EDORIUM Journals

Int J Case Rep Images 2019;10:101041Z01HB2019.
www.ijcasereportsandimages.com

CASE REPORT PEER REVIEWED | OPEN ACCESS

Unique association of Lhermitte–Duclos disease and frontal encephalocele

Habib Bellamlih, Aymane El Farouki, Moulay Rachid El Hassani, Mohammed Jiddane, Meriem Fikri

ABSTRACT

Lhermitte–Duclos disease, also known as dysplastic gangliocytoma of the cerebellum, is an extremely rare cerebellar tumor characterized by an enlargement of cerebellar folia. Its etiology remains controversial. Imaging modalities, especially magnetic resonance imaging, contribute to establish a correct and accurate diagnosis with pre and postoperative assessment. The characteristic "tiger striping" appearance is pathognomonic. The recognition of this unusual and benign condition is particularly important, given the possible association with Cowden's syndrome and therefore the need to identify concurrent malignancies.To our knowledge, we describe a rare and unique presentation of histologically proven Lhermitte–Duclos disease associated with a frontal encephalocele surgically treated.

Keywords: Dysplastic gangliocytoma, Frontal encephalocele, Lhermitte–Duclos disease, Magnetic resonance imaging

How to cite this article

Bellamlih H, El Farouki A, El Hassani MR, Jiddane M, Fikri M. Unique association of Lhermitte–Duclos disease and frontal encephalocele. Int J Case Rep Images 2019;10:101041Z01HB2019.

Habib Bellamlih[1], Aymane El Farouki[1], Moulay Rachid El Hassani[1], Mohammed Jiddane[1], Meriem Fikri[1]

Affiliation: [1]Department of Neuroradiology, Ibn Sina Hospital, Mohamed V University, Rabat, Morocco.

Corresponding Author: Habib Bellamlih, Ibn Sina Hospital, Mohamed V University, Rabat, Morocco; Email: bellamlih-habib@gmail.com

Received: 06 February 2019
Accepted: 24 June 2019
Published: 29 July 2019

Article ID: 101041Z01HB2019

****

doi: 10.5348/101041Z01HB2019CR

INTRODUCTION

Lhermitte–Duclos disease (LDD) or dysplastic gangliocytoma (DG) of the cerebellum is a rare pathological cerebellar entity with features of both hamartoma and benign neoplasm [1].

It typically presents with chronic headaches, gait disturbance, and cranial nerve dysfunctions. Until recently, the diagnosis of LDD was made only postoperatively [2].

Currently, the characteristic magnetic resonance imaging (MRI) features, avoid the need for a diagnostic biopsy [3]. It appears as nonenhancing lesion of the cerebellar hemisphere with a gyriform "tiger-striped" appearance. Surgical excision remains the mainstay of treatment [1, 2]. Over the past decade, the association with Cowden syndrome (CS) has been known with increasing frequency [4].

To our knowledge, no instance of frontal encephalocele associated with LDD has been described thus far. Here, we report the first case of a frontal encephalocele in an adult patient with LDD.

CASE REPORT

A 37-year-old man was referred to our department for persistent and chronic headache associated with dizziness. On admission, neurological examination revealed a nystagmus, a cerebellar gait ataxia with tendinous hyperreflexia, and vibratory hemihypesthesia. The cognitive tests were normal.

Computed tomography scan showed a hypodense and nonenhancing left cerebellar lesion without calcifications compressing the brain stem and effacing the fourth ventricle, responsible for a passive triventricular hydrocephalus (Figure 1). In the left frontal sinus, there

was an isodense mass communicating with the cranial cavity through a posterior wall defect.

At the MRI, the cerebellar lesion appeared hypointense on T1-weighted sequence with linear striations. The lesion was hyperintense with areas of linear hypointensity on T2-weighted sequence, associated with a passive hydrocephalus. The parallel linear corresponding to the particular "tiger striping" aspect. Contrast-enhanced T1-weighted MRI showed no enhancement of the linear striations. On diffusion, there was a high signal intensity of the mass in weighted images. Sagittal MRI revealed a small tonsillar herniation up to the C1 posterior arch without any signs of syringomyelia (Figure 2). The spectroscopic analysis revealed an increased Cho/Cr ratio, lactate peak, and reduction in the choline peak.

