CASE REPORT

Neuromyelitis Optica Spectrum Disorder: An Interesting Case Report

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Abstract

We describe an interesting case of neuromyelitis optica (NMO). It should be understood and differentiated from multiple sclerosis because the treatment for both these conditions varies significantly. NMO spectrum disorder has recurrent episodes of vision loss and immunosuppressive agents remain the mainstay of treatment.

Keywords: Aquaporin-4, Magnetic resonance imaging, Neuromyelitis optica, Rituximab

INTRODUCTION

Neuromyelitis optica (NMO) is a rare condition of the central nervous system and is an inflammatory autoimmune disorder. Patients usually present with optic neuritis (ON) or transverse myelitis. Previously, NMO was thought to be one of the variants of multiple sclerosis and was treated accordingly with poor outcomes. It has now been recognized as a distinctly separate entity with characteristic clinical, radiological, pathological, and serological features. Hence, it is important for ophthalmologists and neurologists to have a high index of suspicion and thorough knowledge of NMO to provide early diagnosis and appropriate treatment. This is essential to preserve the visual and neurological function of these young patients, thereby reducing morbidity.

MATERIALS AND METHODS

A 19-year-old girl presented with complaints of difficulty in using all four limbs and difficulty in passing urine and stool for 20 days. She initially developed itching over the left side of her face, neck, and left shoulder without any visible skin lesions 20 days ago. Over the next 3 days, numbness developed in the left upper limb descending to the left side of the trunk and left lower limb which was insidious in onset and gradually progressive. Ten days later the patient developed weakness which was gradually progressing and she had difficulty holding objects. She then noticed numbness over her right shoulder which gradually descended to the right upper limb, right side of the trunk, and right lower limb over the next 5 days. Four days later, patient gave a history of difficulty in passing urine and inability to pass stools. She developed a diminution of vision suddenly in onset and was painless in nature. Her best corrected visual acuity in the right eye was 1/60 and in the left eye was 6/60. Pupillary reflexes in both eyes were reactive to both direct and consensual light reflexes. Fundus examination in both eyes showed an optic disc with well-defined margins and a cup disc ratio of 0.3. Color vision and visual field assessment could not be done because of poor vision. Magnetic Resonance Imaging of the Cervical spine showed ill-defined long segment intramedullary T1 hypointense/T2 Hyperintense lesions causing spinal cord expansion from the medulla oblongata to the C5 vertebral level. VEP of both eyes showed Bilateral prolonged P 100 latency RE>LE suggestive of bilateral demyelinating optic neuropathy. Cerebrospinal fluid tested positive for Aquaporin-4 (AQP4). The patient was started on intravenous immunoglobulins, steroids, and injection Rituximab. After the 2nd dose of immunosuppressant visual acuity, both eyes improved to 6/6.

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Table 1: Summary of 2015 international diagnostic criteria for neuromyelitis optica spectrum disorder (3)

Diagnosis with AQP4-Abs
> At least one core clinical characteristic
> Positive AQP4-Abs with the best available method
> Exclusion of alternative diagnosis (e.g. sarcoidosis, neoplastic/ paraneoplastic, vascular, chronic infection).
Diagnosis without AQP4-Abs/unknown status
> At least two core clinical characteristics resulting from one or more clinical attacks and fulfilling the following:
> At least one of optic neuritis, LETM, APS
> Dissemination in space (two or more different core clinical characteristics)
> Fulfilment of additional MRI requirements as applicable
> Negative for AQP4-Abs with the best available method, or testing unavailable
> Exclusion of alternative diagnoses.
Core clinical characteristics
> Optic neuritis
> Acute myelitis
> APS
> Brainstem syndrome
> Symptomatic narcolepsy or acute diencephalic syndrome with NMOSD-typical diencephalic MRI lesions
> Symptomatic cerebral syndrome with NMOSD-typical brain lesions.
Additional MRI requirements for NMOSD without AQP4-Abs/unknown status
> Acute optic neuritis: normal or only non-specific white matter lesions on MRI brain; or optic nerve MRI with T2-hyperintense lesion or T1-weighted gadolinium-enhancing lesion extending over >1⁄2 optic nerve length or optic chiasm involvement
> Acute myelitis: MRI spinal cord demonstrating attack-associated lesion spanning ≥3 contiguous vertebral segments (LETM); or ≥3 contiguous segments of focal cord atrophy with a previous history of acute myelitis
> APS: dorsal medulla/area postrema lesion on MRI brain
> Acute brainstem syndrome: periependymal brainstem lesion

MRI: Magnetic resonance imaging, NMOSD: Neuromyelitis optica spectrum disorder, AQP4: Aquaporin-4

DISCUSSION

NMO is a severe autoimmune inflammatory demyelinating disease of the central nervous system characterized by the production of antibodies against water channels in the foot processes of astrocytes.[1] NMO is recognized as an autoimmune water channelopathy.

It is crucial to differentiate NMO from Multiple Sclerosis to ensure patients achieve good visual outcomes. Recognizing typical brain lesions in NMO spectrum disorder (NMOSD) involves considering areas with consistent AQP4 expression, such as the subpial regions, periependymal regions, circumventricular organs, brainstem, chiasm/hypothalamus, and corpus callosum. Although the corticospinal tract is not highly expressive of AQP4, it is observed in 23–44% of NMOSD patients.[2] The cerebrospinal fluid typically shows mixed pleocytosis (lymphocytes, neutrophils, and monocytes) with a median cell count of 19 cells/μL (range: 6–380).[3] Unlike MS, CSF-restricted oligoclonal bands are uncommon.

ON in NMOSD is typically longitudinally extensive, affecting the posterior optic nerve segments, especially the optic chiasm. Bilateral simultaneous involvement and severe visual loss with poor recovery (<6/60) are often indicative of NMOSD.

MRI of the brain and spinal cord, along with Anti-AQP4 antibody assay, play crucial roles in diagnosing NMOSD. Although the Anti-AQP4 antibody is highly specific (75%), a negative assay for NMO-immunoglobulin G (IgG) does not rule out the diagnosis.[4] NMOSD can coexist with other autoimmune disorders such as myasthenia gravis, Sjogren's syndrome, and rheumatoid arthritis.

Before confirming the diagnosis through seropositivity or radiological imaging, patients suspected of having NMOSD-ON should be started on high-dose steroids for 5 days and observed. For confirmed NMOSD, long-term immunosuppressive treatment with drugs, such as azathioprine and mycophenolate mofetil is recommended. Recurrent attacks are common and can be severe, thus requiring maintenance doses of steroids (>10 mg/day). Immunotherapy should be administered for at least 24 months if the patient is seropositive, with azathioprine, mycophenolate, and rituximab being commonly used. Plasmapheresis is indicated to maximize functional recovery in case of relapse. Unlike in multiple sclerosis, interferon therapy worsens NMOSD progression.

Campetella *et al.* postulated that the AQP4-IgG positivity in myelitis (AIM) score predicted AQP4-IgG positivity with 85% sensitivity and 95% specificity.[5] A study on Malaysian patients with idiopathic inflammatory demyelinating diseases at high risk for NMO/NMOSD found that 65 out of 96 patients (67.7%) were positive for anti-AQP4 IgG.[6]

CONCLUSION

This case is being presented to highlight the neurological and visual manifestations of NMO which should be

closely differentiated from multiple sclerosis for a better prognosis.

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