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Neuromyelitis Optica Spectrum Disorder with Longitudinally Extensive Transverse Myelitis and Intractable Vomiting: A Case Report

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Abstract: ***Background: Neuromyelitis optica spectrum disorder (NMOSD) is a rare autoimmune demyelinating disorder of the central nervous system characterized by recurrent episodes of optic neuritis, longitudinally extensive transverse myelitis (LETM), and area postrema involvement. Early diagnosis and prompt initiation of immunotherapy are essential to prevent irreversible neurological disability (1, 2). Case Presentation: We report the case of a 25-year-old female who presented with tingling and numbness over the neck and both upper limbs, followed by progressive quadriparesis. Initial evaluation elsewhere suggested vitamin B_{12} deficiency, but symptoms persisted. On admission, she developed intractable nausea and vomiting, and neurological examination revealed exaggerated deep tendon reflexes with bilateral extensor plantar responses. MRI of the cervical spine revealed long-segment T2/STIR hyperintensity extending from C2 to C7, consistent with longitudinally extensive transverse myelitis. Cerebrospinal fluid analysis showed mild lymphocytic pleocytosis with elevated protein, and serum testing was positive for AQP4-IgG antibodies. She was treated with high-dose intravenous methylprednisolone, followed by therapeutic plasma exchange due to clinical deterioration. Significant neurological recovery was achieved after five cycles of plasma exchange (3, 7). She was discharged on oral corticosteroids and mycophenolate mofetil for maintenance immunosuppression and remained relapse-free on follow-up. Conclusion: This case highlights the importance of considering NMOSD in patients presenting with LETM and unexplained vomiting. Early recognition and prompt initiation of immunotherapy can result in excellent neurological recovery and prevention of relapses (4, 10)***

Keywords: Neuromyelitis optica spectrum disorder, AQP4 antibody, longitudinally extensive transverse myelitis, plasma exchange, area postrema syndrome.

1. Introduction

Neuromyelitis optica spectrum disorder (NMOSD) is a severe, relapsing, autoimmune astrocytopathy that primarily affects the optic nerves and spinal cord (1, 2). Historically regarded as a variant of multiple sclerosis (MS), NMOSD is now recognized as a distinct clinical entity following the discovery of pathogenic autoantibodies directed against the aquaporin-4 (AQP4) water channel on astrocytic end-feet. The detection of serum AQP4-IgG has become the cornerstone of diagnosis, offering high specificity for NMOSD and distinguishing it from MS and other inflammatory demyelinating diseases.

The clinical spectrum of NMOSD extends beyond the classical triad of optic neuritis and longitudinally extensive transverse myelitis (LETM) to include brainstem and diencephalic syndromes, such as area postrema syndrome, characterized by intractable nausea, vomiting, or hiccups.

Magnetic resonance imaging (MRI) findings of NMOSD typically reveal LETM, defined as a spinal cord lesion extending over three or more vertebral segments, often central in location and involving the gray matter (1, 6) Early diagnosis and initiation of high-dose corticosteroids, followed by plasma exchange or immunosuppressive therapy, are crucial for minimizing irreversible neurological damage and preventing relapses (3, 7, 10)

We present a case of AQP4-IgG–positive NMOSD in a young female who presented with cervical LETM and area postrema symptoms, illustrating the importance of early recognition and aggressive immunotherapy in achieving favorable outcomes.

2. Case Presentation

A 25-year-old married female, with no known comorbidities, presented to our outpatient department with complaints of tingling and numbness over the neck and both upper limbs for the past 10 days. The symptoms were insidious in onset and gradually progressive. She denied any weakness, visual disturbance, headache, limb pain, or bladder/bowel involvement at the time of presentation.

Two weeks prior, she had been evaluated at a local hospital, where she was provisionally diagnosed with vitamin B_{12} deficiency and treated with intravenous vitamin B_{12} supplementation for five days without any improvement in symptoms.