Magnetic resonance imaging demonstrated also, a frontal sinus mass isointense to adjacent brain, confirming the diagnosis of left frontal encephalocele (Figure 3).

A left paramedian suboccipital craniectomy revealed a yellowish gray avascular tumor of soft to mildly tenacious consistency. Gross total removal of the tumor was achieved. The histologic examination was compatible with LDD. A subsequent resection of the encephalocele has been programed six months later. The encephalocele

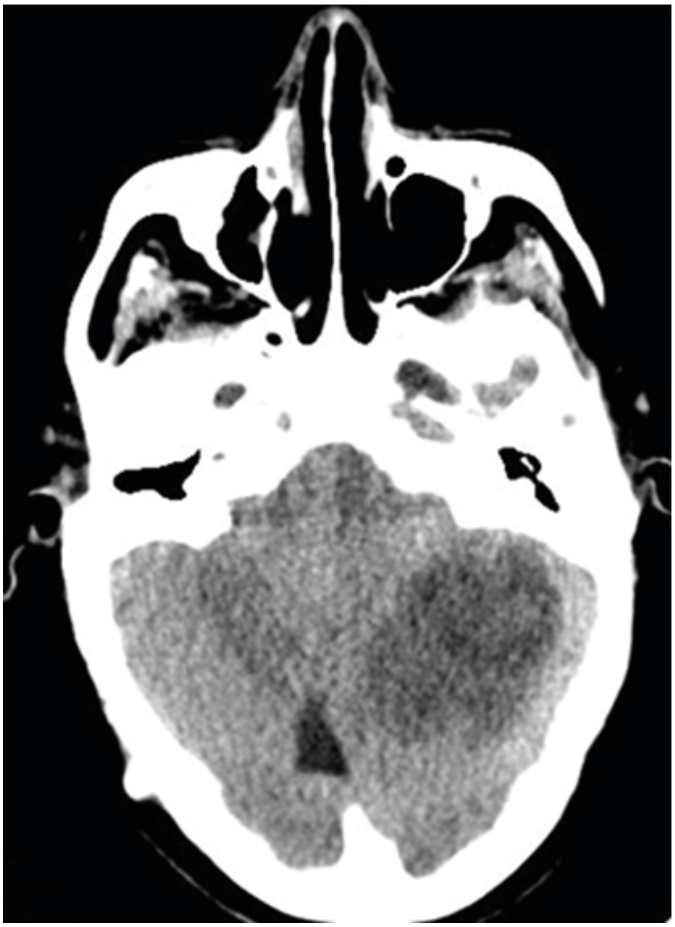


Figure 1: Computed tomography scan: hypodense and nonenhancing left cerebellar lesion compressing the brain stem and effacing the fourth ventricle.

