Neuromyelitis Optica Spectrum Disorder (Nmosd): A Rare Report

Daniel Sundar Singh, Shailaja Krishnamoorthy, Divya Thirumaran, Ektha Promoth Kumar, and Harshidha Dharmalingam

ABSTRACT

Neuromyelitis Optica spectrum disorder (NMOSD), also known as Devic disease, is a chronic disorder of the brain and spinal cord dominated by inflammation of the optic nerve (optic neuritis) and inflammation of the spinal cord (myelitis). We present the case of a 25-year-old female patient who presented with complaints of lower back ache, decreased vision and double vision, generalized body pain, neck pain and vomiting. On examination, Extraocular Movement (EOM) showed right gaze restriction with nystagmus; her serum Neuromyelitis Optica (NMO) result was positive; and her MRI showed lesions in the brain. Insight of her presenting illness and investigations, she was successfully treated with IV steroids, plasmapheresis, and immunosuppressive therapy and was neurologically stable. We present this rare case to clinically enlighten the diagnosis and treatment of NMOSD.

Keywords: Brain, Devic’s disease, Myelitis, Optic-neuritis, Neuromyelitis Optica spectrum disorder (NMOSD), Spinal cord.

I. BACKGROUND

Neuromyelitis Optica spectrum disorder (NMOSD), also known as Devic disease, is a chronic disorder of the brain and spinal cord dominated by inflammation of the optic nerve (optic neuritis) and inflammation of the spinal cord (myelitis) [1]. According to [2], consistently, the prevalence range of NMOSD is 0.5–4/100,000 and may be up to 10/100,000 in certain racial groups. Aquaporin-4 (AQP4) antibody-seropositive, the average age of onset for NMOSD is around 40 years, and the female-to-male ratio can reach up to 9:1 [2].

Optic neuritis or myelitis are the defining signs of NMOSD; either one may appear as the initial symptom [1]. Optic neuropathy causes sudden loss of vision in one or both eyes within days to weeks, which then develops into transverse myelitis and variable degrees of weakness or paralysis in the arms or legs, a lack of sensation, and/or problems with the bladder and bowels [3], [4]. NMOSD and multiple sclerosis (MS) can cause optic neuritis and myelitis as symptoms, it may be challenging to distinguish between both conditions early in the course of the disease. Although the brain MRI is frequently normal and the spinal fluid study typically does not reveal oligoclonal bands in NMOSD, the optic neuritis and myelitis in NMOSD tend to be more severe than in MS. A highly specific and only modestly sensitive diagnostic test for NMOSD is Aquaporin-4 Immunoglobulin G (AQP4-IgG). It has been demonstrated that it can identify antibodies that are specific to the astrocyte protein AQP4 [1].

II. CASE PRESENTATION

A 25-year-old female presented with complaints of lower back ache for 10 days, decreased vision and double vision for 3 days, generalized body pain, neck pain, and vomiting. She had a history of fever and initially had weight loss. On examination, the patient was conscious, oriented, and awake. Her BP (160/100 mmHg) and HR (105/ min) were found to be elevated. Her Extraocular Movement (EOM) showed right gaze restriction with nystagmus, pupils were normal and reactive to light, right sided diplopia and ataxia were present. Based on the laboratory investigations, this particular individual was anemic and her platelets, neutrophils, CRP, and ESR were elevated. Urine analysis revealed hematuria and proteinuria. Antinuclear antibodies (ANA) were found to be positive. CSF proteins and lymphocytes are found to be elevated. Her MRI BRAIN P+C showed multiple patchy foci of altered signal T1 hypo T2/FLAIR hyperintensity seen involving body of corpus callosum, right cingulate gyrus,
right median temporal lobe, right caudate, left gangliocapsular region, mid brain, pons (predominantly in dorsum) bilateral middle cerebellar peduncle and medulla. Diffuse restriction was seen in the right caudate (Fig. 1), left gangliocapsular region, and medulla in the median aspect. WHOLe SPINE SCREENING revealed linear T2 hyperintense signal seen within anterior cord at level of D3 likely syrinx and mild diffuse disc bulge at L4/L5 and L5/S1 causing bilateral neural foraminal compromise. Her serum Neuromyelitis Optica (NMO) reports were positive and lupus anticoagulants were detected. With the above-mentioned history and complaints, she was initially treated with IV steroids and symptomatically treated after getting an opinion from a clinical specialist in regard to this. She was started on plasmapheresis. She showed considerable improvement after 5 days of plasma exchange. However, the need for further escalation of treatment (Rituximab or Cyclophospham ide) if needed has been explained. Hence, she was started on 1 gram of Rituximab infusion as per protocol after premedication. Then she got her second dose of injection rituximab after one month, and the patient was stable.

