Neuromyelitis optica spectrum disorder: a challenge in daily practice

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Introduction

The article describes the case of a 31-year-old woman, presenting with occipital headache, paraesthesia of the lower limbs with progressive motor deficits, sensory loss and retention-like sphincter disorders. Although these clinical features were investigated in a number of hospitals, no proper diagnosis was established. Once a diagnosis of neuromyelitis optica spectrum disorder was confirmed, the treatment protocol was initiated and a clear improvement was noticed. The patient was discharged with a follow-up appointment in 1 month and a recommendation of continued immunosuppressive therapy and physical rehabilitation.

Discussion

Neuromyelitis optica spectrum disorder (previously known as 'Devic's disease') is a rare autoimmune astrocytopathy, characterised by immune-mediated episodes of demyelination involving the optic nerves or/and spinal cord, more frequently diagnosed in Asian people and often occurring concurrently with systemic or organ-specific autoimmune disorders (Alvarenga et al, 2021).

Case report

A 31-year-old woman, with a recent history of autoimmune thyroiditis, was referred to hospital after 10 days of occipital headache, paraesthesia of the lower limbs with progressive motor deficits, sensory loss and retention-like sphincter disorders. Over the previous 2 weeks, the patient had presented at various hospitals for lower limb weakness, without a cause being identified. Cerebral and thoracic computed tomography scans were within normal limits, as well as a complete blood count and serum chemistry. A lumbar puncture was performed showing hypotensive, clear fluid, with a high protein content. CSF was collected for polymerase chain reaction and cultures, and full bacterial, viral and parasitic screenings were performed, all with negative results. Systemic corticosteroid therapy was initiated, but the paraplegia persisted with hypoaesthesia evident up to T6 of the spinal cord, associated with postural tremor in the upper limbs.

Cerebral and spinal magnetic resonance examinations with intravenous radiocontrast agent were performed, showing extensive inflammatory lesions of the spinal cord extending from the medulla to the T10 level, an appearance compatible with acute myelitis (**Figures 1** and **2**). The thyroid-stimulating hormone level was within normal range. An immunological profile was made, with negative results for anti-angiotensin-converting enzyme antibodies, antinuclear antibodies, anti-Ro/anti-SSA (anti-Sjögren's-syndrome-related antigen A autoantibodies) and anti-La antibodies/anti-SSB (autoantibodies directed against the La/SS-B protein). In addition, the patient was tested for neuromyelitis optica spectrum disorder, with the result (obtained after 2 weeks) positive for aquaporin-4 antibodies (AQP4), with a titre of 1/100 (the reference interval was 1/10), confirming the diagnosis.

The patient showed an incomplete response to systemic corticosteroid therapy and insufficient clinical improvement, so was admitted to the intensive care unit for therapeutic plasma exchange. Every other day she underwent five cycles, with 1–1.5 plasma volumes (30–40 ml/kg of plasma) exchanged. In face of persistent retention-like sphincter disorder, she developed a lower urinary tract infection, needing specific antibiotic therapy. The postural tremor of the upper limbs disappeared, sensitivity disorders improved and minimal mobility in the toes of the lower limbs was obtained, so the patient started intravenous immunoglobulin treatment, with 2 g/kg (total of 100 g) given over 5 days. Over the following days, the bladder dysfunction and hypoaesthesia improved, with sequelae affecting only the lower limbs. Rituximab was prescribed to prevent relapses. She was discharged home, with a follow-up appointment in 1 month and referral to a physical recovery centre.

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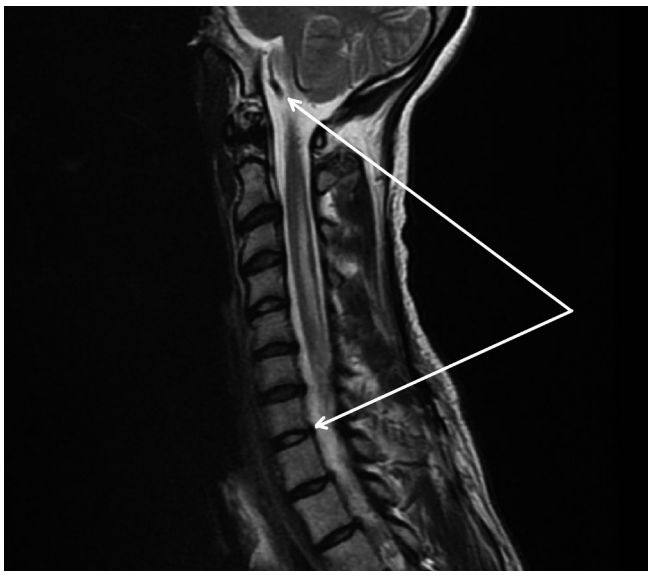

Figure 1. Sagittal spinal cord magnetic resonance imaging: T2-hyperintense lesion extending from the medulla throughout the cervical spinal cord (white arrows) to the T10 level.

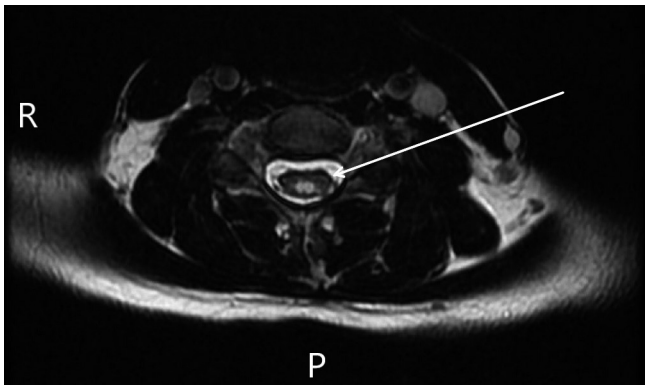

Figure 2. Axial spinal cord magnetic resonance imaging: typical involvement – T2-weighted image revealing a bright spotty lesion (white arrow).