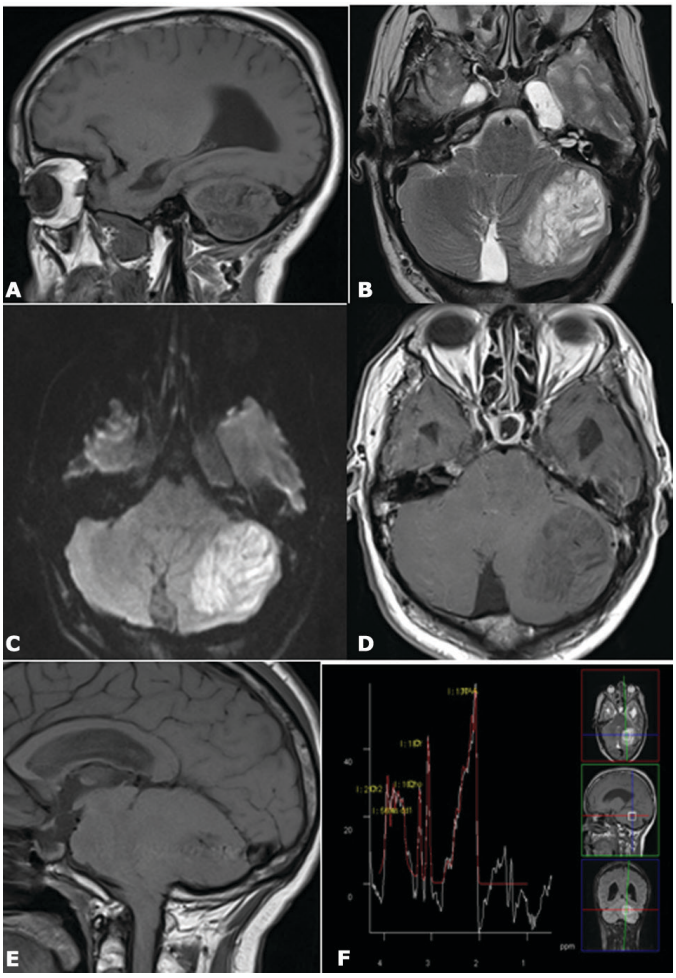


Figure 2: Brain MRI. (A) Sagittal T1-weighted sequence: left cerebellar hypointense lesion with obstructive hydrocephalus. (B) Axial T2-weighted sequence: left cerebellar "tiger-striated" hyperintense lesion. (C) Diffusion-weighted sequence: high signal intensity of the left mass. (D) Axial T1-weighted sequence with gadolinium: no enhancement of the linear striations. (E) Axial T1-weighted sequence: tonsillar herniation up to the C1 posterior arch without any signs of syringomyelia. (F) The spectroscopic analysis: an increased Cho/Cr ratio, lactate peak, and reduction in the choline peak.

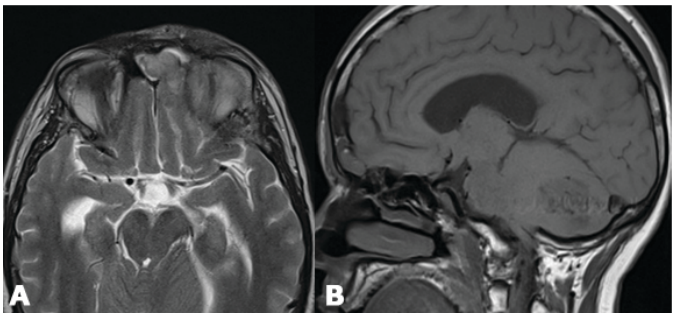


Figure 3: Brain MRI. (A) Axial T2-weighted sequence. (B) Sagittal T1-weighted sequence: a left frontal sinus mass isointense to adjacent brain, communicating with the cranial cavity through a posterior wall defect: left frontal encephalocele.

was encountered within the left frontal sinus protruding through the posterior table and dural defects. The encephalocele was resected and the dural defect closed. The postoperative course was unremarkable. The patient remained symptomfree at a 12-months follow-up examination.

DISCUSSION

Lhermitte–Duclos disease is a highly unusual and controversial condition characterized by a circumscribed enlargement of cerebellar folia.

Lhermitte and Duclos first reported the disorder in 1920. They described this rare tumor as a combination of a congenital malformation and a neoplasm arising from ganglion cells [5]. Other observers of this particular entity suggested the lesion to be a "hamartoma" or "hyperplasia" [1]. However, the debate about the exact ethiopathogeny of this entity is still in progress.

Dysplastic gangliocytoma of the cerebellum most frequently presents in the third and fourth decades of life, but the age at clinical manifestation ranges from birth to the sixth decade. There is no obvious sex predominance [1, 6–8].

Clinically, the duration of symptoms ranges from a few months to more than 10 years. Typically, the cause of clinical presentation is a posterior fossa mass lesion with headaches, cerebellar ataxia, visual disturbances, and other cranial nerve palsies. Symptoms of increased intracranial pressure, such as headaches, vomiting, papilledema, mental disturbances, and loss of consciousness occur in a later stage of the disease caused by the progressive mass effect of the growing lesion [1, 6].

