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ACUTE BILATERAL OPTIC NEURITIS IN A CASE OF NEUROMYELITIS OPTICA SPECTRUM DISORDER – A CASE REPORT FROM TERTIARY CARE CENTRE.

Opthalmology

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ABSTRACT

Background: Neuromyelitis optica spectrum disorder (NMOSD) is an uncommon autoimmune demyelinating disease of the central nervous system, characterized by recurrent attacks of optic neuritis and longitudinally extensive transverse myelitis. **Case History:** We report a case of a 17-year-old female who presented with acute bilateral optic neuritis as the initial manifestation of NMOSD. She had sudden, painless diminution of vision in both eyes, more severe in the right eye. Clinical examination showed marked visual impairment, relative afferent pupillary defects, and optic disc pallor on fundus evaluation. MRI of the brain and orbits revealed bilateral optic nerve inflammation, and serum aquaporin-4 (AQP-4) IgG antibodies were positive, confirming the diagnosis of NMOSD. Other laboratory parameters were within normal limits. **Conclusion:** NMOSD is a rare but serious autoimmune disorder primarily affecting the optic nerves and spinal cord through AQP-4 antibody-mediated astrocyte injury. Accurate diagnosis using clinical features, antibody testing, and MRI findings is crucial. While the disease can lead to significant and lasting neurological deficits, prompt and appropriate treatment—especially in cases without spinal cord involvement—can improve outcomes and reduce relapse frequency. Longterm immunosuppression and careful monitoring are essential for disease control.

KEYWORDS

Acute bilateral optic neuritis, neuromyelitis optica spectrum disorder, AQP-4 IgG, autoimmune demyelination, vision loss.

INTRODUCTION

Acute bilateral optic neuritis is a severe presentation of neuromyelitis optica spectrum disorder (NMOSD), a multifocal, inflammatory, demyelinating autoimmune condition of the central nervous system. Unlike the more common unilateral optic neuritis seen in multiple sclerosis, bilateral involvement—particularly in young individuals—should raise suspicion for NMOSD, especially when associated with poor visual recovery.

NMOSD primarily affects the optic nerves and spinal cord. Isolated optic neuritis is a common presenting feature. It is now recognized as a distinct entity from multiple sclerosis, owing to the identification of aquaporin-4 (AQP4) IgG antibodies—pathogenic autoantibodies directed against astrocytic water channels.

Historically, neuromyelitis optica spectrum disorder (NMOSD), formerly known as neuromyelitis optica or Devic's disease, was first described by Dr. Eugène Devic while evaluating a patient with optic neuritis accompanied by neuromuscular symptoms.

In NMOSD, optic neuritis tends to be more severe, recurrent, and often bilateral, potentially leading to profound and permanent vision loss if not diagnosed and treated early. Given its devastating potential, acute bilateral optic neuritis must be approached with urgency and a high index of suspicion for NMOSD.

Case History

A 17-year-old female presented to the Ophthalmology OPD at G.G.G. Hospital, Jamnagar, with complaints of sudden, painless diminution of vision in both eyes for the past 15 days, more pronounced in the right eye. There was no history of eye redness, pain, photophobia, metamorphopsia, or floaters. There was also no history of trauma or any major systemic illness.

At presentation, the patient's visual acuity was severely impaired. The right eye had perception of light (PL+), but projection of rays (PR) was faulty. The left eye could count fingers at a distance of 2 meters; however, vision did not improve further with pinhole. Colour vision could not be assessed in either eye due to the poor level of visual acuity. Intraocular pressure, measured using non-contact tonometry, was 17 mmHg in the right eye and 15 mmHg in the left.

Anterior Segment Findings:-

Right eye	Anterior segment	Left eye
Normal	Lid	Norma
Normal	Conjunctiva	Normal
Normal	Sclera	Normal
Clear	Cornea	Clear
++	Anterior chamber	++
Normal	Iris	Normal
Afferent reactivity of pupil (RAPD grade-3)	Pupil	Afferent reactivity of pupil (RAPD grade-3)
Clear	Lens	Clear
Following commands	Ocular movements	Following commands

Dilated Fundus Examination (fig. 1&2):-

Right eye	Fundus	Left eye
Clear	Media	Clear
Disc pallor	Disc	0.4 CDR with Temporal disc pallor
Normal vessels	Blood vessels	Normal vessels
Wandering fixation	Macula	Foveal fixation
No significant findings	General fundus	No significant findings

