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MODESTUM

**Case Report** **OPEN ACCESS**

# A diagnostic challenge of neuromyelitis optica spectrum disorder in a young woman with recurrent urinary symptoms

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**ABSTRACT**

Neuromyelitis optica spectrum disorder (NMOSD) is a rare, severe inflammatory autoimmune disease of the central nervous system, identified with the discovery of aquaporin-4 immunoglobulin G antibodies. Its ability to mimic other conditions often leads to misdiagnosis and delayed treatment. We report a 25-year-old woman with recurrent urinary tract infection symptoms. Despite antibiotics, her condition worsened with progressive myelitis symptoms such as urinary retention, back pain and lower limbs weakness. She was admitted to further imaging and blood investigations that confirmed NMOSD diagnosis. With appropriate treatment, she improved significantly. This is the diagnostic challenges of NMOSD in primary care. It is important to explore other symptoms such as neurological features as myelitis, in patients with recurrent urinary symptoms. Early recognition and intervention can substantially improve outcomes and quality of life.

**Keywords:** neuromyelitis optica spectrum disorder, NMOSD, systemic lupus erythematosus

## INTRODUCTION

Neuromyelitis optica spectrum disorder (NMOSD) is a rare, severe autoimmune disease of the central nervous system characterized by distinct core clinical features and following the discovery of aquaporin-4 immunoglobulin G antibodies (AQP4-IgG) status. The most frequent symptoms are optic neuritis, often causing severe vision loss, and acute myelitis, typically longitudinally extensive, leading to weakness, sensory impairment, and bladder or bowel dysfunction [1]. Other recognized features include area postrema syndrome with intractable nausea, vomiting, or hiccups; brainstem syndromes such as diplopia, vertigo, or dysphagia; and diencephalic or cerebral involvement presenting with narcolepsy, hypothalamic dysfunction, or encephalopathy. These attacks are usually severe, tend to relapse, and often leave residual disability [2]. Early recognition and treatment are therefore essential to improving long-term outcomes.

According to the 2015 international consensus diagnostic criteria, NMOSD can be diagnosed regardless of AQP4-IgG serostatus. However, seropositive cases are considerably more common [3]. Epidemiological based studies estimate the prevalence of NMOSD in Europe and North America range from < 1.0/100,000 to 4.4/100,000. Onset typically occurs between 35 and 45 years of age, although cases have been reported in both adolescents and the elderly. Compared with seronegative patients, seropositive individuals show a higher female predominance, approximately 9-10:1 vs. about 2:1. Although rare familial occurrences have been reported, most cases are sporadic and episodic in nature [4].

All patients presenting with acute transverse myelitis and/or acute optic neuritis, whether unilateral or, less commonly, bilateral should be evaluated for NMOSD. Those exhibiting both of these clinical features, together with suspected encephalitis or brainstem encephalitis, also should be evaluated for NMOSD as potential differential diagnosis. Optic neuritis and myelitis may occur concurrently or, more frequently, in, sequentially [5].

## CASE DESCRIPTION

A 25-year-old lady with no prior medical history presented with recurrent UTIs symptoms for two months. It was associated with back pain and bilateral feet pain. Despite antibiotics treatment, she progressed to urinary retention and was admitted under urology ward. She was also seen by orthopedic team, and they did not attribute her symptoms to cauda equina syndrome (CES). Urine cultures persistently grew *pseudomonas aeruginosa* which was sensitive to cefepime. Initial blood investigations revealed pancytopenia, liver transaminitis, elevated ESR, and LDH. Autoimmune screening showed negative ANA and with low levels of complement. She was treated with IV antibiotics and intermittent catheterization. She was discharged with a diagnosis of acute urinary retention secondary to UTI.

However, two weeks later, she was re-presented with fever, headache and intermittent vomiting in addition to her back pain and lower limb weakness. She also had oral ulcers and hair loss. On examination, she was hypotensive and tachycardic. Neurological examination revealed bilateral

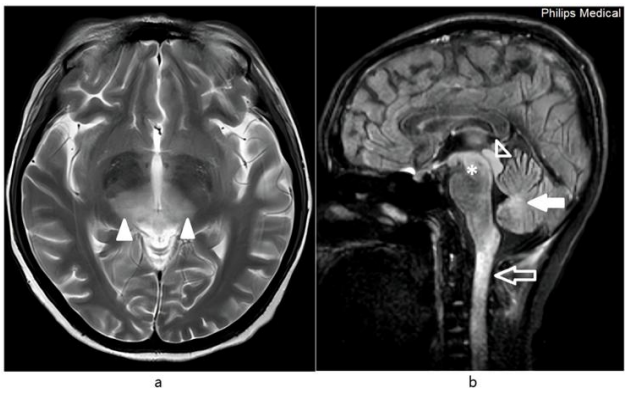


**Figure 1.** MRI brain in axial T2 weighted image (a) & sagittal FLAIR (b) shows abnormal signal intensities at bilateral thalami (arrowhead), quadrigeminal plate (open arrowhead), midbrain (*), middle cerebellar peduncle (arrow), & upper cervical spine (open arrow) (Reprinted with permission of the patient)

