Challenges of Diagnosis for Seronegative Postrema Area Syndrome and Long Myelitis

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Background:

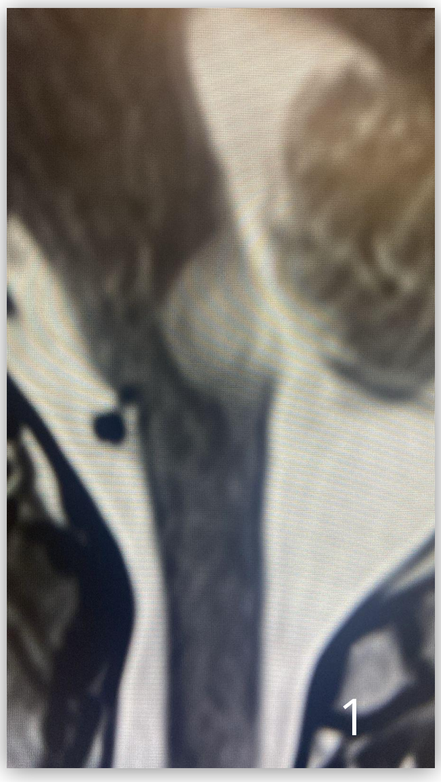
Neuromyelitis optica spectrum disorder (NMOSD) is a rare autoimmune disease often associated with anti-aquaporin-4 antibodies (AQP4-IgG). Diagnosing seronegative NMOSD is particularly challenging due to overlapping clinical and radiological features with other autoimmune, infectious, and neoplastic conditions.

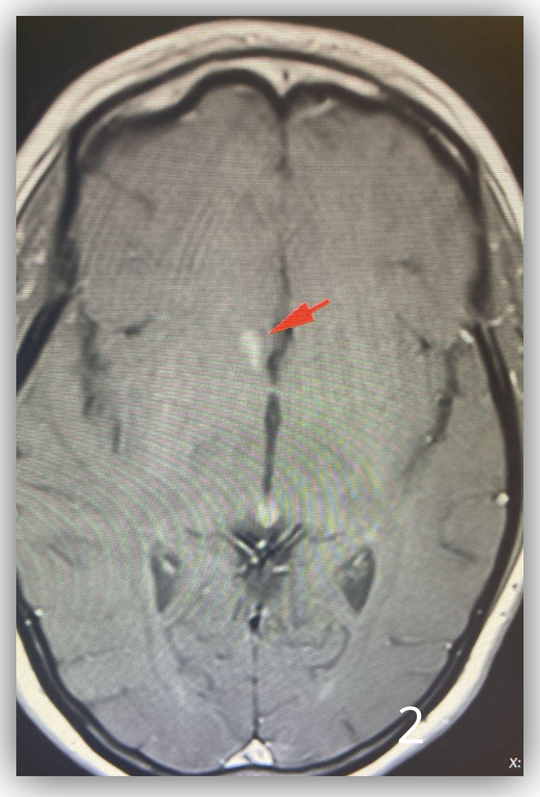
Case Description:

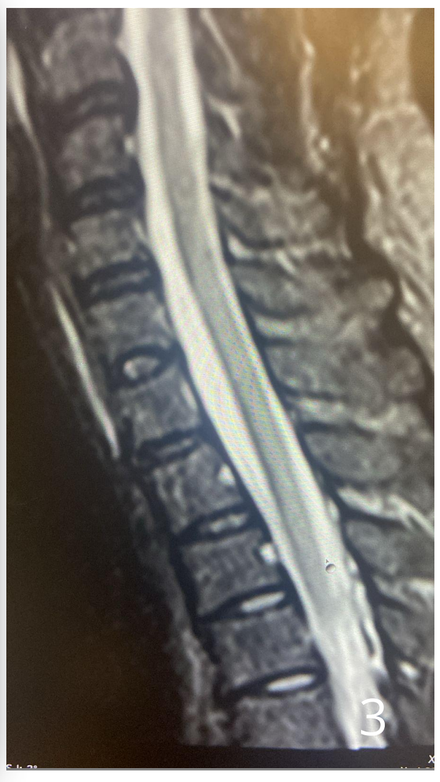
A 37-year-old woman with type I diabetes, a history of dermatological pseudofolliculitis, dental caries in 2022, and periodontitis presented two years ago with tingling in her feet, progressing to difficulty running and climbing stairs. By late October 2024, she reported severe dysphagia, hypophonia, instability requiring bilateral hand support to walk, urinary urgency, and anal spasms. Neurological examination revealed hyperreflexia with bilateral Babinski sign, hypoesthesia, severely diminished proprioception in the lower limbs, an ataxic gait, and dysmetria in the right upper limb.

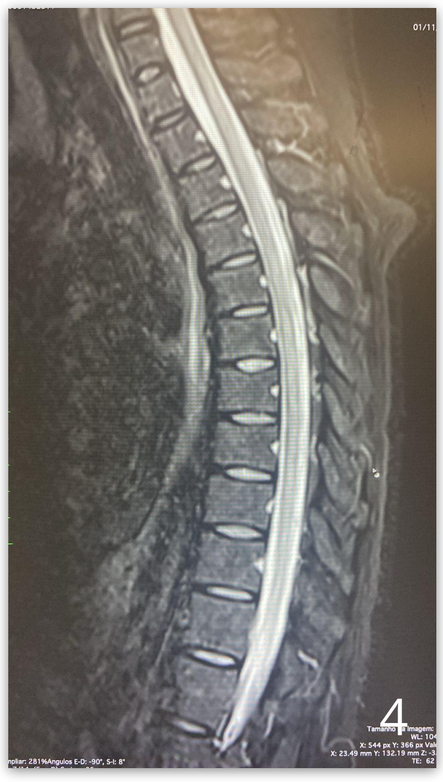
She also reported a localized livedo episode after sun exposure in June 2024 and rarely experienced oral aphthae. Initial MRI showed extensive T2 and STIR hyperintensity diffusely distributed across the cervical and thoracic spine, with gadolinium-enhanced lesions and postrema area enhanced lesion. Multiple tests for AQP4-IgG and MOG-IgG were negative, along with extensive autoimmune and paraneoplastic panels including HLA-B51, CRMP5, GFAP, NMDA, anti-Jo, anti-La, GAD, ANA and amphiphysin antibodies.

The patient received methylprednisolone pulse therapy (1 g/day for five days), followed by plasma exchange and cyclophosphamide. Despite aggressive treatment, follow-up MRI showed persistent contrast enhancement, indicating ongoing inflammatory activity despite clinical improvement.









1 -Lesion in Area Postrema
2 -Gadolinium-enhancing Periventricular Lesion
3- Longitudinal Extensive Hyperintensity in Cervical Cord (FLAIR/STIR)
4 - Longitudinal Extensive Hyperintensity in Dorsal Cord (FLAIR/STIR)

Discussion:

This case highlights the diagnostic complexities of seronegative NMOSD, particularly when mimics must be excluded through extensive testing. Persistent enhancement on follow-up imaging underscores the importance of close monitoring and optimization of immunosuppressive strategies.

Conclusion:

Seronegative NMOSD remains a diagnostic challenge, requiring a systematic and multidisciplinary approach for timely diagnosis and management. Continued research into seronegative cases is essential to improve outcomes for this subgroup.