A case of nasopharyngeal carcinoma presenting as Susac syndrome

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Introduction

Susac syndrome is a rare disease characterised by the clinical triad of encephalopathy, branch retinal artery occlusions and sensorineural hearing loss (Dörr et al, 2013). Immune- and inflammatory-related syndromes affecting the brain, eye and ear are collectively referred to as 'BEE syndromes', which include Susac syndrome (Triplett et al, 2019). Nasopharyngeal carcinoma is considered to be a differential diagnosis. This article presents a case with BEE symptoms. The main diagnosis was nasopharyngeal carcinoma and there was a considerable delay in recognition of the condition because of the overlap with Susac syndrome.

Discussion

Nasopharyngeal carcinoma is one of the most difficult tumours to diagnose correctly at the initial phase because of the lack of nasal symptoms.

Case report

A 51-year-old man with 'possible Susac syndrome' was diagnosed with nasopharyngeal carcinoma after a relapse. The patient was initially admitted with transient expressive aphasia and lower limb weakness, but the medical history revealed that 4 months earlier, he suffered from an intermittent headache accompanied by severe hearing loss in the right ear. The hearing loss was acute onset without tinnitus and secretions, and worsened with each headache attack. A week before admission, the vision in his left eye had gradually decreased to only light perception. He also had a history of hypertension and chronic rhinitis. Magnetic resonance imaging and angiography of the head showed no abnormalities. Fundus photography and optical coherence tomography indicated branch retinal artery occlusion (Figure 1). He was referred to the authors as having a suspected case of Susac syndrome.

Physical examination revealed that the patient could not hear the sound of rubbing fingers in his right ear but could in his left ear. Tuning fork tests were consistent with sensorineural hearing loss in the right ear. The left eye had vision loss and relative afferent pupillary defect. Blood and CSF tests, including those for infections, immunological indicators and tumour markers, were normal or negative. Tests for demyelinating disease were negative. No vascular stenosis was shown on computed tomography angiography of the head and neck. Fundus fluorescein angiography returned normal results. A hearing test and computed tomography and magnetic resonance scans of the nasopharynx were recommended by a consulting otolaryngologist, but the patient refused further tests. Susac syndrome was suspected because there was direct evidence of branch retinal artery occlusion along with possible sensorineural hearing loss and transient ischaemic attack. Without further testing, 'possible Susac syndrome' was considered as the diagnosis (Kleffner et al, 2016). Treatment with intravenous methylprednisolone was begun at 0.5 g daily for 3 days, and the patient made a rapid recovery. Therapy continued with aspirin and a low dose of oral prednisone.

Outpatient follow up after 1 month saw a sustained improvement of clinical symptoms. However, the magnetic resonance scan revealed an abnormal mass with ring enhancement in the nasopharynx, which raised the possibility of a nasopharyngeal tumour. A tissue biopsy was declined by the patient.

Three months later, the patient presented with persistent headache, aural fullness and hearing loss in the right ear, exophthalmos and limited movement in the left eye. A magnetic resonance scan showed multiple enhanced mass shadows in the bilateral retropharyngeal space and enlarged cervical lymph nodes (Figure 2). Pathology confirmed differentiated non-keratinising squamous cell carcinoma (Figure 3), and he received radiation treatment.

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Susac syndrome is an autoimmune vasculopathy resulting in microvessel thrombosis in the brain, retina and inner ear (Dörr et al, 2013; Kleffner et al, 2016). It is rare, with the prevalence estimated at 0.14 per 100 000 (Seifert-Held et al, 2017). The complete triad is present in only 10–15% of patients at disease onset, which leads to a high rate of misdiagnosis. Features of encephalopathy can include headache, focal neurological deficits owing to transient ischaemic attack or stroke, and global deficits such as encephalopathy and dementia. Magnetic resonance imaging of the brain reveals characteristic callosal lesions (Dörr et al, 2013; Vishnevskia-Dai et al, 2016). The main ophthalmological feature is recurrent multiple branch retinal artery occlusion (Sauma et al, 2020). Sensorineural hearing loss typically affects low to mid frequencies and eventually progresses to total loss.

Both Susac syndrome and nasopharyngeal carcinoma are difficult to diagnose correctly at the initial presentation and need to be carefully differentiated. In addition, this case contains some aspects that have not been reported before, which can be summarised as follows.

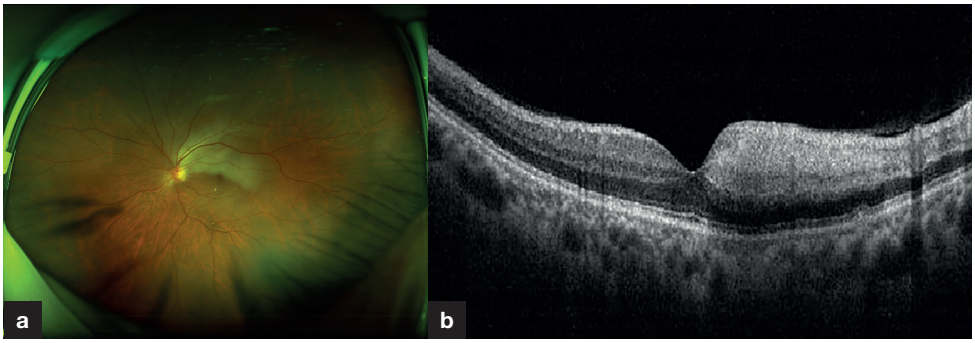

Figure 1. a. Fundus photography showing paleness and oedema in part of the retina. b. Optical coherence tomography showing slight oedema above the neuroepithelium in the macular area, with enhanced reflection in the inner layer.

