Multiple sclerosis with pseudotumoral demyelinating lesions in a female adolescent presenting with an optic neuritis

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DESCRIPTION

A previously healthy 13-year-old female adolescent presented with right retrobulbar eye pain, worse with vertical eye movements, followed by decreased visual acuity, evolving for 4 days. She complained of fatigue in the previous month and she reported no similar previous episodes. On admission, she had mild visual loss (0.8) in the right eye with a relative afferent pupillary defect, blurring of optic margins of the right papilla and a right central scotoma. She showed no dyschromatopsia and no other changes in the neurological examination. The brain MRI showed two tumefactive demyelinating lesions with an open-ring enhancement after gadolinium administration (figure 1) and additional typical demyelinating lesions (disseminated in space) without enhancement. Cerebrospinal fluid analysis showed no changes, oligoclonal banding was negative and anti-AQP4 and anti-MOG antibodies were negative. These findings were consistent with multiple sclerosis (MS) with pseudotumoral demyelinating lesions (MS-PL) and she completed 5 days of intravenous methylprednisolone, with full visual recovery. At the follow-up consultation, she started

Learning points

- Multiple sclerosis with pseudotumoral demyelinating lesions (MS-PL) is a rare variant of the disease and it can represent a diagnostic problem.
- MS-PL may present with clinical features disproportionate to the lesion dimensions and may show a good prognosis despite the great size of lesions.

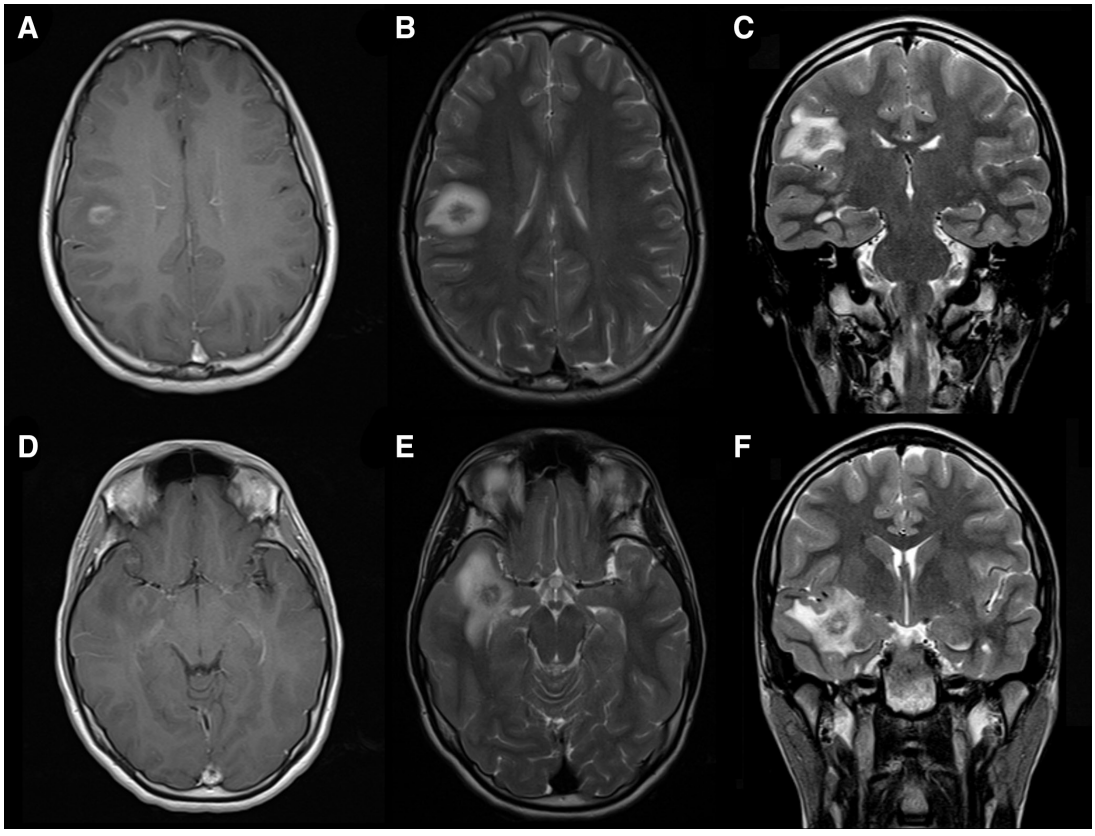

Figure 1 Tumefactive demyelinating lesions: A–C A large subcortical lesion in the posterior-inferior area of the right frontal lobe and D–F A large lesion in the anterior and medial area of the right temporal lobe, with extension to the insular and opercular regions. A,D Axial T1-weighted images after gadolinium injection, with the typical incomplete ring enhancement pattern. B,E Axial T2-weighted images. C,F Coronal T2-weighted images.

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intravenous natalizumab. She showed regression of previous lesions and no new injuries in subsequent MRI at 6, 12, 18 and 36 months. Three years after diagnosis, she is clinically stable (Expanded Disability Status Scale: 1.5), with no new relapses or complaints related to treatment.

MS, defined by recurrent episodes of demyelination in the central nervous system (CNS), disseminated in space and time, is the most prevalent chronic inflammatory disease of the CNS. In children, it has a wide range of clinical manifestations, including optic neuritis, or a range of focal or multifocal neurological symptoms. In this case, although the clinical presentation was limited to ocular symptoms, brain MRI showed large demyelinating lesions. Typical MS lesions identified in MRI are usually smaller than 0.6 cm.[1] Rarely, some patients present with larger demyelinating lesions at their first event. When larger than 2 cm, these lesions are considered tumefactive or pseudotumoral.[2–4] Although rare (incidence of 3:1 000 000/year), MS-PL is more common in women in their second and third decades of life and is usually manifested by multifocal symptoms such as persistent headache, impaired consciousness, focal signs or cognitive deficits.[2 5] Surprisingly, in our case, it occurred with few neurological deficits disproportionate to the lesion dimensions. MS-PL may pose diagnostic challenges: solitary lesions may be mistaken for primary tumours, abscess, metastasis or infarction, while multiple pseudotumoral lesions may suggest acute disseminated encephalomyelitis.[2–9] In our case, the presence of additional typical demyelinating lesions fulfilling McDonald's criteria for dissemination in time and space assisted in diagnosing MS.

MS-PL is associated with higher hospitalisation rates and morbidity than conventional MS.[10] Due to its significant inflammatory component, it may respond to corticosteroids, as shown in our case, with a reduction of the size of the lesions.[9] Although its long-term evolution is still unclear, the complete disappearance of the lesions is unusual and residual deficits are frequent.[10] Recurrence of pseudotumoral lesions, although uncommon, may occur.[6] The present case adds up to the few cases that have been described in the literature and may contribute to further understand the clinical picture of MS-PL in children and adolescents.

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