There was no history of fever, recent infection, vaccination, or drug intake, and no similar neurological episodes in the past. Family history for autoimmune or demyelinating diseases was negative.

Following admission, the patient developed persistent nausea and vomiting, which did not respond to standard antiemetic therapy. After three to four days, she noticed progressive weakness in her arms and legs.

3. Examination

On admission, the patient was hemodynamically stable.

Central Nervous System Examination:
The patient was alert, conscious, and oriented to time, place, and person. Cranial nerves were intact; vision, pupillary reflexes, and extraocular movements were normal. Fundoscopic examination revealed normal optic discs with no signs of optic neuritis.

Motor system evaluation initially demonstrated normal muscle bulk and tone, with power 5/5 in all four limbs. However, deep tendon reflexes were brisk and exaggerated in both upper and lower limbs, and bilateral plantar responses were extensor.

Over the next three days, the patient developed progressive weakness of all four limbs. By the fourth day, muscle power had declined to Right upper limb – 2/5, Left upper limb – 3/5, and both lower limbs – 3/5 (Medical Research Council grading). Muscle tone remained increased, and coordination could not be assessed due to weakness.

Sensory examination revealed diminished touch and pinprick sensation over C3–C7 dermatomes bilaterally, while vibration and joint position senses were preserved.

No meningeal signs or cerebellar involvement were noted. Other systemic examinations were unremarkable.

Investigations and Hospital Course
Routine hematological and biochemical investigations, including complete blood count, liver and renal function tests, serum electrolytes, calcium, and magnesium, were within normal limits. Fasting blood glucose, thyroid profile, and serum vitamin B_{12} levels were normal. Serological tests for HIV and HBsAg were negative.

Magnetic Resonance Imaging (MRI) of the brain and cervical spine with gadolinium contrast revealed a diffuse long-segment T2/STIR hyperintensity extending from C2 to C7, involving more than two-thirds of the spinal cord circumference, associated with mild cord expansion and subtle patchy enhancement. The brain parenchyma appeared normal, with no demyelinating lesions, thereby effectively ruling out multiple sclerosis.

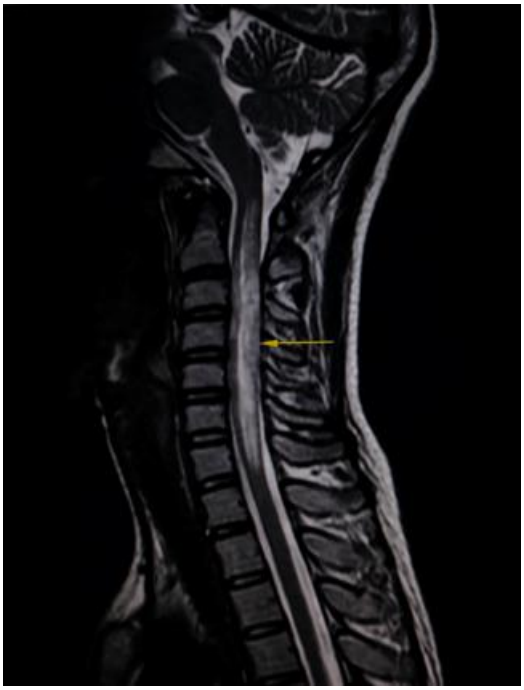

Figure 1: Sagittal T2-weighted MRI of the cervical spine shows a long-segment hyperintense lesion extending from C2 to C7, involving more than two-thirds of the spinal cord circumference, with mild cord expansion.

