When squint and proptosis signal more: A case of probable paediatric myelin oligodendrocyte glycoprotein antibody-associated disease presenting as optic neuritis.

Ruhaya Razali[1], Nor Amalina Abdul Rahman[1], Lim Thiam Hou[1], Siti Norzalehawati Sepian[1], Norshamsiah Md Din[2]

[1]Hospital Sultanah Nur Zahirah, Terengganu, Malaysia
[2]Ophthalmology Department, Hospital Cancelor Tuanku Muhriz, Universiti Kebangsaan Malaysian, MYS, Malaysia

Abstract

Background: Myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) is an inflammatory demyelinating disorder of the central nervous system affecting individuals of all age groups.[1,2] Although optic neuritis, acute disseminated encephalomyelitis, and transverse myelitis are typical presentations, paediatric patients demonstrate a distinct clinical phenotype with age-related variation in disease expression.[2] Orbital manifestations are uncommon and may lead to diagnostic uncertainty.

Case Presentation: A previously healthy 4-year-old boy presented with acute-onset outward deviation and apparent proptosis of the right eye following a recent episode of pneumonia. Visual acuity was 20/600 in the right eye and 20/40 in the left eye, with a right relative afferent pupillary defect and marked right optic disc swelling with perivascular sheathing. Initial evaluation focused on excluding orbital cellulitis, inflammatory orbital disease, orbital tumour, and orbital apex syndrome. Computed tomography of the orbit and brain was performed to exclude an orbital mass or infectious orbital pathology and demonstrated enlargement of the right optic nerve. Subsequent magnetic resonance imaging revealed optic nerve enhancement consistent with optic neuritis, along with large, poorly defined T2/FLAIR hyperintense lesions involving the bilateral frontal and parietal white matter and the hemicerebellum, suggestive of an inflammatory demyelinating process. Aquaporin-4 antibody was negative, while serum MOG-IgG was reported as borderline positive with a reference cut-off of 1:10 using indirect immunofluorescence antibody testing. However, the quantitative titre and specific criteria for borderline interpretation were unavailable. The patient was treated with intravenous methylprednisolone followed by intravenous immunoglobulin because of an inadequate initial response. Significant clinical improvement was observed at the three-week follow-up, with visual acuity improving to 20/40 in the right eye and resolution of ocular deviation, relative afferent pupillary defect, and optic disc swelling. The patient completed immunotherapy and remained free from ocular relapse or neurological deterioration during two years of follow-up.

Conclusion: This case highlights that optic neuritis associated with MOGAD may present with atypical orbital features, including proptosis and ocular deviation, mimicking primary orbital disease. Early recognition of inflammatory demyelinating optic neuropathy and appropriate neuroimaging are essential for timely diagnosis and treatment. Although definitive serological confirmation was limited by borderline MOG-IgG positivity, the clinical-radiological features and favourable response to immunotherapy supported a diagnosis of probable MOGAD.

Keywords: Paediatric MOGAD, Optic neuritis, Squint, Proptosis, Demyelinating disease, Paediatric neuro-ophthalmology

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Correspondence to:
Ruhaya Razali, *Hospital Sultanah Nur Zahirah, Terengganu, Malaysia, Email: ruhayarazali00@gmail.com*
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Introduction

Myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) is a rare immune-mediated demyelinating disorder of the central nervous system associated with antibodies directed against myelin oligodendrocyte glycoprotein (MOG).[1,2] Although MOGAD can affect individuals of all ages, paediatric cases represent a distinct subgroup within the disease spectrum and constitute a smaller proportion of reported cohorts than adult populations.[1,2]

The clinical spectrum of paediatric MOGAD is heterogeneous and varies according to age, with acute disseminated encephalomyelitis (ADEM) being more commonly observed in younger children, while optic neuritis and transverse myelitis are more frequently reported in older children.[1-3] Other phenotypes, including relapsing optic neuritis and neuromyelitis optica spectrum disorder-like presentations, have also been described.