Isolated orthostatic hypotension and tinnitus are unusual symptoms that can be associated with LDD. Occasionally patients present with sudden neurological deterioration, either spontaneously or following surgical intervention, presumably because of acute or decompensated chronic occlusive hydrocephalus [1, 6, 7].

Imaging plays a determinant role in the diagnostic of DG [1]. The tumor appears hypoattenuated with no appreciable enhancement on computed tomographic (CT). The diagnostic clue, in such case, is the mass effect, which manifests as compression of the fourth ventricle, effacement of the cerebellopontine angle cistern and hydrocephalus. However, CT remains of limited value in the exploration of a posterior fossa lesion due to the beam-hardening artifacts caused by the petrous temporal bone that obscure the details [1, 9].

Conventional MRI, associated with diffusion imaging and spectroscopy, is actually the most imaging method able to furnish a definite and preoperative diagnosis [9, 10]. Characteristically, the cerebellum lesion is typically hypointense on T1-weighted sequence. On T2-weighted sequence, LDD appears as a "tiger-striped" cerebellar lesion with unilateral hemispheric expansion and preservation of the gyral pattern. This particular aspect is reported as a specific sign for the diagnosis [9]. The T2 hyperintensity can be explained by the presence of morphological alterations of the cells constituting the inner molecular layer and granular cells, associated with atrophy of the cerebellar white matter, thus confirming the dysplastic and malformative LDD [11].

Diffusion MR shows a hyperintense area at the level of the lesion, due to the "residual T2-contrast." The administration of paramagnetic contrast material shows no enhancement of the mass, indicating a nonsignificant alteration of the brain blood-barrier and the absence of extracellular edema.

Magnetic resonance spectroscopy reveals a typical spectrum of appearance of a lactate peak and reduction in the choline peak. A significant reduction in choline levels indicates a cellular turnover associated with demyelination and supports the dysplastic nature of the lesion [9, 10].

The contribution of MR perfusion imaging is also major as it shows an increase in relative cerebral blood volume, relative cerebral blood flow, and mean transit time in the lesion [9–11].

Although the detailed relationship is still unknown, LDD is considered to be associated with CS, an autosomal dominant hereditary syndrome characterized by systemic hamartoma lesions and high incidences of systemic malignancies [1, 6, 12]. It is reported that approximately 80% of patients with CS have germline mutation of phosphatase and tensin homolog, which is known as a tumor suppressor gene that maps to chromosome 10q23. As phosphatase and tensin homolog mutations are often found in adult-onset LDD, it is recommended that these patients be investigated for concomitant neoplasms to exclude CS [6, 12, 13].

Various additional abnormalities have been described in association with LDD. These include megalocephaly, syringomyelia, skeletal anomalies, multiple hemangioma, and mucocutaneous lesions [1, 6, 12–14].

To our knowledge, no instance of encephalocele associated with LDD has been reported thus far. Encephaloceles are rare congenital abnormalities; it is characterized by herniation of intracranial components through the cranial and facial bones due to a defect of closure of the anterior neuropore of the neural tube. Frontoethmoidal meningoencephaloceles are common [15]. Encephaloceles can be congenital or acquired secondary to tumor, hydrocephalus or other cause. Its etiology remains poorly understood. Some authors suggest that it could be due to ethnic, genetic, environmental factors, and paternal age [15, 16].

The association between LDD and encephalocele may be, in our opinion, part of a larger syndrome, and we suspect that many patients with asymptomatic encephalocele have gone undiagnosed in the setting of LDD. The theory of secondary development of encephalocele due to hydrocephalus by the mass effect generated by LDD is very plausible.

Gross total resection is frequently considered to be the foremost treatment of choice. However, due to the absence of a clear margin between normal cerebellum and the tumor intraoperatively, total surgical resection remains challenging, subtotal or partial resection common in the literature [1, 17].

The prognosis of Lhermitte–Duclos disease has improved over time with advances in neuroimaging, anesthesia, and neurosurgical technique [17, 18].