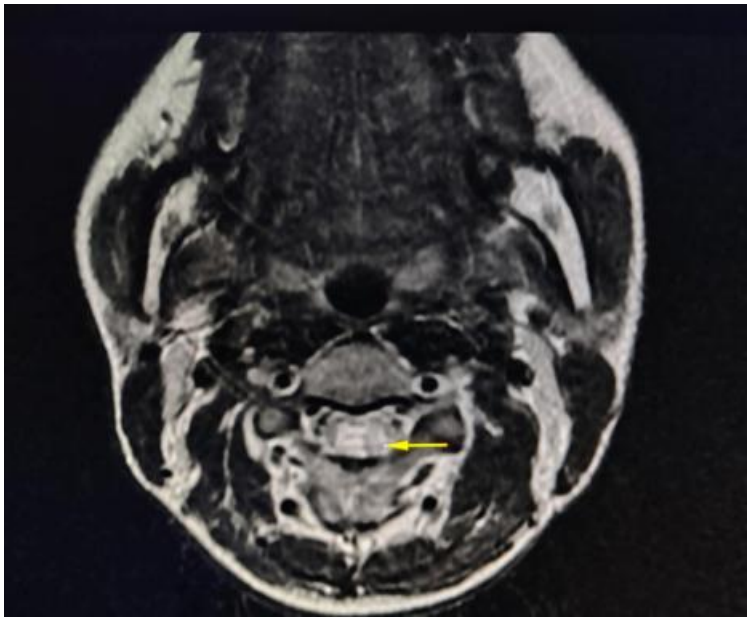

Figure 2: Axial T2-weighted MRI shows diffuse hyperintensity within the cervical cord, corresponding to the same segment

Nerve conduction studies demonstrated normal sensory and motor conduction parameters, excluding peripheral neuropathic involvement.

Cerebrospinal fluid (CSF) examination showed the following: Protein: 70 mg/dL; Glucose: 84 mg/dL; Total cells: 32

cells/mm³ (90% lymphocytes); Oligoclonal bands: Negative; ADA: 1 U/L; GeneXpert MTB: Not detected; ACE: 58.88 U/L.

Autoimmune and infectious panels were negative for ANA blot, C-ANCA, P-ANCA, and MOG antibodies. However, AQP4/NMO-IgG antibody was moderately positive (1.32 titre) on ELISA (Reference Range 1.10), confirming the diagnosis of AQP4-positive Neuromyelitis Optica Spectrum Disorder (NMOSD) presenting as cervical longitudinally extensive transverse myelitis (LETM).

Treatment and Hospital Course

Following the diagnosis of longitudinally extensive transverse myelitis secondary to suspected NMOSD, the patient was started on intravenous methylprednisolone (1 g/day) (7, 8) Despite therapy, there was no improvement in her symptoms, and over the next three days, muscle power began to deteriorate from normal strength at admission to Right upper limb – 2/5, Left upper limb – 3/5, and both lower limbs – 3/5.

After the third dose of methylprednisolone, due to the progressive neurological decline, a decision was made to initiate therapeutic plasma exchange (PLEX) (3, 10) The procedure was performed on alternate days using a plasma filter, exchanging approximately 1.5 plasma volumes per session, with 5% human albumin and fresh frozen plasma as replacement fluids.

After the first cycle of PLEX, mild improvement in upper limb strength was noted. However, following the third session, the patient developed sudden deterioration in sensorium (GCS 8/15) with labile blood pressure and tachycardia, requiring ICU transfer and ventilatory support. No metabolic or infectious cause was identified. After stabilization, PLEX was resumed and completed for a total of five cycles.

By the end of treatment, the patient demonstrated significant neurological recovery, with gradual improvement in limb power and resolution of sensory symptoms. By the second week, motor power improved to 5/5 in all four limbs, and plantar responses became flexor bilaterally.

She was transitioned to oral prednisolone (1 mg/kg/day) with a gradual taper and started on oral mycophenolate mofetil (1 g/day in two divided doses) as maintenance immunosuppression to prevent relapse (5, 7) The patient tolerated therapy well and was discharged after three weeks of hospitalization with complete neurological recovery. She is on regular follow up in the OPD since 4 months.