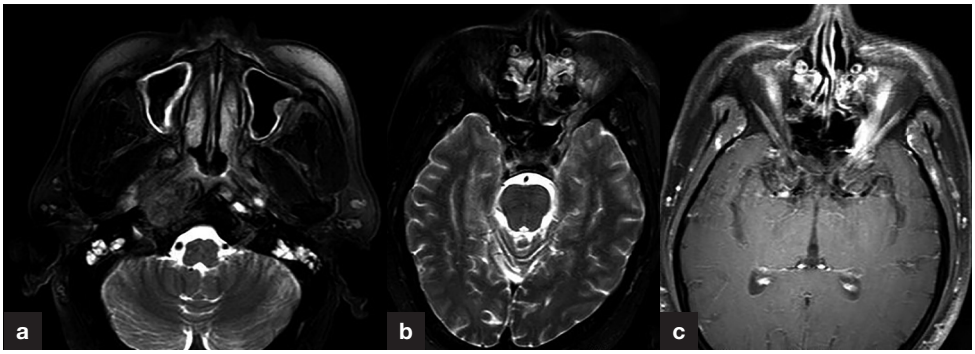

Figure 2. a. Magnetic resonance image showing diffuse soft tissue thickening in the nasopharynx, and the lesions involved the left eustachian tube. b. The lesions involved the right orbit. c. The lesions involved the right optic nerve, and was enhanced on contrast enhancement magnetic resonance.

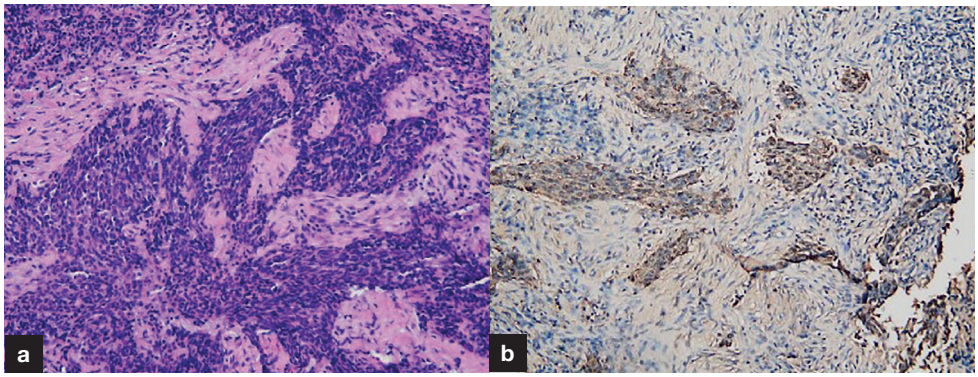

Figure 3. a. Haematoxylin-eosin staining confirmed a poorly differentiated carcinoma. b. Immunohistochemistry showed Epstein–Barr virus-encoded small nuclear RNA (EBER)+.

Learning points

- Nasopharyngeal carcinoma is an important differential diagnosis for brain, eye and ear (BEE) syndromes, including Susac syndrome.
- Neurologists should perform a timely examination of the nasopharynx and skull base when encountering patients with BEE syndromes.
- Cases of 'possible Susac syndrome' need to be thoroughly evaluated for other causes. Whether Susac syndrome is associated with neoplasms needs to be further explored.
- More data are needed to determine whether nasopharyngeal carcinoma increases the risk of cerebrovascular disease.

Since ophthalmic manifestations may be the sole presenting sign of Susac syndrome (Gass et al, 1986; Johnson et al, 1994), Susac syndrome was initially suspected. However, the magnetic resonance imaging and fundus fluorescein angiography results did not support this. Progressive loss of the entire visual field in this patient is inconsistent with the sudden partial vision loss known to occur in branch retinal artery occlusion (Egan, 2019). Improvements in clinical symptoms after methylprednisolone may be explained by a reduction in compression and tissue oedema, or a consequent reduction in tumour volume. Taken together, there was insufficient evidence for a diagnosis of Susac syndrome. However, it is not common for a patient with nasopharyngeal carcinoma to have both branch retinal artery occlusion and transient ischaemic attack at disease onset in the absence of vascular stenosis. Furthermore, the sensorineural hearing loss contradicts the conductive hearing loss often caused by nasopharyngeal carcinoma. These discrepancies led the authors to consider that the diagnosis of Susac syndrome could not be completely rejected, despite there being no previous reports of Susac syndrome being associated with neoplasms. This merits further study.

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