Turkish Journal of Neurology

Turk J Neurol 2024;30(4):276-278
Doi: 10.4274/tnd.2023.09634

LETTER TO THE EDITOR

Lhermitte-Duclos disease presenting as cognitive impairment: A case report with a seven-year follow-up

Bekir Burak Kılboz, **Raziye Tıraş**

Department of Neurology, University of Health Sciences Türkiye, Prof. Dr. Cemil Tascioglu City Hospital, Istanbul, Türkiye

Lhermitte-Duclos disease (LDD), also known as dysplastic cerebellar gangliocytoma, is a condition marked by the presence of a cerebellar hamartomatous lesion. This lesion is typically identified on magnetic resonance imaging (MRI) by its distinctive "tiger-striped" appearance.[1] Typically affecting young adults, LDD can present with headache, cerebellar dysfunction, cranial nerve palsies, visual symptoms, and even hydrocephalus; however, cognitive impairment as an initial presentation is rare.[2] Herein, we report a case of a 33-year-old male with cognitive impairment who was diagnosed with LDD and followed up for seven years.

In January 2015, the patient presented to our neurology outpatient clinic, reporting a six-month history of forgetfulness concerning names, numbers, and daily activities-a problem the patient noticed, but his close friends did not. The patient had worked as an accountant after university, where he was a good student. Despite these memory issues, the patient conveyed a positive mood and recent happiness. The patient denied having any issues with balance, ambulation, headaches, dizziness, vertigo, or double vision and reported no family or personal history of benign or malignant tumors. A written informed consent was obtained from the patient.

Neurological examination, including mental status examination, was normal without papilloedema. Blood tests, including thyroid hormones and vitamin B12, were within normal limits. Brain MRI showed left cerebellar T1 hypointense and T2 hyperintense linear tiger-striped striations without intravenous gadolinium contrast enhancement, consistent with LDD (Figure 1). The patient was started on escitalopram 10 mg/day, and after two months, the patient reported more than 50% improvement in symptoms. However, the patient voluntarily discontinued escitalopram in the third month. The patient was followed up with brain MRIs semiannually for the first two years and biennially for the next six years. The lesion remained stable, and the patient's attention and recall were minimally impaired. No other hamartomas or malignancies were detected.

Lhermitte-Duclos disease is a rare disease caused by a mutation in the PTEN (phosphatase and tensin homolog) gene on chromosome 10q23. This same mutation can also lead to Cowden syndrome, an autosomal dominant disorder characterized by hamartomatous lesions and an increased risk for cancer. Patients with LDD should receive regular assessments and screenings for these associated conditions.[3] Although histopathological findings include loss of white matter and Purkinje cells, outer molecular layer hypermyelination, mitochondria-rich inner granular layer, and dysplastic ganglion cells with rounded nuclei, histopathological confirmation is not usually required, and the diagnosis is based on imaging features.[1]

The mechanism of cognitive impairment in LDD may be due to the cerebellar cognitive affective syndrome, described as executive dysfunction, inattention, visuospatial disorganization, and issues with language production.[4] The cerebellar cognitive affective syndrome is thought to stem from damage

Correspondence: Bekir Burak Kılboz, MD. SBÜ Dr. Cemil Taşçıoğlu Şehir Hastanesi, Nöroloji Kliniği, 34384 Şişli, İstanbul, Türkiye.
E-mail: bbkilboz@gmail.com
Received: April 19, 2023 **Accepted:** August 25, 2023 **Published online:** December 20, 2024
Cite this article as: Kılboz BB, Tıraş R. Lhermitte-Duclos disease presenting as cognitive impairment: A case report with a seven-year follow-up. Turk J Neurol 2024;30(4):276-278. doi: 10.4274/tnd.2023.09634.