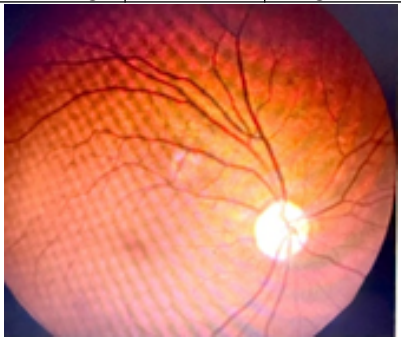

Fig. 1: Right Eye

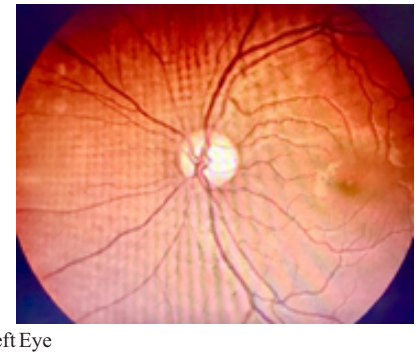

Fig. 2: Left Eye

Radiological evaluation with MRI of the brain and orbits revealed bilateral inflammation of the optic nerves—findings suggestive of optic neuritis and highly indicative of neuromyelitis optica spectrum disorder (NMOSD). Serological testing confirmed the diagnosis, with a positive serum aquaporin-4 (AQP4) IgG antibody. Other clinical investigations, including serum antinuclear antibody (ANA), Quantiferon–Gold TB assay, cerebrospinal fluid (CSF) examination, complete blood count (CBC), renal function tests (RFT), and liver function tests (LFT), were all unremarkable.

The prognosis was explained to the patient, and she was initiated on a five-day course of intravenous methylprednisolone. She was subsequently started on long-term systemic corticosteroid therapy.

DISCUSSION

Neuromyelitis optica spectrum disorder (NMOSD) is an inflammatory and demyelinating disorder of the central nervous system. It is classified as an autoimmune astrocytopathy, where AQP4-IgG antibodies mediate perivascular lymphocytic infiltration, ultimately leading to axonal loss in specific regions.[1]

Aquaporin-4 is a transmembrane water channel located on the foot processes of astrocytes. It is highly concentrated in the optic nerves, spinal cord, area postrema, and other parts of the central nervous system. Circumventricular regions, including the periaqueductal gray matter, are rich in AQP4 and are more commonly involved in NMOSD.[1]

The binding of AQP4-IgG antibodies initiates complement-mediated cytotoxicity, resulting in astrocytic injury, oligodendrocyte damage, demyelination, and neuronal loss.[5]

IgG antibodies against aquaporin-4 are detected in approximately 60% to 90% of patients with NMOSD, with a specificity of 90% and sensitivity ranging from 70% to 90%.[4]

Whole-genome sequencing has identified several at-risk genotypes for NMOSD, including major histocompatibility complexes such as HLA-DRB103:01.[3]

In this case, the bilateral presentation of optic neuritis, along with MRI findings showing posterior optic nerve involvement and positive AQP4-IgG antibodies, confirms the diagnosis of NMOSD. The absence of spinal cord involvement and early initiation of treatment are associated with a more favorable visual prognosis.

NMOSD is a rare condition with an estimated prevalence of 0.3 to 4.4 cases per 100,000 population.[6] It exhibits a strong female predominance, with a female-to-male ratio of nearly 10:1. The age of onset varies significantly, with about one-sixth of AQP4-IgG-positive cases occurring in pediatric or elderly patients. The median age of onset is approximately 40 years. NMOSD is also associated with other autoimmune disorders, including systemic lupus erythematosus (SLE), celiac disease, Sjögren's syndrome, and sarcoidosis.

Clinical Manifestations

Ophthalmic involvement is a prominent feature of NMOSD, with approximately half of the patients presenting with isolated optic neuritis, and about 20% of these cases exhibiting bilateral involvement. The resulting visual loss is often profound and persistent.[17]

Patients with optic neuritis in NMOSD typically present with decreased visual acuity, visual field defects, or impaired color vision (red desaturation). A relative afferent pupillary defect (RAPD) may be observed in unilateral or bilaterally asymmetric optic nerve involvement.[8] Fundus examination commonly reveals optic nerve head elevation, while optical coherence tomography (OCT) demonstrates significant peripapillary retinal nerve fiber layer (RNFL) loss.[10] Macular thinning may also be observed and serves as an additional, indirect indicator of axonal damage.[11]

In addition to ocular findings, patients may present with neurological symptoms such as intractable hiccups, vomiting, narcolepsy, brainstem encephalitis, and longitudinally extensive transverse myelitis.[2]

