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Case Report

Two Unusual Presentations of MOGAD Mimicking Pseudotumor Cerebri in Children

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Abstract Optic neuritis (ON) is a common clinical manifestation of myelin oligodendrocyte glycoprotein (MOG) antibody-associated disorders (MOGAD), while myelitis is less often manifestation in children. Other clinical manifestations include acute demyelinating encephalomyelitis and neuromyelitis optica spectrum disorder (NMOSD). Coexistence of brain lesion and myelitis is an uncommon presentation of MOGAD and there have been no cases reported in children. In MOGAD, ON typically involves the anterior portion of both optic nerves, leading to bilateral disc swelling. During the early stages of ON, in the setting of bilateral disc swelling and pain, patients may initially be suspected as having pseudotumor cerebri (PTC). In this study, we report two cases who were admitted to the emergency department with early symptoms of overlapping ON and myelitis in the course of MOG-IgG-related disorders and were misdiagnosed as having PTC. MOG-IgG-positive ON requires timely treatment to prevent retinal nerve fiber layer (RNFL) and vision loss due to the high relapse rate associated with these antibodies. Our aim was to increase the awareness of unique findings of MOG-IgG-positive ON and myelitis, which may initially mimic PTC very high cerebrospinal fluid opening pressure and thereby delay treatment in children.

Key words Children; Mimicking; Myelin oligodendrocyte glycoprotein (MOG) antibody-associated disorders (MOGAD); Pseudotumor cerebri (PTC)

Introduction

Myelin oligodendrocyte glycoprotein (MOG) antibody-associated disorders (MOGAD) describe a new entity of demyelinating neurological syndromes defined by the presence of serum IgG autoantibodies against MOG detected by cell-based assays.[1-3] MOGAD occur in children and comprise a heterogeneous disease spectrum. The latest proposed diagnostic criteria for MOGAD include[1] a presentation of acute disseminated encephalomyelitis (ADEM), optic neuritis (ON), myelitis, cerebral focal deficit, brainstem syndrome, cerebellar presentations or cortical encephalitis often with seizures,[2] positive serum MOG-IgG test by cell-based assay,[3] and exclusion of a better diagnosis.[4]

In this report, we describe two paediatric cases of MOGAD with confounding initial presentations and highlight their diagnostic challenges which may initially mimic pseudotumor cerebri (PTC).

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Case 1

A 10-year-old obese boy (body mass index [BMI] 36 kg/m^2) presented with there was no illness before suddenly there was a bilateral blurred vision and

headaches. Vision was as low as 20/70 and optic discs were bilaterally swollen. Color vision was normal with no relative afferent pupillary defect (RAPD). Visual fields demonstrated a nasal step defect in both eyes. Average peripapillary retinal nerve fiber layer (RNFL) on Optical Coherence Tomography (OCT) was significantly thickened in both eyes (RE 135 µm, LE 152 µm). Brain tomography (CT) and CT-venography were normal. Cerebrospinal fluid opening pressure was 117 cm H_2O with three white blood cells and normal protein, glucose levels and cerebrospinal fluid (CSF) culture. The patient was initially treated with oral acetazolamide with a working diagnosis of PTC, but he returned within a few days due to progressive deterioration of vision. On magnetic resonance imaging (MRI) of the optic nerve, coronal short tau inversion recovery (STIR) showed high signal intensity of the left perioptic nerve sheath (arrow) and coronal contrast-enhanced fat-saturated T1-weighted image revealed enhancement of left perioptic nerve sheath and surrounding orbital fat (arrow) (Figure 1), which rapidly pulse steroid treatment was given for 5 days responded to intravenous (IV) methylprednisolone. AQP4-IgG antibodies were negative, while serum IgG autoantibodies against MOG detected by cell-based assays and titer 1/360 (Figure 1). At 3-month follow-up, the patient was in remission, with intact visual acuity and normal fundus and MRI examination and titer 1/240 so there were no findings on control imaging and one year later, he was still in remission and no detect in serum IgG autoantibodies against MOG detected by cell-based assays.