Optic neuritis is the most common onset feature of MOGAD, especially in adults, but is also frequent in children.[2-4] However, atypical presentations may overlap with orbital disorders. Previous reports have described ocular motility abnormalities associated with MOGAD-related optic neuritis, including cases presenting with ocular deviation or orbital apex syndrome-like features, although prominent proptosis has not been reported in these cases.[5-7] Optic nerve enlargement, optic disc swelling, and inflammatory changes may further contribute to diagnostic confusion with primary orbital pathology. To our knowledge, the coexistence of apparent proptosis and ocular deviation as presenting features of paediatric MOGAD-associated optic neuritis has rarely been described.

This case describes an unusual presentation of paediatric MOGAD-associated optic neuritis with proptosis and ocular deviation, initially mimicking orbital pathology.

Case Report

A previously healthy 4-year-old Malay boy presented with a one-week history of outward deviation of the right eye associated with apparent proptosis. The symptoms developed following an episode of pneumonia three weeks prior to presentation. There was no history of ocular pain, diplopia, fever, seizures, headache, vomiting, behavioural changes, focal neurological deficits or features suggestive of raised intracranial pressure.

He was born at term with an uneventful neonatal history. Developmental milestones were appropriate for his age. There was no history of recurrent infections, autoimmune disease, trauma, or thyroid-related symptoms. Immunisations were complete according to the national schedule. Family history was notable only for breast cancer in a maternal aunt, with no history of autoimmune disorders.

On ocular examination, best-corrected visual acuity was 20/600 in the right eye and 20/40 in the left eye. A relative afferent pupillary defect was present in the right eye, with mild right-sided proptosis. Ocular motility assessment revealed limitation of adduction with superior deviation of the right eye. Anterior segment examination was unremarkable bilaterally. Fundus examination of the right eye demonstrated marked optic disc swelling with elevated disc margins and perivascular sheathing, while the left eye showed mild optic disc hyperaemia without swelling.

Although the patient appeared lethargic on presentation, he remained haemodynamically stable with preserved oral intake.

Initial laboratory investigations showed a normal full blood picture with haemoglobin of 10.4 g/dL, a total white blood cell count of 6.7×10^9 /L with lymphocyte predominance (64%), and a platelet count of 454×10^9 /L. The erythrocyte sedimentation rate was elevated at 96 mm/hour. Renal and liver function tests, complement levels (C3 and C4), and autoimmune and infectious screening, including antinuclear antibody and serology for toxoplasmosis, rubella, herpes simplex virus, and syphilis, were all unremarkable. Tuberculosis screening and chest radiography were also negative.

Neuroimaging demonstrated right optic nerve enlargement and enhancement consistent with optic neuritis. Magnetic Resonance Imaging (MRI) T2/FLAIR showed large fluffy hyperintense white matter lesions involving the bilateral frontal and parietal white matter and the hemicerebellum, together with circumferential thickening and enhancement of the right optic nerve sheath, consistent with optic perineuritis. Computed tomography was performed initially to exclude an orbital mass, space-occupying lesions and infectious orbital

disease, followed by magnetic resonance imaging for further characterization.

Serological evaluation for autoimmune demyelinating disorders demonstrated a negative aquaporin-4 (AQP4) antibody result and a borderline positive myelin oligodendrocyte glycoprotein immunoglobulin G (MOG-IgG) result. The serum sample was collected during treatment with intravenous methylprednisolone. MOG-IgG testing was performed using an indirect immunofluorescence antibody (IFA) method, with a reported cut-off/reference value of 1:10. However, the available laboratory report did not provide the quantitative antibody titre or specific interpretation criteria for borderline positivity. Lumbar puncture was not performed due to parental refusal.

The patient was initially treated with intravenous methylprednisolone at a dose of 10 mg/kg every 8 hours for 5 days, in view of suspected inflammatory demyelinating optic neuritis. Empirical intravenous ceftriaxone was administered concurrently because of the initial concern for possible infectious orbital pathology.

