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Acute CNS Demyelinating Disorder Presenting with Optic Neuritis, Myelitis, and Seizures in a Young Female: A Diagnostic Challenge

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Abstract: ***Acute demyelinating disorders of the central nervous system (CNS) can present with overlapping clinical and radiological features, making diagnosis challenging. We report a case of a 19-year-old female presenting with acute unilateral visual loss, seizures, and upper motor neuron signs. MRI revealed right retrobulbar optic neuritis and a cervical spinal cord lesion. Extensive infectious and autoimmune workup, including neuromyelitis optica spectrum disorder (NMOSD) and myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD), was negative. The patient responded dramatically to corticosteroids and antiepileptics, with radiological and clinical improvement. This case highlights the importance of considering seronegative demyelinating disorders and early steroid therapy in atypical presentations.***

Keywords: central nervous system demyelination, optic neuritis, spinal cord lesion, seronegative disorder, corticosteroid therapy

1. Introduction

Demyelinating disorders of the CNS such as multiple sclerosis (MS), NMOSD, MOGAD, and acute disseminated encephalomyelitis (ADEM) often present with optic neuritis and myelitis. However, seizures and encephalopathy may obscure diagnosis, especially in younger patients. Distinguishing between these entities is essential due to differences in treatment and prognosis. We present a diagnostically challenging case of acute CNS demyelination with negative autoimmune markers.

2. Case Presentation

A 19-year-old female presented with sudden onset defective vision in the right eye followed by new-onset involuntary movements suggestive of seizures. On admission, the patient was in a postictal state and afebrile.

Following recovery, neurological examination revealed:
Dilated right pupil with impaired light reflex
Reduced accommodation reflex in the right eye
Decreased visual acuity in the right eye
Normal tone in all four limbs
Exaggerated deep tendon reflexes
Presence of ankle and patellar clonus
Bilateral extensor plantar responses
Fundus examination was normal.

Investigations
Routine blood investigations: Within normal limits
MRI Brain: Right retrobulbar optic neuritis
MRI Spine: Hyperintense lesion in cervical spinal cord (~4 cm)

CSF Analysis:
Normal opening pressure
Normal glucose and protein
No oligoclonal band
Viral panel negative

Autoimmune Workup:
Autoimmune encephalitis panel: Negative
NMOSD antibodies: Negative
MOG antibodies: Negative
Differential Diagnosis
Multiple sclerosis (MS)
MOG antibody-associated disease (MOGAD)
Neuromyelitis optica spectrum disorder (NMOSD)
Acute disseminated encephalomyelitis (ADEM)
Infectious encephalomyelitis

Treatment
The patient was treated with:
Intravenous high-dose corticosteroids
Antiepileptic drugs
Outcome and Follow-Up

The patient showed significant clinical improvement:
Visual acuity improved
Reflexes normalized
No further seizures
Repeat MRI brain showed resolving optic neuritis.

3. Discussion

This case highlights a probable acute inflammatory demyelinating disorder with overlapping features. The combination of optic neuritis and longitudinal spinal cord lesion raises suspicion for NMOSD or MOGAD; however, seronegativity and favorable steroid response suggest a monophasic inflammatory process.

Seizures are atypical in classic demyelinating disorders but may be seen in:
- ADEM
- Cortical demyelination
- Encephalitic involvement

Normal CSF findings and negative autoimmune markers made diagnosis challenging. Early steroid therapy resulted in rapid recovery, emphasizing its importance in suspected inflammatory demyelination.

4. Conclusions

Seronegative acute CNS demyelinating disorders should be considered in young patients presenting with optic neuritis, myelitis, and seizures. Early diagnosis and prompt corticosteroid therapy can significantly improve outcomes. Long-term follow-up is necessary to detect recurrence or evolution into a chronic demyelinating condition. "Seronegative acute inflammatory CNS demyelinating disorder presenting with unilateral retrobulbar optic neuritis, cervical myelitis, and seizures."

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