Fig. 1. MRI Brain.

III. DISCUSSION

The phrase “acute neuromyelitis optic” was initially coined by Eugene Devic (1858–1930) to describe a new syndrome characterized by myelitis and acute optic neuritis. IgG-NMO or IgG-AQP4, the distinctive antibodies that distinguish NMOSD from MS, were found by [5].

IgG1-isotype antibodies make up the majority of AQP4- Abs. According to experimental studies, AQ P4- Abs cause astrocytes that express AQP4 to produce interleukin-6 (IL-6), and IL-6 signaling to endothelial cells impairs blood-brain barrier function. Aside from internalizing the glutamate transporter EAAT-2, AQ P4- Abs cause complement- and cell-mediated astrocytic damage after they are attached to the extracellular domain of the AQP4 receptor. As a result, the astrocyte is rendered helpless, ultimately leading to the loss of support for neighboring cells, including oligodendrocytes and neurons. Following granulocyte infiltration, oligodendrocyte destruction and demyelination occur. In contrast to MS, the demyelination found in NMOSD is a secondary event that results from primary astrocyte injury [6]. In NMOSD, clinical syndromes like bilateral optic neuritis, longitudinally extensive transverse myelitis, region postrema syndrome, acute brain stem syndrome, diencephalic syndrome, and symptomatic cerebral syndrome were seen. This individual was diagnosed based on the International Panel for NMOSD Diagnostic Criteria. The patient fulfilled the diagnostic criteria for “NMOSD with unknown AQP4-IgG” as evidenced by the presence of two core clinical characteristics such as optic neuritis and acute myelitis [7].

In our case, the serum NMO-IgG was found to be positive, whereas in other case it was found to be negative, which was reported by [9].

Treatment of NMOSD in Acute exacerbation phase therapy to lower the probability of relapse and long-term care. Oral corticosteroids, immuno suppres sant therapy, Therapeutic Plasma Exchange (TPE), immunosuppressive therapy, and other novel medicines are available as treatment options for preventing relapse [8]. This patient was initially treated with corticosteroids, in spite of that she developed new lesions and hence she was started on plasmapheresis. Then, to optimize the treatment, this patient was put on 1 gm Rituximab infusion as per protocol after premedication. During infusion, she had a complaint of numbness over her throat region. Hence, infusion was stopped and 100 mg of hydrocortisone injection stat and 1 ampule of pheniramine injection were administered. After resolving complaints, the infusion was restarted and completed. Other supportive medication was given. She became neurologically stable.

The elimination of B cells as antigen-presenting cells and a decrease in the CD20+ early plasma blast population that produces anti-aquaporin-4 antibodies are two of the theorized mechanisms of action of rituximab in NMOSD [8].

In this report, the patient was administered rituximab as a drug of choice, which was different from the case report in which azathioprine was used as the treatment option, reported by [6].

IV. CONCLUSION

NMOSD is an autoimmune disease of the central nervous system with a predilection for the optic nerves, spinal cord, and brain stem regions. This case demonstrates that neuromyelitis optica spectrum illness can be found in clinical settings in underdeveloped countries. To prevent delayed diagnosis and treatment, a high index of suspicion for this rare condition is necessary. This patient had been successfully treated with intravenous Steroids, Plasma exchange therapy and Immunosuppressive drugs.

V. PATIENT’S CONSENT

Written informed consent was obtained from the patient for the publication of this case report.

ACKNOWLEDGMENT

We would like to thank the patient and her family for their cooperation and full support.

CONFLICT OF INTEREST

The authors have no conflicts of interest.
REFERENCES