Case 2

An 11-year-old girl (BMI 18 kg/m^2) presented to the emergency department with severe right frontal headache and pain radiating to the neck and back with healthy development before, without vision problems. Initial evaluation revealed normal neurological, fundoscopic, and head CT examinations. Three days later, the headaches worsened, blurred vision developed in the left eye, and funduscopy revealed bilaterally swollen discs. CSF opening pressure was 70 cm H_2O with eight white blood cells and normal protein and glucose levels and culture. She was treated with acetazolamide for presumed PTC until an MRI was performed. The MRI examination was normal. One week later, her headache immediately improved following the lumber puncture and the patient had no focal motor or sensory deficits, but bladder and bowel dysfunction, or impaired coordination. On cerebral MRI, axial FLAIR image showed bilateral areas of high signal intensity in subcortical and deep white matter (arrows) and axial FLAIR images revealed areas of high signal intensity in the pons (arrows) (Figure 2). STIR images showed hyperintense lesions in two segment the cervical spinal cord and in two segment the conus medullaris. MRI of the optic nerve demonstrated no contrast enhancement. Laboratory tests including serum and CSF paediatric autoimmune panels were performed and treatment with methylprednisolone resulted in visual acuity was 20/70 in the first application, it was 20/20 on the three day of treatment. Subsequently, the treatment was continued with oral prednisone followed by a slow taper. Serology for AQP4-IgG was negative, while autoantibodies against MOG detected by cell-based assays and titer 1/840. The patient was diagnosed as having ON and myelitis. The symptoms were resolved with the administration of IV methylprednisolone 1000 mg daily for 5 days. The patient was discharged from the hospital on corticosteroid taper. At 6-month follow-up, she was in remission, with intact visual acuity and normal fundus and servico-thoracolumbar MRI examination. One year later, the patient was still in remission and no autoantibodies against MOG detected by cell-based assays

Discussion

Clinical manifestations of MOGAD vary in part by location of nervous system involvement, typically including vision impairment, cranial nerve deficits, focal motor or sensory deficits, bladder or bowel dysfunction, and/or impaired coordination.[1,2]

Both patients had CSF opening pressure measured and, for the first time in the literature, had extremely high CSF pressure documented in children. Accordingly, we consider that Intracranial hypertension (ICH) may have a similar pathophysiologic mechanism to that of MOGAD. Nevertheless, both patients showed good tolerance. In transgenic mice, a deficiency of AQP-4 will lead to a PTCS-like pathology, however, no studies have been able to translate these findings to the human PTCS presentation.[5,6] Other studies with AQP-1 have found that acetazolamide will lower its expression, while steroids and retinoids will increase the expression[7] Accordingly, different markers such as AQp may be useful in the differential diagnosis. Research has also begun into the

aquaporins (AQP).[5,7]

Clinical symptoms of ON include blurred vision and reduced visual acuity or visual loss as well as eye pain, particularly retrobulbar pain with eye movement. Longitudinal extensive transverse myelitis (LETM), defined as a spinal cord lesion spanning three or more vertebral segments in length, is a characteristic finding in MOGAD.[3] Typical symptoms of LETM include motor and/or sensory deficits (numbness), bladder, bowel and erectile function in males.[3] Myelitis in MOG-IgG-related disease most frequently occurs as the initial manifestation and may occur in isolation or concurrently with ON and/or cerebral involvement with a neuromyelitis optica spectrum disorder (NMOSD)/ADEM-phenotype, and such presentations should prompt consideration of MOG-IgG testing.[7-9] Similar to prior studies, we detected urinary retention/incontinence and erectile dysfunction in both patients, which typically accompany myelitis, fitting with