The prevalence varies from 0.05 to 4.4/100 000, with a female predominance, in the third or fourth decade of life (Asgari et al, 2011; Bruscolini et al, 2018), similar to the case discussed in this article.

The presence of serum-specific antibodies (immunoglobulin G) targeting aquaporin-4 antibodies and the pathognomonic magnetic resonance imaging features are used to detect and classify neuromyelitis optica spectrum disorder (Kim et al, 2017; Bruscolini et al, 2018). This also allows it to be differentiated from multiple sclerosis, the principal differential diagnosis (Alvarenga et al, 2021). After excluding all possible differential diagnoses, diagnostic criteria include specific clinical elements such as optic neuritis, acute myelitis with longitudinally extensive transverse myelitis lesions, acute brainstem and area postrema syndrome, and neuromyelitis optica spectrum disorder-typical brain and diencephalic lesions (Wingerchuk et al, 2015).

The main differential diagnoses include amyotrophic lateral sclerosis, acute disseminated encephalomyelitis, primary lateral sclerosis, poliomyelitis, transverse myelitis, Sjögren's syndrome, systemic lupus erythematosus, Behçet's disease, sarcoidosis, intrathecal tumours, infections and exposure to toxins (Huda et al, 2019).

Corticosteroids are used as the first-line treatment in acute cases, with the aim of restoring neurological function. If, after 5 days, the symptoms do not significantly improve, therapeutic plasma exchange should be considered (Kleiter and Gold, 2016; Moore and Tanner, 2019). In cases with severe spinal inflammation, as seen in this case, and where deficits persist after therapeutic plasma exchange, human immunoglobulins are used as second line treatment (Wu et al, 2019).

Learning points

- Specific clinical elements, anti-aquaporin-4 immunoglobulin G status and magnetic resonance imaging-specific lesions define neuromyelitis optica spectrum disorder and differentiate it from multiple sclerosis.
- Preventing relapses is the key to prevent the appearance of disabling sequelae.
- In patients with bladder dysfunction, there is a high risk of lower urinary tract infection.

For this patient, rituximab was chosen as long-term immunosuppressive therapy, as this is suitable for preventing relapses in patients with severe attacks or incomplete response to other therapies (Kim et al, 2011; Pellkofer et al, 2011).

The patient also presented with retention-like sphincter disorders and a lower urinary tract infection. In neuromyelitis optica spectrum disorder with severe transverse myelitis, lower urinary tract symptoms can be expected (in 80% of the cases), as these patients present a progressive degradation of the functional abilities (Declemy et al, 2018). A nephrological and urological evaluation is recommended when the general assessment is performed.

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References

Alvarenga MP, do Carmo LF, Vasconcelos CCF et al. Neuromyelitis optica is an HLA associated disease different from multiple sclerosis: a systematic review with meta-analysis. Sci Rep. 2021;11(1):152. https://doi.org/10.1038/s41598-020-80535-3

Asgari N, Lillevang ST, Skejoe HP et al. A population-based study of neuromyelitis optica in Caucasians. Neurology. 2011;76(18):1589–1595. https://doi.org/10.1212/WNL.0b013e3182190f74

Bruscolini A, Sacchetti M, La Cava M et al. Diagnosis and management of neuromyelitis optica spectrum disorders - an update. Autoimmun Rev. 2018;17(3):195–200. https://doi.org/10.1016/j.autrev.2018.01.001

Declemy A, Chesnel C, Charlanes A et al. Specificity of lower urinary tract symptoms in neuromyelitis optica in comparison with multiple sclerosis patients. Int Neurourol J. 2018;22(3):185–191. https://doi.org/10.5213/inj.1836050.025

Huda S, Whittam D, Bhojak M et al. Neuromyelitis optica spectrum disorders. Clin Med (London). 2019;19(2):169–176. https://doi.org/10.7861/clinmedicine.19-2-169

Kleiter I, Gold R. Present and future therapies in neuromyelitis optica spectrum disorders. Neurotherapeutics. 2016;13(1):70–83. https://doi.org/10.1007/s13311-015-0400-8

Kim S-H, Kim W, Li XF, Jung I-J, Kim HJ. Repeated treatment with rituximab based on the assessment of peripheral circulating memory B cells in patients with relapsing neuromyelitis optica over 2 years. Arch Neurol. 2011;68(11):1412–1420. https://doi.org/10.1001/archneurol.2011.154

Kim S-M, Kim S-J, Lee HJ et al. Differential diagnosis of neuromyelitis optica spectrum disorders. Ther Adv Neurol Disord. 2017;10(7):265–289. https://doi.org/10.1177/1756285617709723

Moore RR, Tanner M. The latest diagnostic criteria and treatment options for neuromyelitis optica. JAAPA. 2019;32(7):1–6. https://doi.org/10.1097/01.JAA.0000558352.01234.c0

Pellkofer HL, Krumbholz M, Berthele A et al. Long-term follow-up of patients with neuromyelitis optica after repeated therapy with rituximab. Neurology. 2011;76(15):1310–1315. https//doi.org/10.1212/WNL.0b013e3182152881

Wingerchuk DM, Banwell B, Bennett JL et al. International consensus diagnostic criteria for neuromyelitis optica spectrum disorders. Neurology. 2015;85(2):177–189. https://doi.org/10.1212/WNL.0000000000001729

Wu Y, Zhong L, Geng J. Neuromyelitis optica spectrum disorder: pathogenesis, treatment, and experimental models. Mult Scler Relat Disord. 2019;27:412–418. https//doi.org/10.1016/j.msard.2018.12.002