The treatment is surgical decompression of the posterior fossa to relieve the mass effect and hydrocephalus. The ill-defined margins of the lesion usually preclude complete excision.

Prior to the widespread availability of MRI, unfavorable outcomes were seen as a consequence of mass effect. The disease may recur, but malignant transformation has not been reported. Due to the indolent nature of this disease, radiation therapy is not indicated [1, 17, 19].

CONCLUSION

Lhermitte–Duclos disease is a rare tumor. It must be considered as one of the diagnosis when a young adult presents with a nonenhancing mass in the posterior fossa. It is associated with many developmental anomalies. To our knowledge, this is the first report of associating LDD with frontal encephalocele. Even if it is a benign condition, patients with LDD require a long-term follow-up and familial screening.

REFERENCES

1. Nowak DA, Trost HA. Lhermitte–Duclos disease (dysplastic cerebellar gangliocytoma): A malformation, hamartoma or neoplasm? Acta Neurol Scand 2002;105(3):137–45.
2. Shimanskiy VN, Karnaukhov VV, Shishkina LV, Vinogradov EV. The successful treatment of a patient with Lhermitte–Duclos disease (A case report and literature review). [Article in English]. Zh Vopr Neirokhir Im N N Burdenko 2015;79(4):78–83.
3. 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. AJNR Am J Neuroradiol 2001;22(5):824–30.
4. Koch R, Scholz M, Nelen MR, et al. Lhermitte–Duclos disease as a component of Cowden's syndrome. Case report and review of the literature. J Neurosurg 1999;90(4):776–9.
5. Lhermitte J, Duclos P. Sur un ganglioneurome diffus du cortex du cervelet. Bull Assoc Fr Etude Cancer 1920;9:99–107.
6. Robinson S, Cohen AR. Cowden disease and Lhermitte–Duclos disease: Characterization of a new phakomatosis. Neurosurgery 2000;46(2):371–83.
7. Lobo CJ, Mehan R, Murugasu E, Laitt RD. Tinnitus as the presenting symptom in a case of Lhermitte–Duclos disease. J Laryngol Otol 1999;113(5):464–5.
8. Pérez-Núñez A, Lagares A, Benítez J, et al. Lhermitte–Duclos disease and Cowden disease: Clinical and genetic study in five patients with Lhermitte–Duclos disease and literature review. Acta Neurochir (Wien) 2004;146(7):679–90.
9. Nagaraja S, Powell T, Griffiths PD, Wilkinson ID. MR imaging and spectroscopy in Lhermitte–Duclos disease. Neuroradiology 2004;46(5):355–8.
10. Moeninghoff C, Kraff O, Schlamann M, Ladd ME, Katsarava Z, Gizewski ER. Assessing a dysplastic cerebellar gangliocytoma (Lhermitte–Duclos disease) with 7T MR imaging. Korean J Radiol 2010;11(2):244–8.
11. Moonis G, Ibrahim M, Melhem ER. Diffusion-weighted MRI in Lhermitte–Duclos disease: Report of two cases. Neuroradiology 2004;46(5):351–4.
12. Derrey S, Proust F, Debono B, et al. Association between Cowden syndrome and Lhermitte–Duclos disease: Report of two cases and review of the literature. Surg Neurol 2004;61(5):447–54.
13. Vantomme N, Van Calenbergh F, Goffin J, Sciot R, Demaerel P, Plets C. Lhermitte–Duclos disease is a clinical manifestation of Cowden's syndrome. Surg Neurol 2001;56(3):201–4.
14. Marcus CD, Galeon M, Peruzzi P, Bazin A, Bernard MH, Pluot M, Menanteau B. Lhermitte–Duclos disease associated with syringomyelia. Neuroradiology 1996;38(6):529–31.
15. Mahapatra AK, Agrawal D. Anterior encephaloceles: A series of 103 cases over 32 years. J Clin Neurosci 2006;13(5):536–9.
16. Dubey D, Pande S, Dubey P, Sawhney A. A case of naso-ethmoidal meningoencephalocele. Int J Collab Res Intern Med Public Health 2011;3(8):666–9.
17. Gama I, Almeida L. Lhermitte–Duclos disease associated to Cowden syndrome: De novo diagnosis and management of these extremely rare syndromes in a patient. BMJ Case Rep. 2017;2017. pii: bcr2016217974.
18. Jiang T, Wang J, Du J, et al. Lhermitte–Duclos disease (dysplastic gangliocytoma of the cerebellum) and Cowden syndrome: Clinical experience from a single institution with long-term follow-up. World Neurosurg 2017;104:398–406.
19. Zak M, Ledbetter M, Maertens P. Infantile Lhermitte–Duclos disease treated successfully with rapamycin. J Child Neurol 2017;32(3):322–6.