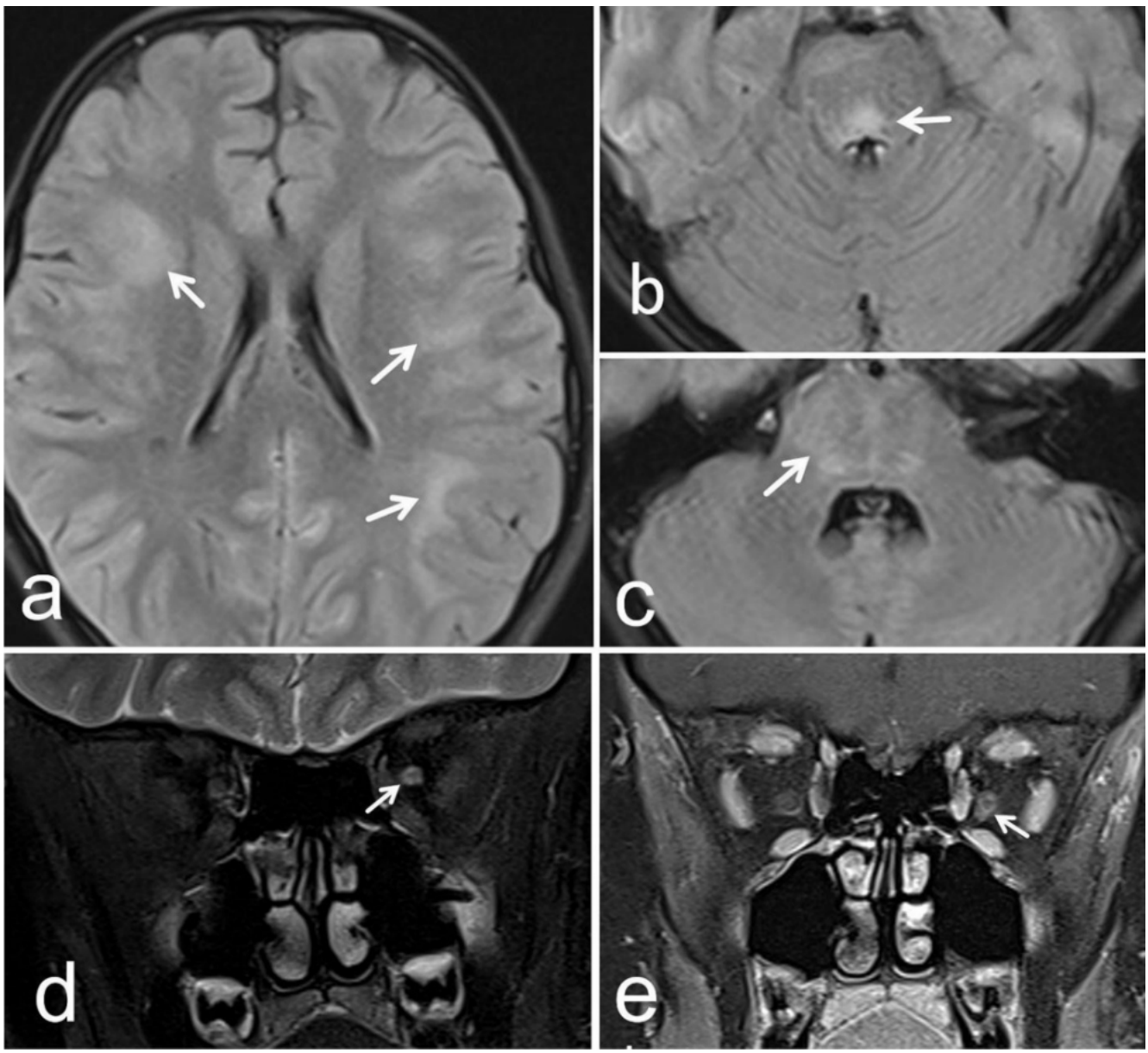

Figure 1 Axial FLAIR MR image (a) shows bilateral areas of high signal intensity (arrows) in subcortical and deep white matter and axial FLAIR MR images (b, c) reveal areas of high signal intensity in pons (arrows). Coronal short tau inversion recovery (STIR) image (d) shows high signal intensity of left optic nerve sheath (arrow) and coronal contrast-enhanced fat-saturated T1-weighted image (e) reveals enhancement of left optic nerve sheath and surrounding orbital fat (arrow).