Diagnostic Evaluation

According to Wingerchuk et al., optic neuritis is a core clinical characteristic in the diagnosis of neuromyelitis optica spectrum disorder (NMOSD), along with others such as acute myelitis, area postrema syndrome, acute brainstem syndrome, acute diencephalic clinical syndrome, and symptomatic cerebral s opyndrome, as outlined in the 2015 International Panel for NMO Diagnosis (IPND) criteria. In patients who are seropositive for aquaporin-4 immunoglobulin G (AQP4-IgG), a single episode of optic neuritis—when accompanied by positive serology and exclusion of alternative diagnoses—is sufficient to establish the diagnosis of NMOSD. However, in AQP4-IgG-negative or untested individuals, optic neuritis alone is not diagnostic; it must occur in conjunction with at least one other core clinical feature, such as longitudinally extensive transverse myelitis (LETM) or area postrema syndrome, and meet specific MRI criteria. NMOSD-related optic neuritis tends to be more severe than that seen in multiple sclerosis (MS), often presenting with bilateral involvement, chiasmal extension, or lesions affecting more than half the length of the optic nerve, potentially leading to profound visual loss (visual acuity ≤20/200). Certain features raise red flags for alternative diagnoses, such as partial optic neuritis without LETM, the presence of typical MS lesions on brain MRI, or positive cerebrospinal fluid (CSF) oligoclonal bands. These distinguishing features aid in differentiating NMOSD from MS and other demyelinating conditions, particularly when AQP4-IgG is absent or testing is unavailable.[2]

AQP4 antibody serum levels are not only specific to NMOSD but also correlate with disease activity. Testing for AQP4 is recommended during an acute attack and before initiating immunosuppressive therapy. A small number of patients with clinical characteristics of NMOSD—mostly AQP4-IgG seronegative—may have detectable serum myelin oligodendrocyte glycoprotein (MOG) antibodies. The absence of CSF oligoclonal bands supports a diagnosis of NMOSD over multiple sclerosis, although they may be transiently detectable during an acute NMOSD attack. CSF pleocytosis with more than 50 leukocytes per microliter, or the presence of neutrophils or eosinophils, is also helpful in distinguishing NMOSD from MS.[12] The glial fibrillary acidic protein (GFAP) level is markedly elevated in the CSF of AQP4-antibody-seropositive NMOSD patients during acute exacerbations, whereas CSF GFAP is not elevated in typical multiple sclerosis.[16]

MRI with or without gadolinium contrast typically shows T2 hyperintense enhancement of the optic nerves, often involving both sides and extending to the posterior segments and optic chiasm—findings that are highly suggestive of NMOSD. The anterior visual pathway is anatomically divided into ten segments, including the orbital, intracanalicular, and intracranial parts of both optic nerves, halves of the chiasm, and the optic tracts. Involvement of the spinal cord with longitudinal lesions extending over three or more vertebral segments is another hallmark feature.[2] Bright spotty lesions in the central gray matter of the spinal cord that are intensely hyperintense on T2 (brighter than cerebrospinal fluid) and hypointense on T1 are considered specific for NMOSD. Acute lesions are usually enhanced with gadolinium contrast.[1] In seronegative cases, where antibody sensitivity may be limited, these imaging and clinical criteria are essential for diagnosis.

Treatment

Acute management of optic neuritis in NMOSD involves high-dose intravenous methylprednisolone at 1,000 mg per day for 5 to 10 days. In steroid-resistant cases, plasma exchange (PLEX) is indicated. For long-term treatment, immunosuppressive agents such as azathioprine, mycophenolate mofetil, and rituximab are commonly used. Novel biologic therapies targeting interleukin-6, complement pathways, and AQP-4 antibodies are currently under investigation and show promise as future treatment options.[9]

Prognosis

Although NMOSD generally carries a poor prognosis due to its relapsing nature, factors such as younger age at onset, a monophasic presentation, absence of myelitis, and prompt treatment can significantly improve outcomes.[15]

CONCLUSION

Neuromyelitis optica spectrum disorder (NMOSD) is a rare but serious autoimmune condition that primarily affects the optic nerves and spinal cord through AQP-4 antibody-mediated astrocyte injury. Accurate diagnosis using clinical features, antibody testing, and MRI

findings is crucial. Although the disease can lead to significant and lasting neurological deficits, prompt and appropriate treatment—particularly in cases without spinal cord involvement—can improve outcomes and reduce relapse frequency. Long-term immunosuppression and careful monitoring are essential for effective disease control.

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