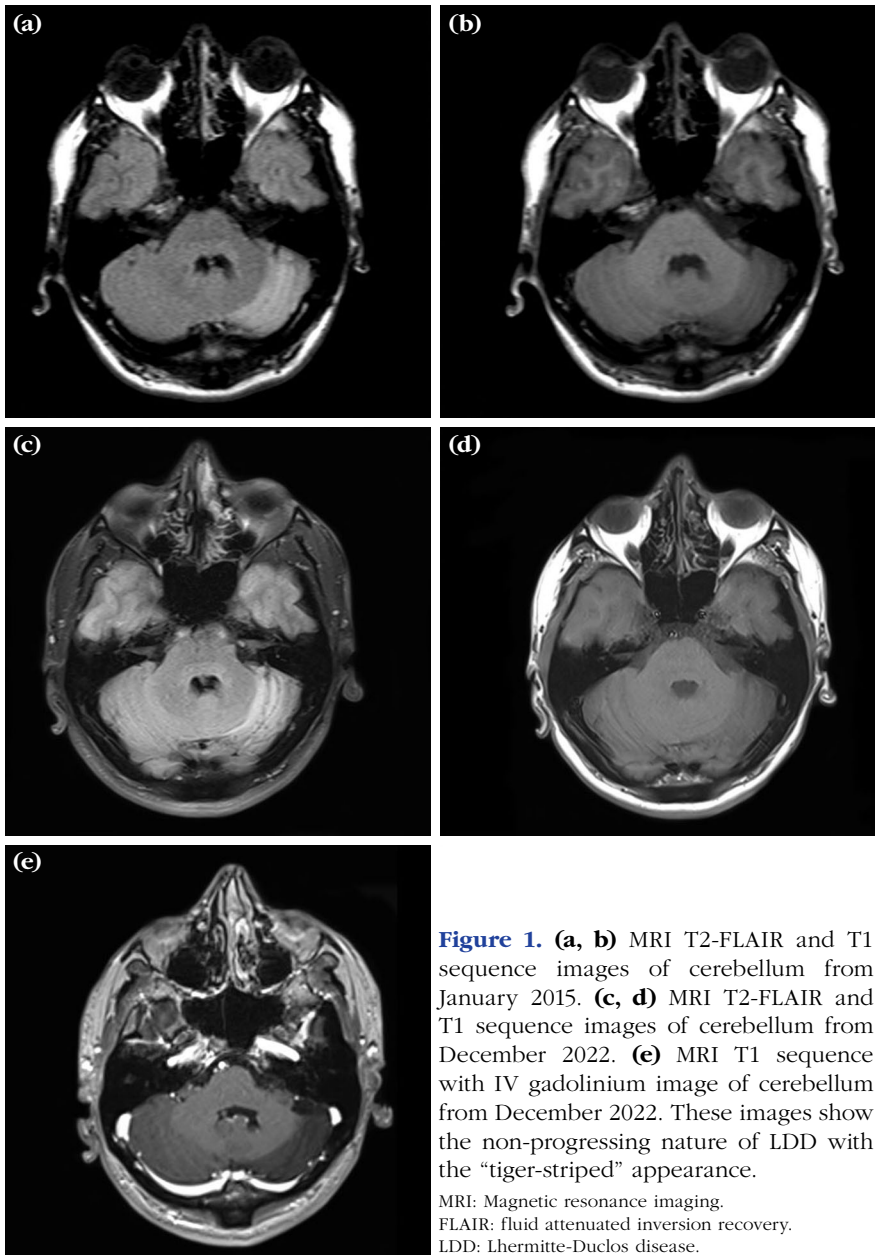

Figure 1. **(a, b)** MRI T2-FLAIR and T1 sequence images of cerebellum from January 2015. **(c, d)** MRI T2-FLAIR and T1 sequence images of cerebellum from December 2022. **(e)** MRI T1 sequence with IV gadolinium image of cerebellum from December 2022. These images show the non-progressing nature of LDD with the "tiger-striped" appearance.

MRI: Magnetic resonance imaging.
FLAIR: fluid attenuated inversion recovery.
LDD: Lhermitte-Duclos disease.

to the posterior cerebellar lobe and theorized to result in a dysfunction in the thought process known as "dysmetria of thought."[4] The PTEN gene is also associated with learning disabilities and autism spectrum disorder.[5] In our case, the patient's cognitive impairment responded to escitalopram treatment, which is a selective serotonin reuptake inhibitor.

In conclusion, we presented a rare case of LDD with cognitive impairment that responded well to escitalopram treatment and exhibited a stable follow-up. Although LDD is a benign condition and progression of the lesion is not expected, systemic evaluation and follow-up are necessary due to the associated conditions.

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

Author Contributions: Design, literature review, writing, references: B.B.K.; Idea, control/supervision, data collection, review, analysis: R.T.

Conflict of Interest: The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

Funding: The authors received no financial support for the research and/or authorship of this article.

REFERENCES

1. Meltzer CC, Smirniotopoulos JG, Jones RV. The striated cerebellum: An MR imaging sign in Lhermitte-Duclos disease (dysplastic gangliocytoma). Radiology 1995;194:699-703. doi: 10.1148/radiology.194.3.7862965.
2. Milbouw G, Born JD, Martin D, Collignon J, Hans P, Reznik M, et al. Clinical and radiological aspects of dysplastic gangliocytoma (Lhermitte-Duclos disease): A report of two cases with review of the literature. Neurosurgery 1988;22:124-8. doi: 10.1227/00006123-198801010-00020.
3. Zhou XP, Marsh DJ, Morrison CD, Chaudhury AR, Maxwell M, Reifenberger G, et al. Germline inactivation of PTEN and dysregulation of the phosphoinositol-3-kinase/Akt pathway cause human Lhermitte-Duclos disease in adults. Am J Hum Genet 2003;73:1191-8. doi: 10.1086/379382.
4. Schmahmann JD. The cerebellum and cognition. Neurosci Lett 2019;688:62-75. doi: 10.1016/j.neulet.2018.07.005.
5. Cummings K, Watkins A, Jones C, Dias R, Welham A. Behavioural and psychological features of PTEN mutations: A systematic review of the literature and meta-analysis of the prevalence of autism spectrum disorder characteristics. J Neurodev Disord 2022;14:1. doi: 10.1186/s11689-021-09406-w.