CASE REPORT

Melkersson-Rosenthal syndrome

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Melkersson-Rosenthal syndrome (MRS) is a rare neuro-mucocutaneous disorder which is defined by a triad of orofacial oedema, furrowing of the tongue, and recurrent episodes of facial nerve palsy [1]. Due to the rarity of the disease and inconsistent presentation, MRS remains under-diagnosed and understanding of disease progression and management is still limited.

Case Report

A 30-year-old Caucasian male presented with sequential bilateral painless periorbital oedema (Figure 1). The remaining ophthalmic examination was unremarkable. He denied any recent foreign travel or trauma. He had no history of allergies or atopy.

Extensive investigations including infectious and autoimmune screen, thyroid function tests, immunoglobulin levels and C1 esterase inhibitor function were all normal. MRI orbits showed periocular subcutaneous oedema with no orbital involvement. The patient did not respond to oral antihistamines and was started on high doses of oral prednisolone (1mg / kg / day). He responded well with complete resolution.

Soon after stopping the steroids the oedema recurred and as a result an eyelid biopsy was scheduled six weeks later. Histology demonstrated oedema of the lamina propria with increasing number of mast cells that stained positive with mast cell tryptase and CD117. The patient was once more started on oral steroids (1mg / kg / day) and his symptoms resolved but the tapering off was more gradual.

A few months later he presented with upper lip oedema which eventually involved the lower lip (Figure 2). A lip biopsy showed aggregates of macrophages and T-cells mainly surrounding small vessels and nerves within the submucosa and striated lip muscles without evidence of granulomas. Six months later he developed bilateral sequential peripheral facial nerve paralysis with no other neurological deficit.

Figure 1: A 30-year-old male with a subacute onset of periorbital painless non-pitting subcutaneous oedema.

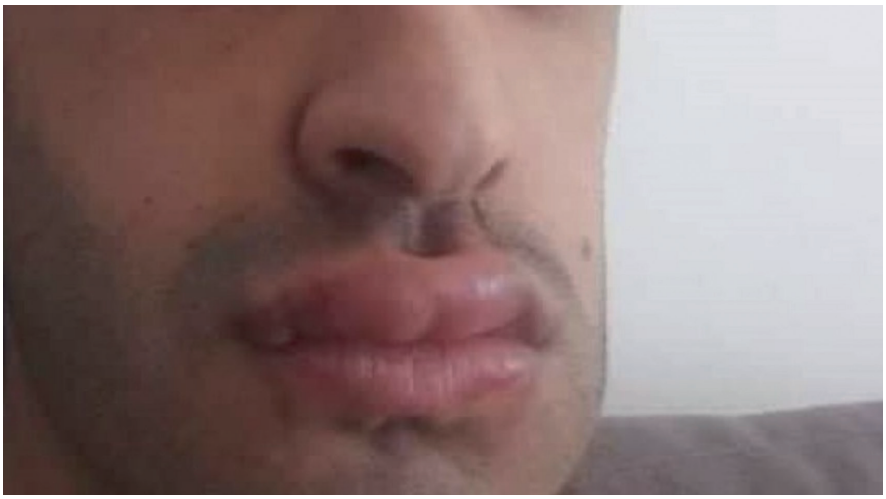

Figure 2: The patient presented several months later with upper and lower lip painless oedema.

As a result of the orofacial oedema and facial paralysis the diagnosis of MRS was made, more than one year after initial presentation. The patient underwent further investigations to exclude malignancy, Crohn's and sarcoid disease and a lumbar puncture for which the results were all normal. He has been on azathioprine 100mg twice daily for 12 months with no further relapses but his facial paralysis has shown little improvement.

> "In patients with recurring orofacial oedema, MRS should be on the differential list"

Discussion

Melkersson-Rosenthal syndrome is a rare, neuro-mucocutaneous disorder characterised by a triad of lingua plicata, painless and non-pitting oedema involving the lips and eyelids, and recurrent episodes of complete or partial peripheral facial nerve paralysis. Disease course is often recurrent or progressive, with episodes presenting over years or decades and spontaneous regression being rare. Diagnosis is often challenging as the presence of the complete triad is seen in less than 20% of patients [2] leading to diagnosis delay with a median time of four to nine years [1].

Histological evidence is not necessary but the presence of one of three signs with positive histopathological features is also enough for diagnosis [1]. Common histopathological features include lymphoedema, noncaseating epithelioid granulomas, Langerhans giant cells, perivascular mononuclear inflammatory infiltrates, and at later stages tissue fibrosis. Current literature suggests a plethora of

aetiologies including autoimmune, infectious, genetic factors, hypersensitivity and allergic reactions [3].

Even though management of MRS remains mostly symptomatic, early diagnosis and treatment can prevent cosmetic sequelae from recurrent swelling and nerve injury. High dose of oral prednisolone (1mg / kg / day) over a period of three to six weeks has shown good results in 50–80% of patients, with symptomatic relief and reduction in relapse frequency. Surgical decompression of the facial nerve can be offered to patients with relapsing or refractory facial palsy [4]. Cosmetic surgery for fibrosis in chronically affected areas, such as the upper lip, has also shown good results [5].

Our patient had two of the three clinical signs. The histology results were most likely influenced by the systemic steroids and were therefore not typical. In patients with recurring orofacial oedema, MRS should be on the differential list. Considering the lack of clinical understanding and awareness of the disease, this case report aims to help raise its profile, in particular its presentation, progression, and treatment.

References

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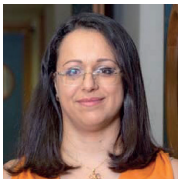

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