Following completion of intravenous methylprednisolone, there was no significant improvement in visual acuity or reduction in optic disc swelling on repeat ophthalmological assessment. Therefore, intravenous immunoglobulin (IVIG) at 1 g/kg daily was commenced for 2 days as an escalation of immunotherapy. The patient was subsequently transitioned to oral prednisolone for tapering.

After initiation of intravenous immunoglobulin (IVIG), the patient demonstrated significant clinical improvement. At the three-week follow-up, visual acuity improved from 20/600 to 20/40 in the right eye and from 20/40 to 20/25 in the left eye. The right ocular deviation resolved completely, with marked reduction of optic disc swelling on fundoscopic examination and resolution of the right relative afferent pupillary defect.

The patient subsequently completed a three-month course of corticosteroid and immunosuppressive therapy. Two months after completion of treatment, he was admitted with prolonged fever and was diagnosed with bronchopneumonia. Despite this intercurrent infective episode, there was no evidence of ocular relapse or recurrence of optic neuritis. The patient remained clinically stable during two years of follow-up. Serial ophthalmological assessments demonstrated sustained visual improvement, with visual acuity maintained at 20/40 in the right eye, no recurrence of the relative afferent pupillary defect or optic disc swelling, and no further episodes of ocular or neurological relapse.



Figure 1: Right eye showing outward ocular deviation (exodeviation) at presentation.



Figure 2: Axial contrast-enhanced CT orbit showing unilateral right-sided proptosis

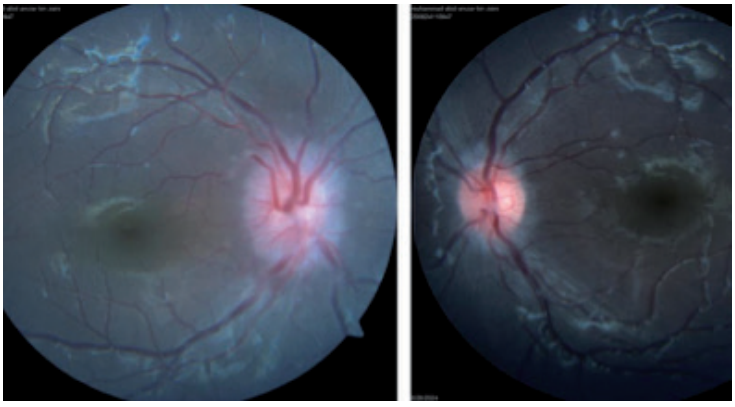


Figure 3: Fundus photo on initial presentation shows swollen right optic disc, perivascular sheathing of the retinal vessels and slight hyperemic left optic disc.

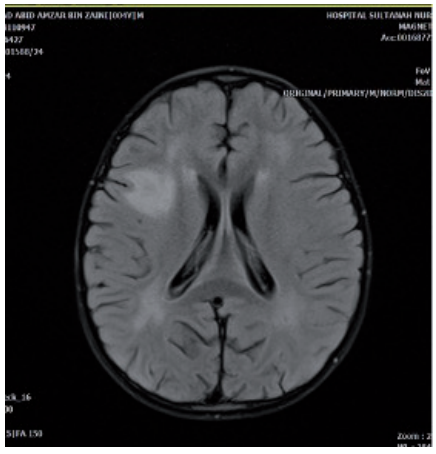


Figure 4: Axial T2-weighted fluid-attenuated inversion recovery (T2-FLAIR) magnetic resonance imaging (MRI) of the brain demonstrating a hyperintense lesion in the right frontal periventricular/deep white matter with mild surrounding edema

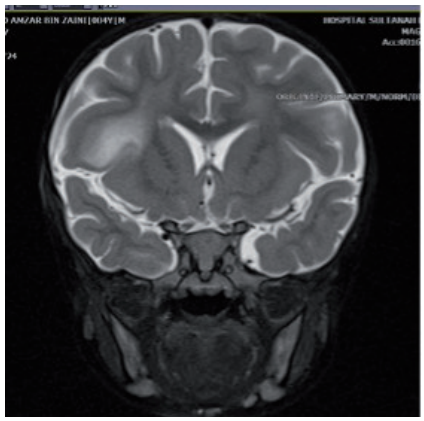


Figure 5: Coronal T2-weighted MRI demonstrating a large hyperintense lesion in the right frontal subcortical and deep white matter, consistent with an inflammatory demyelinating lesion.

