IgG4-related disease at the foramen magnum and craniovertebral junction compressing the medulla

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DESCRIPTION

An 8-year-old normal weight boy with a medical history of obstructive sleep apnoea since 4 years of age using a continuous positive airway pressure machine at night and with bronchial asthma was in his usual state of health until 1 month prior to presentation, when his mother noticed significant right eyelid swelling. He had no known risk factors for obstructive sleep apnoea; however, he had a nasal turbinectomy performed 2 years ago, which did not improve his condition. About a year earlier, the mother started noticing bilateral eyelid oedema. She denies weight loss, fever, headache, visual changes, dizziness, ataxia, nausea or vomiting. On

Figure 1 Brain and orbit gadolinium-enhanced T1-weighted MRI. Top row: sagittal and coronal images show two enhancing intracranial extra-axial lesions at the anterior and posterior aspects of the foramen. Middle row: axial images show the severe stenosis at the foramen magnum causing cervicomedullary compression and a temporoparietal dural base enhancing extra-axial lesion. Bottom row: axial and coronal images show bilateral symmetric enlargement and enhancement of the parotid glands and lacrimal glands.

Figure 2 Ultrasound-guided core needle biopsy of the parotid gland. (A) Fibrous scarring, prominent lymphoplasmacytic infiltrates, lymphoid follicles and few residual acini (H&E 20 $\times$). (B) Prominent lymphoplasmacytic infiltrate (H&E 60 $\times$). (C) Kappa light chain immunostain positive cells around unstained salivary gland acini (60 $\times$). (D) Lambda light chain immunostain positive cells around unstained salivary gland acini (60 $\times$).

physical examination, he was alert and oriented. He presented horizontal nystagmus with right eye limited movement to the right and left side. Fundus showed right-sided creamy-coloured plaques within the choroid in a perivascular distribution that may represent a multifocal infiltrative choroidal process. A normal motor and sensory examination with intact cranial nerves were found.

A CT scan of the orbits showed bilateral soft tissue mass in the lacrimal gland with slight proptosis of both globes. Brain and orbit MRI (figure 1) showed multiple enhancing intracranial extra-axial lesions, the largest at the left temporoparietal region with two smaller lesions at the foramen magnum region, causing severe stenosis. Bilateral enhancing enlargement of the lacrimal glands, parotid glands and submandibular glands was identified. Bilateral upper cervical lymphadenopathy was also noted. CT scan of the chest and abdominopelvic cavity showed no lymphadenopathy or solid organ lesions to suggest disseminated disease. Differential diagnosis includes histiocytosis, IgG4-related disease, lymphoma, leukaemia, sarcoidosis, tuberculosis and Rosai-Dorfman disease. Diagnostic workup performed included a fine-needle aspiration of the parotid gland and open core biopsy. Fine-needle aspiration of the right parotid gland showed atypical lymphoid cells suspicious for a lymphoproliferative

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disorder. The ultrasound-guided core needle biopsy showed fibrous scarring and marked lymphoplasmacytic infiltrates. Kappa and lambda light chain immunostain were positive. The IgG4 phenotype antibody study detected an increased IgG4/IgG ratio of 54%. Histopathologic findings were compatible with IgG4-related disease (figure 2). The patient was diagnosed with IgG4-related disease with intracranial hypertrophic pachymeningitis, extracranial sclerosing sialadenitis and orbital disease. The patient was treated with high-dose oral prednisolone daily for 4 weeks, then tapered slowly over the following months.

Immunoglobulin IgG4-related disease is a disease that forms inflammatory pseudotumors composed of IgG4-positive plasma cells and lymphocytes infiltrating the exocrine gland and other organs.¹² It is a non-neoplastic lesion that can mimic a neoplasm. This lesion most commonly occurs in middle-aged patients and very rarely in children. We present the youngest patient with intracranial involvement. IgG4-related disease should be included in the differential diagnosis of patients with dural-based

mass lesions.^{1–3} Moss *et al* reported the first case involving the foramen magnum area.⁴ Kuroda *et al* presented a patient with multiple lesions at the foramen magnum, causing compression of the medulla.² When present at the craniovertebral junction, it can cause osteolytic destruction.⁵ The IgG4-related disease pseudotumors are likely to respond to steroid therapy.^{1,4}

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Learning points

- ▶ IgG4-related disease should be included in the differential diagnosis of patients with dural-based mass lesions.
- ▶ IgG4-related disease can occur in the paediatric population.
- ▶ IgG4-related disease at the foramen magnum area can significantly compress the medulla.

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