****

Author Contributions

Habib Bellamlih – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Aymane El Farouki – Conception of the work, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final

approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Moulay Rachid El Hassani – Conception of the work, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Mohammed Jiddane – Conception of the work, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Meriem Fikri – Conception of the work, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission
The corresponding author is the guarantor of submission.

Source of Support
None.

Consent Statement
Written informed consent was obtained from the patient for publication of this article.

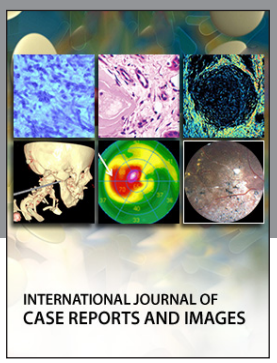
Conflict of Interest
Authors declare no conflict of interest.

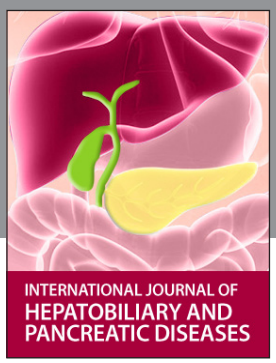
Data Availability
All relevant data are within the paper and its Supporting Information files.

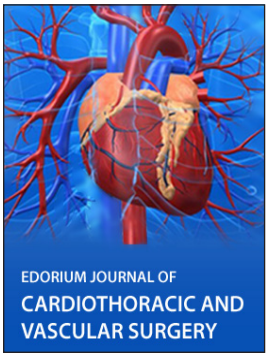
Copyright
© 2019 Habib Bellamlih et al. This article is distributed under the terms of Creative Commons Attribution License which permits unrestricted use, distribution and reproduction in any medium provided the original author(s) and original publisher are properly credited. Please see the copyright policy on the journal website for more information.

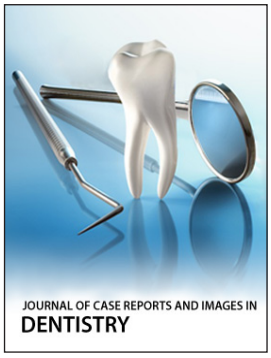
Access full text article on other devices

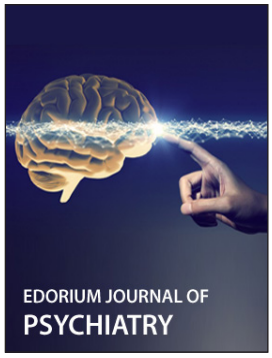
Access PDF of article on other devices

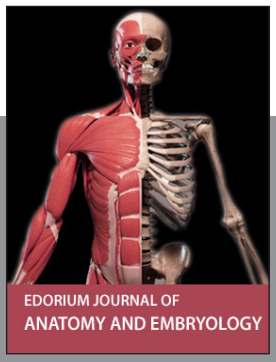


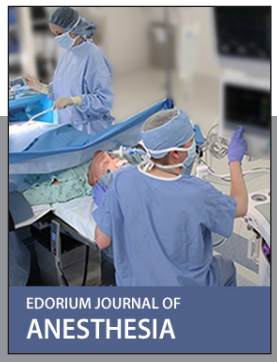












Submit your manuscripts at

www.edoriumjournals.com