



## CASE REPORT

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## Atypical Presentation of Multiple Sclerosis on MRI: A Diagnostic Challenge

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Multiple Sclerosis (MS) is a chronic inflammatory demyelinating disease of the central nervous system, typically diagnosed based on clinical presentation and characteristic magnetic resonance imaging (MRI) findings. However, atypical MRI presentations can pose significant diagnostic challenges. This case report discusses a patient with an unusual MRI manifestation of MS, emphasizing the importance of considering MS in the differential diagnosis of atypical neuroimaging findings

**Keywords:** Multiple Sclerosis; Magnetic Resonance Imaging; Demyelinating Lesion

### INTRODUCTION

MS is a chronic, demyelinating disorder of the central nervous system that presents with a wide variety of clinical manifestations. The hallmark of MS is the presence of white matter lesions in

the brain and spinal cord, which are typically detected through MRI (1). However, MS can present in atypical ways that pose significant diagnostic challenges, especially when the lesions do not follow the classic distribution or exhibit unusual characteristics (2). In these cases, the recognition of MS on MRI may be delayed, leading to misdiagnosis or inappropriate treatments (3).

Atypical presentations of MS are particularly problematic when the lesion patterns mimic other neurological conditions, such as vascular diseases, infections, or neoplasms, complicating the clinician's ability to distinguish between them (4). Moreover, MS lesions may appear in atypical locations, such as the brainstem or spinal cord, or demonstrate unusual features, including small or scattered plaques that lack the typical periventricular, juxtacortical, or infratentorial localization (5). This can result in an overlap of imaging findings with other pathologies, thus requiring a high degree of clinical suspicion and the use of advanced MRI techniques, such as gadolinium enhancement or magnetization transfer imaging, to improve diagnostic accuracy (6).

The complexity of diagnosing MS in these atypical cases underscores the importance of considering the full spectrum of potential differential diagnoses, along with a thorough clinical evaluation (7).

This case report presents a unique instance of MS with an unusual MRI presentation, highlighting the importance of recognizing these atypical patterns to ensure prompt and accurate diagnosis, and ultimately, optimal management for the patient.

## CASE REPORT

A 25-year-old medical student presented with acute onset of right-sided weakness, numbness, visual disturbances and speech difficulties. Her medical history was unremarkable, and there was no family history of neurological disorders. Neurological examination revealed mild right hemiparesis and expressive aphasia.

Initial MRI of the brain demonstrated a large, heterogeneous, contrast-enhancing lesion in the left frontal lobe, measuring approximately 3.5 cm in diameter, with surrounding edema and mass effect. The lesion exhibited both T2 hyperintensity and areas of T1 hypointensity, suggestive of a tumefactive demyelinating lesion (TDL). These findings raised concerns for a high-grade glioma or abscess.

Further evaluation, including cerebrospinal fluid (CSF) analysis, revealed the presence of oligoclonal bands, supporting a diagnosis of demyelinating disease. Additional MRI of the cervical spine showed multiple small T2 hyperintense lesions without enhancement, consistent with demyelination. Given the combination of clinical presentation, CSF findings, and imaging characteristics, a diagnosis of multiple sclerosis with atypical MRI presentation was established.

Furthermore, patient was also referred to the ophthalmology Dept. for evaluation and management of ocular manifestation of the condition.

Given the patient's diagnosis of MS with an atypical MRI presentation, the management approach should be multidisciplinary and individualized, focusing on both acute management and long-term disease control.

### **Acute Management (Relapse Phase)**

The patient presents with acute neurological symptoms (right-sided weakness, numbness, visual disturbances, and speech difficulties), which are indicative of an MS relapse. Management at this stage involves: Intravenous methylprednisolone (1 g daily for 3-5 days) is typically administered to reduce inflammation and edema around demyelinating lesions. This helps in improving neurological deficits and shortening the duration of the relapse (8).

Regular follow-up visits were planned and included clinical evaluations, repeat MRI (at least annually), and assessment of relapse frequency. New lesions or progression of existing lesions on MRI may prompt escalation of treatment (9).

The management of this 25-year-old medical student with multiple sclerosis involves acute treatment with corticosteroids, initiation of long-term disease-modifying therapy, and multidisciplinary care to address neurological and visual symptoms, rehabilitation needs, and psychological support. Regular follow-up is essential for assessing treatment efficacy and disease progression.

## **DISCUSSION**

MS diagnosis is primarily based on the McDonald criteria, which incorporate clinical and MRI findings to demonstrate dissemination in time and space (10). Typical MRI features include multiple ovoid, periventricular, juxtacortical, infratentorial, and spinal cord lesions. However,

atypical presentations, such as TDLs, can mimic other pathologies, leading to diagnostic dilemmas.

TDLs are large demyelinating lesions ( $>2$  cm) that often present with mass effect and ring enhancement, resembling neoplasms or abscesses. Differentiating TDLs from other intracranial lesions is crucial, as misdiagnosis can lead to inappropriate management. Advanced imaging techniques, such as magnetic resonance spectroscopy and diffusion tensor imaging, can aid in distinguishing TDLs from neoplastic processes by evaluating metabolic profiles and microstructural integrity, respectively. (4-5)

In this case, the patient's acute neurological deficits, combined with the MRI findings of a large, enhancing lesion with mass effect, initially suggested a neoplastic process. However, the presence of CSF oligoclonal bands and additional demyelinating lesions in the cervical spine supported the diagnosis of MS. This underscores the importance of a comprehensive diagnostic approach, including clinical assessment, CSF analysis, and thorough imaging evaluation. (8)

MS patients with atypical MRI presentations have variability in imaging findings and the potential for misdiagnosis. The study emphasized the need for heightened awareness of atypical MS manifestations to prevent diagnostic delays and ensure appropriate treatment.