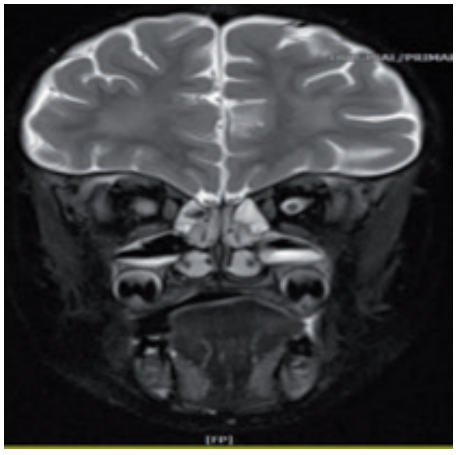


Figure 6: Coronal T2-weighted MRI orbit showing circumferential thickening with enhancement of the right optic nerve sheath (doughnut sign)



Figure 7: Fundus photo after three weeks of treatment

Discussion

Myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) is an inflammatory demyelinating disorder of the central nervous system with a broad clinical spectrum affecting both children and adults.[1,2] In paediatric patients, the most frequently recognised phenotypes include optic neuritis (ON), acute disseminated encephalomyelitis (ADEM), and transverse myelitis.[2-4,8,9] Large paediatric cohort studies have demonstrated age-related differences in disease presentation, with ADEM being more frequently observed in younger children, whereas optic neuritis becomes increasingly predominant with advancing age.[2-4,8,9] The International MOGAD Panel proposed criteria further established these core clinical phenotypes and emphasised the importance of integrating clinical presentation, MRI characteristics, and MOG-IgG testing for accurate diagnosis.[2]

Although optic neuritis is a common ophthalmic manifestation of paediatric MOGAD, orbital signs such as proptosis and ocular deviation are uncommon. Previous paediatric MOGAD cohorts have primarily described visual loss, relative afferent pupillary defect (RAPD), optic disc swelling, and optic nerve abnormalities on imaging as typical ophthalmic findings.[2-4,8,9] In contrast, our patient presented with concurrent proptosis and strabismus, initially raising suspicion of primary orbital pathology. This unusual presentation highlights the potential for MOGAD-associated optic neuritis to mimic orbital disease and expands the recognised ophthalmic spectrum of paediatric MOGAD.

Several isolated reports have described atypical ophthalmic presentations of MOGAD. Nurul-Ain et al. reported paediatric patients with MOGAD-associated optic neuritis presenting with optic disc oedema and ocular motility limitation, although proptosis was not observed.[5] Similarly, Sulaiman et al. described an orbital apex syndrome-like presentation characterised by severe visual loss and extraocular movement restriction without apparent proptosis.[6] Shah et al. further reported optic perineuritis associated with abducens nerve palsy during disease relapse, suggesting that inflammatory involvement beyond the optic nerve may occur in MOGAD.[7] While these reports provide insight into uncommon ophthalmic manifestations, prominent proptosis combined with ocular deviation at initial presentation remains an unusual feature, as demonstrated in our patient.

The diagnosis of MOGAD in this case was challenging because of the atypical orbital presentation and the borderline serum MOG-IgG result. According to the International MOGAD Panel proposed criteria, diagnosis requires a compatible clinical demyelinating event, positive MOG-IgG testing using an appropriate assay, and exclusion of alternative diagnoses.[2] In patients with borderline or low-positive MOG-IgG titres, MOGAD may still be diagnosed when clinical and MRI features are strongly supportive and at least one characteristic supporting feature is present.[2] In our patient, the diagnosis was supported by clinical evidence of optic neuritis, including optic disc swelling and RAPD, together with characteristic MRI findings demonstrating large, poorly demarcated ("fluffy") T2/FLAIR hyperintense lesions involving the bilateral frontal and parietal white matter and the hemicerebellum. These imaging findings are consistent with reported paediatric MOGAD patterns and were more suggestive of an acquired inflammatory demyelinating disorder than compressive, infectious, or neoplastic orbital disease.[1-4]

