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Neuromyelitis Optica Spectrum Disorder -Diagnostic Dilemma

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Abstract
Background: *Neuromyelitis optica or Devics disease is a rare inflammatory demyelinating autoimmune disease of CNS affectingspinal cord and optic nerve with prevalence of 1-10 / 100,000 individuals worldwide. It has varied manifestations like loss of vision in one or both eyes with optic neuritis developing within days to weeks with subsequent transverse myelitis, grouped as spectrum disorder (NMOSD) .*
Case Presentation: *A 24yr old obese female with diminution of vision with pain in LE, headache since 10 days followed by numbness of bilateral upper and lower limbs, difficulty in walking and getting up from squatting position for 3 days. On ocular examination, BCVA in LE was 2/60, RAPD in left pupil. Fundus examination showed LE disc edema. Reduced power in upper and lower limbs. Tests for syphilis, HIV, SLE were negative.AQP4-Ab was negative whereas MOG Ab was weak positive. CE-MRI spine was suggestive of longitudinally extensive transverse myelitis, normal CE-MRI brain. NMO was diagnosed using IPND diagnostic criteria. The patientwas started on IVMethylprednisolone and tapered subsequently.*
CONCLUSION:*NMOSD is confirmed by antibody biomarkers; negative report can lead todiagnostic dilemma. AQP4-Ab testing isimportant for diagnosis with imaging studies; however negative antibody cannot exclude thediagnosis.*

Keywords: *Neuromyelitis optica, optic neuritis, AQP4 antibody*

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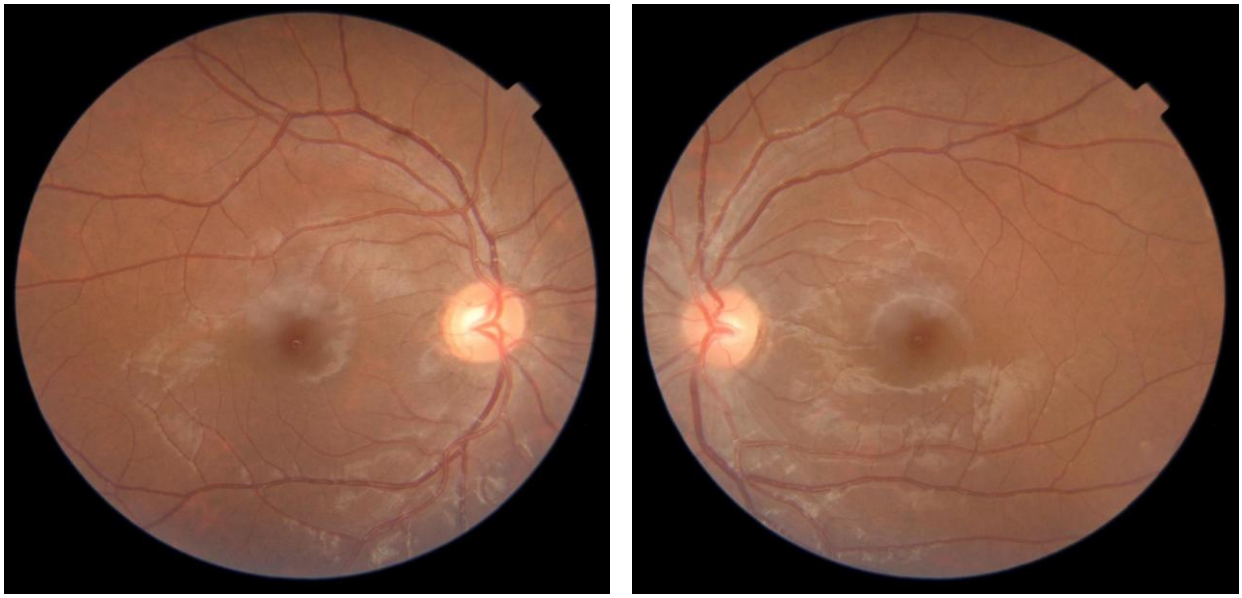
I. Introduction

Neuromyelitis optica / Devic's disease is a rare inflammatory demyelinating autoimmune disease of CNS affecting spinal cord and optic nerve with varied manifestations. It is different from Multiple sclerosis and is associated with serum Aquaporin-4 immunoglobulin G (AQP4-IgG)[3]. There are many patients who meet the criteria for NMO with serum Aquaporin-4-IgG. It has varied manifestations like loss of vision in one or both eyes with optic neuritis developing within days to weeks with subsequent transverse myelitis[1]. This disease is grouped as spectrum disorder (NMOSD) in young to middle aged patients with female preponderance[3].

II. Case Report

A 24 year old obese female presented with pain and diminution of vision in left eye and headache since 10 days followed by numbness of both upper and lower limbs, difficulty in walking and getting up from squatting position for 3 days. There was no history of fever, diplopia, dysarthria, dysphagia, sphincter disturbances, no similar episodes in the past. No history of Diabetes Mellitus, Hypertension, Thyroid disorder. On examination, patient's vitals were normal. Cardiovascular system, respiratory system and abdominal exam were normal. Patient's visual acuity in the right eye was 6/6 whereas visual acuity in the left eye was 2/60. Colour vision in the right eye was normal, left eye colour vision could not be assessed due to low vision. Relative afferent pupillary defect noted in left eye. Power in both upper and lower limbs was 4/5. Head posture was erect, ocular position was orthophoric, facial symmetry was normal. Slit lamp examination was normal for both eyes. Dilated fundus examination showed hyperaemic disc and blurring of nasal margins of left optic disc.