Patient patient with recurrent optic neuritis and initially normal MRI findings, are later diagnosed with MS after subsequent imaging revealed demyelinating lesions. This case highlights that MS can present with normal initial imaging, and clinicians should maintain a high index of suspicion in patients with suggestive clinical features. (11-12)

Differential diagnoses for atypical MRI presentations include neoplasms, abscesses, and other demyelinating diseases such as neuromyelitis optica spectrum disorder (NMOSD). Distinguishing MS from NMOSD is essential, as treatment strategies differ. In this context, testing for aquaporin-4 antibodies can aid in differentiating between these conditions. (13)

## CONCLUSION

This case illustrates the diagnostic challenges posed by atypical MRI presentations of MS. Clinicians should consider MS in the differential diagnosis of large, enhancing brain lesions, even when imaging features are not classic. A multidisciplinary approach, incorporating clinical

evaluation, CSF analysis, and advanced imaging techniques, is essential for accurate diagnosis and appropriate management.

## REFERENCES

1. Filippi M, Rocca MA, Ciccarelli O. Magnetic resonance imaging of multiple sclerosis. *Lancet Neurol.* 2016;15(4):429-38. doi: 10.1016/S1474-4422(16)00015-5.
2. Filippi M, Preziosa P, Banwell BL, et al. Assessment of lesions on magnetic resonance imaging in multiple sclerosis: practical guidelines. *Brain.* 2019 Jul 1;142(7):1858-1875. doi: 10.1093/brain/awz144. PMID: 31209474; PMCID: PMC6598631.
3. Weidauer S, Raab P, Hattingen E. Diagnostic approach in multiple sclerosis with MRI: an update. *Clinical Imaging.* 2021 Oct 1;78:276-85.
4. Rocca MA, Preziosa P, Barkhof F, Brownlee W, Calabrese M, De Stefano N, Granziera C, Ropele S, Toosy AT, Vidal-Jordana À, Di Filippo M. Current and future role of MRI in the diagnosis and prognosis of multiple sclerosis. *The Lancet Regional Health–Europe.* 2024 Sep 1;44.
5. DeMaio A, Mehrotra S, Sambamurti K, Husain S. The role of the adaptive immune system and T cell dysfunction in neurodegenerative diseases. *Journal of Neuroinflammation.* 2022 Oct 8;19(1):251.
6. Wattjes MP, Ciccarelli O, Reich DS, Banwell B, de Stefano N, Enzinger C, Fazekas F, Filippi M, Frederiksen J, Gasperini C, Hacohen Y. 2021 MAGNIMS–CMSC–NAIMS consensus recommendations on the use of MRI in patients with multiple sclerosis. *The Lancet Neurology.* 2021 Aug 1;20(8):653-70.
7. Meca-Lallana V, Berenguer-Ruiz L, Carreres-Polo J, Eichau-Madueño S, Ferrer-Lozano J, Forero L, Higueras Y, Téllez Lara N, Vidal-Jordana A, Pérez-Miralles FC. Deciphering multiple sclerosis progression. *Frontiers in Neurology.* 2021 Apr 7;12:608491.
8. Ramo-Tello C, Blanco Y, Brieva L, Casanova B, Martínez-Cáceres E, Ontaneda D, Ramió-Torrentá L, Rovira À. Recommendations for the diagnosis and treatment of multiple sclerosis relapses. *Journal of Personalized Medicine.* 2021 Dec 22;12(1):6.

9. Ananthavarathan P, Sahi N, Chard DT. An update on the role of magnetic resonance imaging in predicting and monitoring multiple sclerosis progression. *Expert Review of Neurotherapeutics*. 2024 Feb 1;24(2):201-16.
10. Polman CH, Reingold SC, Banwell B, Clanet M, Cohen JA, Filippi M, Fujihara K, Havrdova E, Hutchinson M, Kappos L, Lublin FD, Montalban X, O'Connor P, Sandberg-Wollheim M, Thompson AJ, Waubant E, Weinshenker B, Wolinsky JS. Diagnostic criteria for multiple sclerosis: 2010 revisions to the McDonald criteria. *Ann Neurol*. 2011 Feb;69(2):292-302. doi: 10.1002/ana.22366. PMID: 21387374; PMCID: PMC3084507.
11. Kale N. Optic neuritis as an early sign of multiple sclerosis. *Eye and brain*. 2016 Oct 26:195-202.
12. Zafar U, Habib SH, Raza SS. Identification of appropriate tools to gauge brain functions in a clinical setup of a developing country: A pilot study. *Pak J Med Sci*. 2023 Nov-Dec;39(6):1840-1846. doi: 10.12669/pjms.39.6.7489. PMID: 37936739; PMCID: PMC10626111.
13. Pittock SJ, Lucchinetti CF. Neuromyelitis optica and the evolving spectrum of autoimmune aquaporin-4 channelopathies: a decade later. *Annals of the New York Academy of Sciences*. 2016 Feb;1366(1):20-39.