conus involvement radiologically.[9] Both of our patients presented to the emergency department with headaches and bilateral disc swelling, which were considered initial clinical manifestations of bilateral ON and thus led to a presumed diagnosis of PTC and initiation of acetazolamide treatment. Diagnosis of bilateral anterior ON was made based on MRI of the optic nerve and perioptic enhancement. Despite the fact that similarities in the clinical spectrum of demyelinating diseases can cause difficulties in differential diagnosis, it is important that patients with the symptoms of PTC have differences in the course, since they typically present with progressive spinal findings. To date, there have been few reports of MOGAD cases presenting with isolated ICH.[10] Significantly, the

Figure 2 Sagittal short tau inversion recovery (STIR) images show hyperintense lesion in (a) cervical spinal cord and (b) conus medullaris.

clinical presentation of very high CSF pressure with the continuous progression of spinal involvement in our patient is an asset of great importance. Spinal involvement is a rare phenomenon detected in the follow-up of PTC-mimicking cases. Nonetheless, although spinal involvement of the long segment is a definite diagnostic parameter for the NMOSD spectrum, it has a different topographic character specific to MOGAD. During childhood, MOGAD can come with many different clinics. There may be spinal and cerebral lesions that may occur in two different clinics accompanied by high CSF pressure, one of which may be perineural involvement and progressive rotation without imaging findings at the first admission. Before receiving the diagnosis of PTC, the differential diagnosis of MOGAD should definitely be kept in mind and the gold standart autoantibodies against MOG detected by cell-based assays should be sent to these signs. The overlap of high CSF pressure and favorable clinical parameters at the time of admission and perioptic involvement in imaging should be stimulating for both ON and spinal involvement in PTC-mimicking patients.

Acknowledgement / Disclaimers / Conflict of Interest

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Contest to Participate

Written informed consent was obtained from the parents of the patient reported in this study.

Consent for Publication

The parents of the patient reported in this study have consented to the submission of the case report to the journal and signed informed consent regarding publishing their data and photographs.

Conflict of Interest

The authors declare no competing interests.

References

1. Jarius S, Ruprecht K, Kleiter I, et al. MOG-IgG in NMO and related disorders: A multicenter study of 50 patients. part 2: Epidemiology, clinical presentation, radiological and laboratory features, treatment responses, and long-term outcome. J Neuroinflammation 2016;13:280.
2. Cobo-Calvo A, Ruiz A, Rollot F, et al. Clinical features and risk of relapse in children and adults with myelin oligodendrocyte glycoprotein antibody-associated disease. Ann Neurol 2021;89:30-41.
3. Dubey D, Pittock SJ, Krecke KN, et al. Clinical, radiologic, and prognostic features of myelitis associated with myelin oligodendrocyte glycoprotein autoantibody. JAMA Neurol 2019;76:301-9.
4. Banwell B, Bennett JL, Marignier R, et al. Diagnosis of myelin oligodendrocyte glycoprotein antibody-associated disease: International MOGAD Panel proposed criteria. Lancet Neurol 2023;22:268-82.
5. Mollan S, Ali F, Hassan-Smith G, Botfield H, Friedman D, Sinclair A. Evolving evidence in adult idiopathic intracranial hypertension: pathophysiology and management. J Neurol Neurosurg Psychiatry 2016;87:982-92.
6. Portelli M, Papageorgiou P. An update on idiopathic intracranial hypertension. Acta Neurochir (Wien) 2016;159:491-9.
7. Ameli PA, Madan M, Chigurupati S, Yu A, Chan SL, Pattisapu JV. Effect of acetazolamide on aquaporin-1 and fluid flow in cultured choroid plexus. Acta Neurochir Suppl 2012:113:59-64.
8. Jurynczyk M, Messina S, Woodhall MR, et al.. Clinical presentation and prognosis in MOG-antibody disease: a UK study. Brain 2017;140:3128-38.
9. Sinha S, Banwell B, Tucker A, Storm PB, Huh J, Lang SS. Hemicraniectomy and externalized ventricular drain placement in a pediatric patient with myelin oligodendrocyte glycoprotein-associated tumefactive demyelinating disease. Childs Nerv Syst 2022;38:185-9.
10. Lotan I, Brody J, Hellmann MA, et al. Myelin oligodendrocyte glycoprotein-positive optic neuritis masquerading as pseudotumor cerebri at presentation. J Neurol 2018;265:1985-8.