The absence of aquaporin-4 (AQP4) antibody positivity and the favourable response to immunotherapy further supported the diagnosis.[2] However, lumbar puncture was not performed due to parental refusal, representing a limitation of this report. Cerebrospinal fluid (CSF) examination, including cell count, protein concentration, and oligoclonal band analysis, may provide additional supportive information and assist in differentiating MOGAD from other acquired demyelinating disorders, particularly multiple sclerosis.[4] Furthermore, the MOG-IgG result in this case was reported as borderline positive using an indirect immunofluorescence assay, without an available quantitative titre or assay-specific interpretation criteria. Therefore, although the diagnosis was supported by the clinical phenotype, neuroimaging findings, exclusion of alternative diagnoses, and treatment response, this case is most appropriately classified as probable MOGAD.

Acute treatment of paediatric MOGAD aims to suppress inflammatory activity and achieve rapid neurological recovery.[10] According

to the E.U. Paediatric MOG Consortium consensus, intravenous methylprednisolone (IVMP) is recommended as first-line therapy for acute attacks and is associated with favourable outcomes in the majority of patients. Oral corticosteroid tapering for up to three months may be considered following acute treatment to maintain clinical improvement and reduce the risk of early disease activity.[10] Intravenous immunoglobulin (IVIG) and plasma exchange are considered escalation therapies for patients with severe disease or an inadequate response to corticosteroids.[10] In our patient, an incomplete early response following IVMP prompted the administration of IVIG, resulting in significant improvement in visual function, resolution of ocular deviation, and reduction of optic disc swelling. This favourable response supports the role of timely escalation therapy in achieving recovery in severe or atypical paediatric MOGAD presentations.

Long-term management of paediatric MOGAD remains challenging because the optimal duration of immunotherapy and predictors of relapse are not fully established. Previous studies suggest that children may have different relapse patterns from adults, with some cohorts demonstrating a lower risk of relapsing disease in paediatric patients.[10-11] Therefore, treatment decisions should balance relapse prevention against the potential adverse effects associated with prolonged immunosuppression. In our patient, a three-month course of corticosteroid and immunosuppressive therapy was completed, with no ocular or neurological relapse observed during two years of follow-up. Although the patient required hospitalisation for bronchopneumonia two months after completing therapy, there was no evidence of recurrent demyelinating disease, suggesting a favourable monophasic course.

This case demonstrates that proptosis and strabismus, although rare, may represent the initial manifestations of paediatric MOGAD-associated optic neuritis. Awareness of these atypical orbital presentations is important because early recognition, appropriate neuroimaging, and consideration of inflammatory demyelinating disease can prevent misdiagnosis as primary orbital pathology and facilitate timely immunotherapy.

Conclusion

This case highlights that paediatric MOGAD-associated optic neuritis may occasionally mimic orbital disease and should be considered in atypical presentations involving ophthalmoplegia or apparent proptosis. Although the initial presentation raised concern for primary orbital pathology, careful evaluation of the clinical findings, neuroimaging, and MOG-IgG results supported a diagnosis within the spectrum of MOGAD. Borderline MOG-IgG positivity may pose a diagnostic challenge. However, integration of compatible clinical features, characteristic MRI findings, exclusion of alternative diagnoses, and response to immunotherapy can aid diagnosis. Early recognition of atypical presentations is important to facilitate timely treatment and optimise visual and neurological outcomes.

Declaration of Patient Consent

The authors certify that they have obtained all the appropriate patient consent forms. The patient's guardians provided consent for the use of images and clinical information in this report. Names and initials will not be published, and efforts have been made to maintain their anonymity.

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Nil.

Conflicts of Interest

The authors declare no conflicts of interest.

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