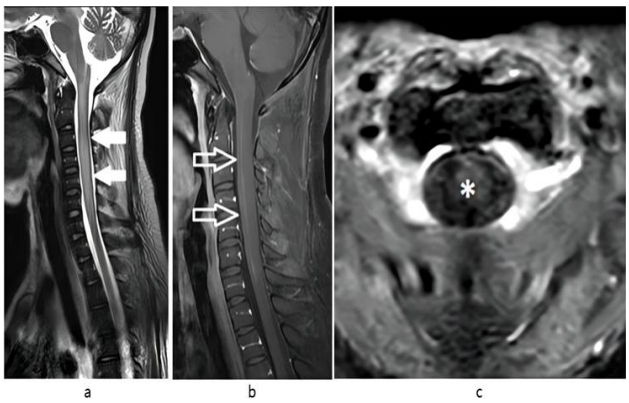


**Figure 2.** MRI upper cervical spine in sagittal T2 weighted (a), sagittal (b), & axial (c) post contrast sequences shows spine enlargement and hyperintensity at the upper cervical spine region (arrow), which showed enhancement (open arrow, *) post gadolinium (Reprinted with permission of the patient)

tremors, bidirectional nystagmus, oral ulcers and headache. Repeat blood tests showed worsening pancytopenia, liver enzyme elevations and this time with, positive ANA, anti-dsDNA, and low C3/C4 levels. With positive autoimmune bloods test, AQP4-IgG was taken but the result was negative. A diagnosis of systemic lupus erythematosus (SLE) with multiorgan involvement was then made.

Magnetic resonance imaging (MRI) brain was done and demonstrated extensive T2/FLAIR hyperintensities in the thalami, midbrain, medulla, cerebellar vermis, and upper cervical cord (**Figure 1** and **Figure 2**). These findings were consistent with neuropsychiatric SLE and probable with NMOSD. She was treated with IV methylprednisolone for five days, had plasma exchanged (PE) (five cycles), and IV cyclophosphamide (six cycles). Her condition improved and she was well upon discharge, even though she had a mild transfusion reaction during the packed cell administration. The final diagnosis for her was SLE with multi organ involvement, including hematologic and mucocutaneous manifestations, and seronegative NMOSD.

## DISCUSSION

NMOSD is diagnosed through clinical evaluation, serological testing for AQP4-IgG, and MRI with and without gadolinium contrast. In 2015, American Academy of Neurology established the international consensus diagnostic criteria for NMOSDs, classifying NMOSD according to AQP4-IgG status (positive, negative, or unknown) and specifying the required core clinical features and MRI features as in **Figure 3** [6]. There are six core clinical features that clinicians need to assess when seeing patients with suspected NMOSD. For our patient, since her serum AQP4-IgG was negative, she need two of the core clinical features. She presented with acute myelitis symptoms (urinary retention, back pain, and lower limbs weakness) and positive MRI findings in both the brain and spinal cord. However, her diagnosis was delayed by two months from the initial presentation, as our low suspicion towards NMOSD.

During her first admission, autoimmune disease was suspected when full blood count revealed pancytopenia with multiorgan involvement however her ANA test was rejected. While in the ward, she developed symptoms suggestive of lower motor neuron involvement, including back pain, bilateral lower limb neuropraxia, and urinary retention, raising suspicion of CES. CES typically results from lumbosacral nerve root compression, presenting with saddle anesthesia, bladder or bowel dysfunction, sexual disturbance, and bilateral radicular pain [7]. However, after ortho review and assessment, it was not CES.

On her second admission, the patient presented with headache, vomiting, urinary retention, and lower limb weakness, raising suspicion of acute myelitis. Investigations confirmed SLE with positive ANA and further serological markers, consistent with current diagnostic recommendations [8]. Core clinical features of NMOSD include acute bilateral or rapidly sequential optic neuritis causing visual loss, acute transverse myelitis leading to limb weakness and bladder dysfunction, and area postrema syndrome characterized by intractable hiccups, nausea, or vomiting [9]. MRI of the brain and spinal cord revealed findings suggestive of NMOSD, despite negative serum AQP4-IgG.

According to international diagnostic criteria, patients with negative or unknown AQP4-IgG require at least two core clinical features, with dissemination in space, and at least one episode of acute optic neuritis, acute myelitis, or area postrema syndrome. Moreover, standard MRI criteria must be met for diagnosis [5].