Left eye optic disc showing hyperaemia and blurring of nasal margins



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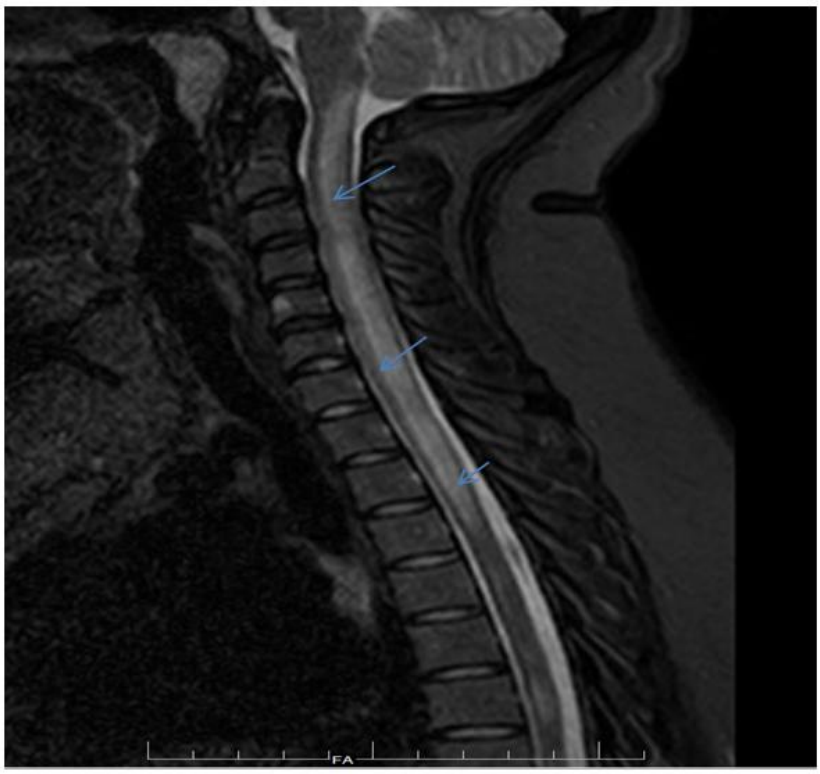
Blood investigations like complete blood picture was normal, renal function tests were normal, serum electrolytes were normal, liver function tests were normal, thyroid function was normal, serology was normal, Serum ACE was normal, chest x ray was normal, CSF Fluid analysis showed predominantly lymphocytes, Serum Aquaporin 4 Antibody was negative, Serum myelo oligodendrocyte Antibody was weak positive. CE MRI Brain was normal, MR Cerebral Angiogram was normal, MR Cerebral Venogram was normal. CE MRI Spine was suggestive of longitudinally extensive transverse myelitis.

T2/STIR high intensity noted along the cervical cord from cervico-medullary junction upto D3 vertebra and cord expansion



Patient was diagnosed as Neuromyelitis Optica Spectrum Disorder (NMOSD) with serum negative AQP4Ab according to 2015 revised International Panel for NMO Diagnosis Criteria for Neuromyelitis Optica Spectrum Disorder.

Patient was started on INJ.Methylprednisolone1g IV OD for 5 days and then changed to oral Prednisolone 40mg OD for 1 month and was tapered subsequently over 3 months. Patient improved symptomatically.

III. Discussion:

Neuromyelitis Optica Spectrum Disorder is an autoimmune disease of the central nervous system with a prediliction for the optic nerve, spinal cord and brain stem region[2]. The epicentre of autoimmunity is defined by IgG antibody to AQP4 water channel protein in astrocyte foot processes of CNS structures[6]. Serum AQP4-IgG is detected in 80% of patients with NMOSD. Half of AQP4-IgG negative NMOSD are categorised under myelin oligodendrocyte glycoprotein (MOG) positive NMOSD[2]. The patient fulfilled the International Panel for NMO diagnosis criteria for NMOSD with seronegative AQP4Ab with presence of 2 core clinical characteristics such as optic neuritis and acute myelitis. It has shown dissemination in space at 2 sites. The T2 spine MR image revealed longitudinally extensive transverse myelitis (LETM) with spinal lesion extending >= 3 contiguous vertebral segments. Differential Diagnosis include Multiple Sclerosis, SLE, Neurosarcoidosis, Subacute combined degeneration, Acute demyelinating encephalomyelitis.

Table 1 The 2015 International Panel for NMO Diagnosis criteria for neuromyelitis optica spectrum disorder (NMOSD)[24]

Diagnostic criteria for NMOSD with AQP4-IgG
At least 1 core clinical characteristic
Positive test for AQP4-IgG using best available detection method (cell-based assay strongly recommended)
Exclusion of alternative diagnoses
Diagnostic criteria for NMOSD without AQP4-IgG or NMOSD with unknown AQP4-IgG status
At least 2 core clinical characteristics occurring as a result of one or more clinical attacks and meeting all of the following requirements:
At least 1 core clinical characteristic must be optic neuritis, acute myelitis with LETM, or area postrema syndrome
Dissemination in space (2 or more different core clinical characteristics)
Fulfillment of additional MRI requirements, as applicable
Negative tests for AQP4-IgG using best available detection method, or testing unavailable
Exclusion of alternative diagnoses

Abbreviations: AQP4-IgG = aquaporin-4 immunoglobulin G antibody; LETM = longitudinally extensive transverse myelitis.

PROGNOSIS:

The clinical course of NMO is more severe as compared to multiple sclerosis, NMO has high relapse rates (high AQP4-Ab titres). Seronegative patients have mild visual impairment as compared to seropositive patients.

IV. Conclusion:

The diagnosis is confirmed by antibody biomarkers, negative report can lead to diagnostic dilemma. Early stages of Multiple sclerosis mimics NMO, making it difficult to distinguish. AQP4Ab testing is important for diagnosis with imaging studies; however negative antibody cannot exclude the diagnosis but rather grouped into seronegative variety.

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