4. Discussion

Neuromyelitis optica spectrum disorder (NMOSD) is an autoimmune inflammatory demyelinating disorder of the central nervous system (CNS) that predominantly affects the optic nerves and spinal cord. Once considered a variant of multiple sclerosis (MS), the identification of the aquaporin-4 (AQP4) antibody has established NMOSD as a distinct clinical entity. The presence of AQP4-IgG, a pathogenic autoantibody directed against the water channel protein expressed on astrocyte foot processes, is highly specific for the disease and plays a crucial role in its diagnosis and pathogenesis (1, 4)

The classical manifestations of NMOSD include optic neuritis, acute transverse myelitis, and area postrema syndrome, characterized by intractable nausea, vomiting, and hiccups due to involvement of the medullary chemoreceptor trigger zone (1, 2). Other core features include acute brainstem syndromes, symptomatic narcolepsy, and diencephalic or cerebral presentations. The present case demonstrated longitudinally extensive transverse myelitis (LETM) extending from C2 to C7, accompanied by persistent nausea and vomiting, likely representing subclinical area postrema involvement- an underrecognized but characteristic feature of NMOSD.

LETM, defined as a spinal cord lesion extending over three or more vertebral segments, is a hallmark imaging feature that differentiates NMOSD from MS, where cord lesions are typically shorter and peripheral. In this case, MRI revealed a long segment T2/STIR hyperintensity spanning C2–C7 with mild cord expansion and patchy enhancement, consistent with an inflammatory demyelinating process. The absence of brain lesions, negative oligoclonal bands in CSF, and seropositivity for AQP4-IgG strongly supported the diagnosis of NMOSD.

CSF analysis in NMOSD often demonstrates mild lymphocytic pleocytosis and elevated protein levels, while oligoclonal bands are usually absent- findings that were consistent with our patient's CSF profile. The differential diagnosis for LETM includes multiple sclerosis, sarcoidosis, systemic lupus erythematosus, Sjögren's syndrome, paraneoplastic myelitis, infectious causes (such as tuberculosis or viral myelitis), and MOG antibody-associated disease (MOGAD). These were systematically excluded through negative autoimmune and infectious panels, normal ACE levels, and negative MOG-IgG (2, 6, 9)

The absence of response to high-dose intravenous methylprednisolone (IVMP) in this case highlights the variability in therapeutic responsiveness among NMOSD patients. Plasma exchange (PLEX) has proven to be an effective rescue therapy in steroid-refractory cases, as it directly eliminates pathogenic autoantibodies and complement components contributing to astrocytic injury. In this patient, the initiation of PLEX after the third steroid dose led to substantial neurological improvement, underscoring the benefit of early therapeutic escalation.

Long-term relapse prevention remains a critical aspect of NMOSD management. The addition of immunosuppressive therapy with mycophenolate mofetil in this patient provided sustained remission and prevented recurrence during follow-up. Early institution of maintenance therapy after acute recovery has been associated with reduced relapse rates and better functional outcomes.

This case emphasizes that NMOSD can initially mimic other metabolic or inflammatory myelopathies, such as vitamin B_{12} deficiency, leading to misdiagnosis or delayed treatment. Therefore, clinicians should maintain a high index of suspicion for NMOSD in patients with longitudinally

extensive spinal cord lesions and associated intractable vomiting, even in the absence of optic nerve involvement.

5. Conclusion

This case demonstrates the diagnostic and therapeutic challenges in managing NMOSD, particularly when initial symptoms are nonspecific. The coexistence of longitudinally extensive transverse myelitis and area postrema symptoms should prompt early consideration of NMOSD and AQP4 antibody testing. Failure to respond to corticosteroids should not delay the initiation of plasma exchange, which can significantly improve outcomes. With timely recognition and appropriate immunotherapy, complete neurological recovery is achievable, as observed in this patient (3, 4, 10)

Consent

The patient provided written informed consent for the publication of all clinical information and images included in this report.

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