Ascher's syndrome: a rare cause of lip swelling

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ABSTRACT

Ascher's syndrome is a rare, benign entity with just over 100 reported cases. The condition is characterised by a 'double' upper lip, blepharochalasis and non-toxic thyroid enlargement. It presents before the age of 20 years in the majority of cases and shows no racial or gender differences. While the exact cause is unknown, hormonal dysfunction and autosomal dominant inheritance have been suggested as possible aetiological factors. We present two cases of Ascher's syndrome referred for investigation of lip swelling.

KEYWORDS

Oral medicine – Lip swelling – Blepharochalasis – Thyroid enlargement

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Introduction

Ascher's syndrome is a rare, benign entity that is characterised by a 'double' upper lip, blepharochalasis and non-toxic thyroid enlargement. There are just over 100 cases described in the literature. We present two cases of Ascher's syndrome referred for investigation of lip swelling and highlight the importance of distinguishing this condition from other more serious ones that may present similarly.

Case 1

A 25-year-old female was referred to the Oral Medicine department for the appearance of a 'double' upper lip. This first became apparent during her teenage years and was becoming more pronounced with age. There was no history of trauma or surgery to the lips. There were no gastrointestinal symptoms and her medical history was unremarkable.

On examination, excess tissue was present on the mucosal aspect of the upper lip, which was more evident when the patient smiled (Fig 1). There was also mild blepharochalasis of the eyelids and diffuse thyroid enlargement, consistent with a goitre (Fig 2). Thyroid function tests revealed normal T3 and T4 levels, and positive antithyroglobulin antibodies. Ultrasonography of the thyroid gland revealed no abnormality.

Based on the characteristic morphology and evolution of the lip enlargement, and the absence of an underlying systemic disease, the diagnosis of Ascher's syndrome was made. The patient was subsequently referred for surgical correction of the upper lip.

Case 2

An 11-year-old boy was referred for the management of a 'double' lip and eyelid swelling. This was first identified at the age of six years. He was asymptomatic and had no systemic symptoms. His medical history was unremarkable. On examination, a fold of excess tissue was evident on the mucosal aspect of the upper lip, which was soft on palpation (Fig 3). There was diffuse soft swelling of the upper eyelids (Fig 4). Haematological assays and radioallergosorbent testing for common allergens were normal. Lip biopsy demonstrated squamous mucosa with non-specific inflammation but no granulomas. Ultrasonography of the thyroid gland was unremarkable.

Based on the clinical phenotype, histopathological findings and absence of systemic involvement, a diagnosis of Ascher's syndrome was made. The patient is undergoing regular review until he reaches an appropriate age to undergo surgical correction.

Discussion

First described in 1920, Ascher's syndrome is commonly characterised by a triad of a 'double' upper lip, blepharochalasis and non-toxic thyroid enlargement. It presents before the age of 20 years in 80% of cases and shows no racial or gender differences.¹ While the exact cause is unknown, hormonal dysfunction and autosomal dominant inheritance have been suggested as possible aetiological factors.² Thyroid enlargement is variable and not considered essential for diagnosis although it is present in 10–50% of cases.³ The aetiology for thyroid enlargement in Ascher's syndrome is unknown.

The appearance of a double lip is caused by a fold of excess or hypertrophic tissue on the mucosal aspect of the lip.⁴ It usually affects the upper lip bilaterally⁵ but can also present unilaterally, involving both upper and lower lips.

Blepharochalasis is a rare condition of inflammation of the eyelids. It is a result of repeated episodes of oedema of the eyelids, which causes the skin to become wrinkled, redundant, discoloured, thin and atrophic, giving the

characteristic appearance of ptosis.⁶ This can result in impairment of vision due to narrowing of the palpebral fissure.

The differential diagnoses of Ascher's syndrome include orofacial granulomatosis, angio-oedema, sarcoidosis, minor salivary gland tumour of the lip, vascular tumours and lymphangioma. The histological changes seen in Ascher's syndrome are not very specific, consisting of prominent salivary glands and mixed inflammatory cell infiltration but no granulomas. Haematological investigations are normal, as are reported findings from imaging studies including ultrasonography and computed tomography. Clinically, the presence of a double upper lip, blepharochalasis and goitre are unique to Ascher's syndrome. When faced with this triad, the diagnosis of Ascher's syndrome should be considered.

Figure 1 Excess tissue noted when smiling, more marked on the left side

Figure 3 Bilateral soft swelling of the upper eyelids

Figure 2 Diffuse enlargement of the thyroid gland

Figure 4 A fold of excess tissue on the mucosal aspect of the upper lip

Management is often surgical correction of the upper lip and eyelids with excision of the excess tissue.^{5,7,8} The use of dapsone 50mg daily has been described in a single case report in an attempt to delay the progression of the condition.⁹

Conclusions

This case series of two patients with Ascher's syndrome highlights the importance of distinguishing this condition from other more serious ones that may present similarly.

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