The conventional acute treatment for both AQP4-IgG-positive and negative NMOSD includes high-dose intravenous glucocorticoids and apheresis therapy. Methylprednisolone is commonly administered at 1,000 mg/day for 3-5 days, followed by a tapering oral regimen (1 mg/kg/day or 20-30 mg/day, reduced to 10-15 mg/day over 2-3 weeks), alongside gastric and thrombosis prophylaxis. In patients with poor response to steroids, early initiation of therapeutic PE or immunoadsorption is recommended, typically over 5 cycles on alternate days, although up to 10 may be required [3]. Without treatment, NMOSD attacks often carry an unfavorable prognosis, with complete recovery achieved in only a minority of cases. However, outcomes can be improved with early initiation of therapy and timely escalation. AS for our patient, she was treated with intravenous methylprednisolone and PE, resulting in significant clinical improvement and normalization of blood investigations.

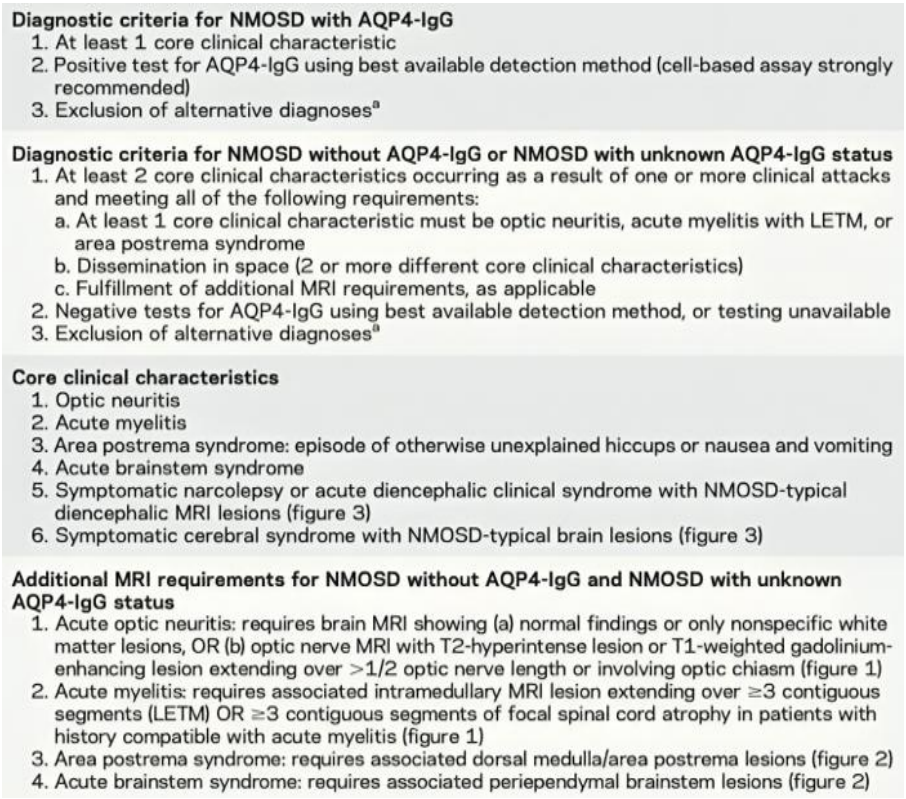


**Figure 3.** NMOSD diagnostic criteria for adult patients (international consensus diagnostic criteria for NMOSDs, 2015 American Academy of Neurology) (AQP4: Aquaporin; lgG: Immunoglobulin; & LETM: Longitudinally extensive transverse myelitis lesions) [6]

## COMPLICATIONS/PROGNOSIS

The prognosis of NMOSD attacks is typically poor without intervention, with complete recovery occurring in only a minority of patients. However, recovery might be enhanced through the timely initiation of therapy during acute episodes and early escalation. Factors like pre-existing conditions, the type and intensity of an attack, age, and, especially, the time interval before treatment begins, may affect recovery [3]. Considering the poor prognosis of the condition, lifelong therapy involving immunosuppressants and corticosteroids is advised by the literature. Fifty percent of patients experience significant movement disability and vision loss within five years, with a death rate reaching up to 25% [10]. Complications of NMOSD include visual field defects and motor dysfunction, potentially leading to blindness and irreversible motor impairments. In severe cases, myogenic respiratory failure contributes to elevated rates of mortality [11].

## CONCLUSION

The presentation of NMOSD can pose a significant diagnostic challenge, particularly in primary care. Recurrent urinary tract infections with lower back pain and bilateral lower limb neuropraxia may suggest alternative conditions such as CES, viral myelitis, paraneoplastic syndromes, or traumatic spinal cord injury. Given its rarity, the index of suspicion for NMOSD is often low, and diagnosis may be delayed or overlooked in the absence of typical features. Timely recognition is therefore essential. Patients presenting with recurrent UTIs and additional neurological symptoms should undergo comprehensive evaluation and detailed history-taking to maintain a high suspicion for NMOSD, thereby reducing the risk of misdiagnosis and treatment delays.

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**Data sharing statement:** Data supporting the findings and conclusions are available upon request from the corresponding author.

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