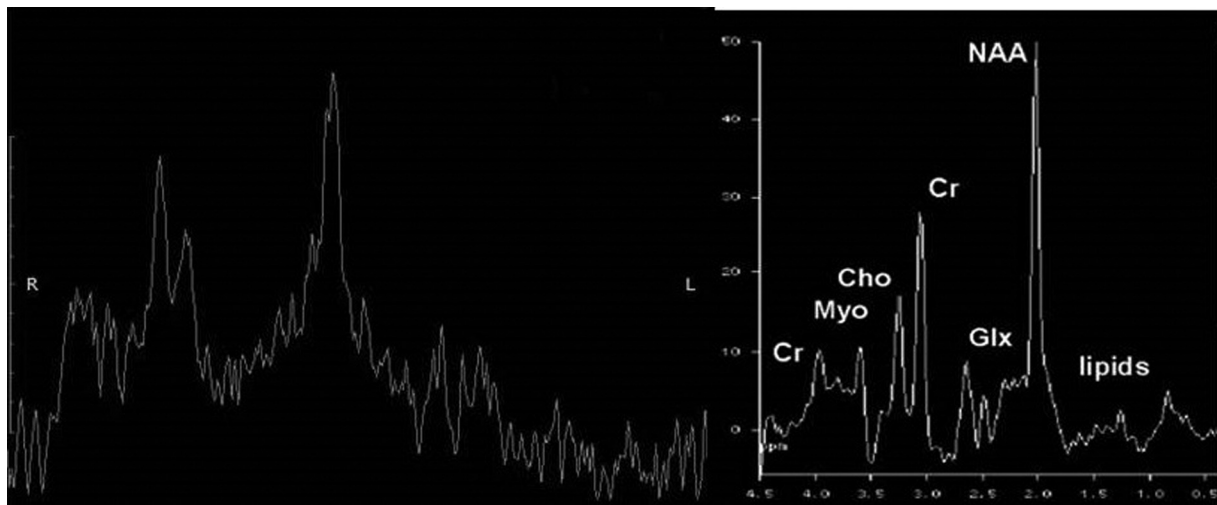


Fig. 3 The magnetic resonance spectroscopy demonstrates an elevated level of lactate and decreased levels of choline and myoinositol.

mucocutaneous lesions, thyroid diseases, and breast and ovarian tumors.¹⁰

LLD commonly presents with posterior fossa clinical manifestations, such as ataxia, hydrocephalus, and cranial nerve palsies. The MRI shows the typical tiger-striped appearance. There is a regionally increased cerebral blood volume in perfusion images. MR spectroscopy reveals an increased level of lactate and decreased level of myoinositol and N-acetyl-aspartate, as observed in low-grade gliomas but with decreased choline levels.¹¹ Undoubtedly, the definitive diagnosis is always set from the histopathology results. However, all currently available evidence shows that MRI, including magnetic resonance spectroscopy, could safely set the diagnosis.¹² Unfortunately, there is no diagnostic accuracy data on the performance of MRI in LDD, since the available evidence originate from a limited number of case reports, due to the rarity of the clinical entity under study. On top of that, an expectant approach with a regular patient follow-up seems to be a more legally defensible strategy than to proceed with an aggressive posterior fossa surgery, considering the potential perioperative risk the patient's preference.

In symptomatic cases, surgical resection remains the treatment of choice. However, the optimal treatment is under debate regarding asymptomatic cases due to the ill-defined lesion's margins. Another indication for surgery is to establish a definite histological diagnosis. The latter shows thickening and hypermyelination of the outer molecular layer, loss of Purkinje cells and white matter, dysplastic ganglion cells with rounded nuclei, and abundant mitochondria invading the inner granular layer.⁵ The prognosis after surgery is encouraging, even though rare occasions of tumor recurrence have been described.¹³

Conclusion

The management of posterior fossa lesions requires a multidisciplinary approach, including an experienced neuroradiologist. LDD should be considered in the differential diagnosis of intra-axial posterior fossa lesions. Advanced MRI is priceless in asymptomatic cases to avoid unnecessary surgical interventions.

Ethical Approval

The study was approved by the institutional ethics committee.

Funding

None.

Conflict of Interest

None declared.

Informed Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given consent for images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal his identity, but anonymity cannot be guaranteed.

References

- 1 Ambler M, Pogacar S, Sidman R. Lhermitte-Duclos disease (granule cell hypertrophy of the cerebellum) pathological analysis of the first familial cases. *J Neuropathol Exp Neurol* 1969;28(04): 622–647
- 2 Barnholtz-Sloan JS, Sloan AE, Davis FG, Vigneaun FD, Lai P, Sawaya RE. Incidence proportions of brain metastases in patients diagnosed (1973 to 2001) in the Metropolitan Detroit Cancer Surveillance System. *J Clin Oncol* 2004;22(14):2865–2872
- 3 Coeli GNM, Silva GC da, Tiengo RR, Fernandes JOM, Fernandes WLM, Lourenço DNL. Acute cerebellitis with tonsillar herniation: a case report. *Radiol Bras* 2012;45:244–246
- 4 De Cocker IJL, Lövblad K-O, Hendrikse J. MRI of cerebellar infarction. *Eur Neurol* 2017;77(3–4):137–146
- 5 Dragoo DD, Taher A, Wong VK, et al. PTEN hamartoma tumor syndrome/Cowden syndrome: genomics, oncogenesis, and imaging review for associated lesions and malignancy. *Cancers (Basel)* 2021;13(13):3120
- 6 Grossman R, Ram Z. Posterior fossa intra-axial tumors in adults. *World Neurosurg* 2016;88:140–145
- 7 Klisch J, Juengling F, Spreer J, et al. Lhermitte-Duclos Disease: Assessment with MR Imaging, Positron Emission Tomography, Single-photon Emission CT, and MR Spectroscopy. *Am J Neuro-radiol* 2001;22(05):824–830
- 8 Lhermitte J, Duclos P. On a diffuse ganglioneuroma of the cortex of the cerebellum. *Bull Assoc Franc Cancer* 1920;9:99–107
- 9 Lloyd KM II, Dennis M. Cowden's disease. A possible new symptom complex with multiple system involvement. *Ann Intern Med* 1963;58:136–142
- 10 Nieder C, Spanne O, Mehta MP, Grosu AL, Geinitz H. Presentation, patterns of care, and survival in patients with brain metastases: what has changed in the last 20 years? *Cancer* 2011;117(11): 2505–2512
- 11 Padberg GW, Schot JDL, Vielvoye GJ, Bots GT, de Beer FC. Lhermitte-Duclos disease and Cowden disease: a single phakomatosis. *Ann Neurol* 1991;29(05):517–523
- 12 Zhang HW, Zhang YQ, Liu XL, et al. MR imaging features of Lhermitte-Duclos disease: case reports and literature review. *Medicine (Baltimore)* 2022;101(04):e28667
- 13 Stapleton SR, Wilkins PR, Bell BA. Recurrent dysplastic cerebellar gangliocytoma (Lhermitte-Duclos disease) presenting with sub-arachnoid haemorrhage. *Br J Neurosurg* 1992;6(02):153–156