Profound bilateral ophthalmoplegia and ptosis as a first presentation of NMOSD: A case report

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Objective

To highlight the following:

- **less typical brainstem syndromes seen in Neuromyelitis optica spectrum disorder (NMOSD)**
- **the importance of recognizing ascending and descending fibers that may be affected in a more diffuse brainstem lesion,**
- **the salient imaging pattern that can be seen in NMOSD.**

Introduction

- NMOSD is an autoimmune astrocytopathy that mainly affects middle-aged women with a female:male ratio of 9:1.
- Optic neuritis (ON) and transverse myelitis (TM) are the predominant presenting features, with 85% of patients presenting with either one or both as a first sign of the disease.
- Brainstem, cerebral, and diencephalic events can occur, including narcolepsy and osmotic demyelination.
- Less commonly involved brainstem nuclei may present with under-recognized symptoms that delay diagnosis and treatment, leading to a poorer treatment response and greater residual disability.
- **Awareness of less common presentations can aid in timely NMOSD recognition and intervention.**

Methodology

- A 38-year-old female of Filipino descent presented with a two-week history of progressive bilateral ptosis, ophthalmoparesis, ageusia, and tinnitus.
- The patient also reported a severely pruritic area on her right back in the T4 dermatome.
- The patient had no relevant family history and was otherwise healthy.
- She was admitted to the Neurology inpatient service for further evaluation and management.

Results

- Initial examination showed symmetric, miotic, but reactive pupils bilaterally and profound bilateral ptosis with near-complete ophthalmoparesis.
- Enhanced MRI brain, orbits, and cervicothoracic spine revealed a large periaqueductal ependymal lesion extending from the midbrain to the cerebellopontine angle, as well as a marbled lesion in the corpus callosum.
- A working diagnosis of NMOSD was made, and the patient was treated with high-dose intravenous methylprednisolone and plasmapheresis.
- AQP-4 antibody testing came back positive at a high titer, confirming the diagnosis of NMOSD.
- The patient had a moderate response to initial treatment and started on rituximab infusions for maintenance therapy.
- At the 2-month follow-up appointment, the patient showed significant improvement in her ocular symptoms, with resolution of ptosis and only mild binocular diplopia in certain directions of gaze.

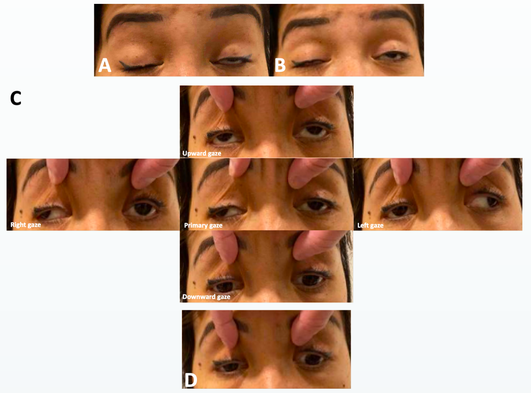

Figure 1: Eye movements at presentation to hospital A. eyes open at rest, B. eyes open voluntarily, C. primary gaze and cardinal eye positions, D. convergence

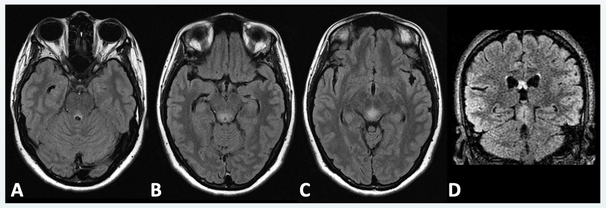

Figure 2: T2 FLAIR MR brain showing a periaqueductal hyperintense lesion spanning from pons to the midbrain, axial (A-C) and coronal (D)

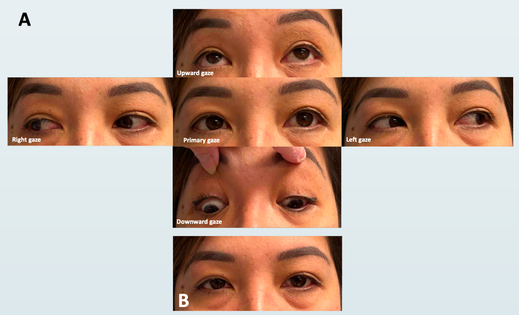

Figure 3: Eye movements after plasmapheresis A. primary gaze and cardinal eye positions, B. convergence.

Conclusion

1. NMOSD can present with atypical brainstem syndromes, making localization and differential diagnosis challenging.
2. Periaqueductal lesions on MRI have a high association with NMOSD and can aid in diagnosis.
3. AQP-4 antibody testing is highly sensitive and specific and can confirm the diagnosis of NMOSD in atypical cases.
4. Delay in diagnosis and treatment of NMOSD can result in greater fixed disability.
5. Healthcare providers should recognize the full range of NMOSD phenotypes to ensure appropriate investigations and rapid treatment.

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