Ascher Syndrome: Report of a Case with Early Manifestations

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Abstract

Keywords

- Ascher syndrome
- cheiloplasty
- blepharochalasis
- double lip

Ascher syndrome is a disease of unknown etiology first described in 1920 by Ascher, an ophthalmologist from Prague. It presents with recurrent edema of the lip and upper eyelid resulting in double lip and blepharochalasis. In 10% of cases the idiopathic nontoxic thyroid enlargement also occurs. Because of its rarity, it is often undiagnosed. A case of early onset is presented with its respective surgical treatment and outcome.

Ascher syndrome is a disease of unknown etiology first described in 1920 by Ascher, an ophthalmologist from Prague.¹ It presents with recurrent edema of the lip and upper eyelid resulting in double lip and blepharochalasis. In 10% of cases the idiopathic nontoxic thyroid enlargement also occurs. The association of blepharochalasis and double lip also had been described by Laffer in 1909.² This is the reason some authors continue calling this as Laffer–Ascher syndrome.

Because of its rarity, it is often undiagnosed. Generally, it begins at puberty with very few cases reporting manifestations before the age of 8 years. This disease which rarely brings functional problems, often cause psychological disturbances of the affected person because of the facial malformation.

A case of early onset is presented with its respective surgical treatment and outcome.

Case Report

A 10-year-old male patient assisted at hospital with upper lip and upper eyelids deformity, with recurrent edemas starting at the age of 6 years, each episode lasting approximately 1 week.

On clinical examination, upper lip prominence formation was observed with double lip when smiling and herniation of its center by the upper lip frenum insertion; bilateral blepharochalasis with redundant, lax and thin skin hiding the lid margins and eyelash, more marked in lateral zone affecting vision, and mild eye protrusion by bilateral enlargement of orbital fat. In the last year, there had been no recurrent edema of the eyelids. Lower lip and lower eyelids had never been affected before (► **Figs. 1 and 2**).

In cervical palpation, no thyroid enlargement was identified and then this was confirmed by ultrasound.

The patient has no family history and no history of trauma or sucking habit. His laboratory tests including thyroid hormone values and renal function were considered normal. Sinus X-ray and panoramic dental X-ray were requested, no change was observed.

It was decided to perform bilateral upper blepharoplasty under general anesthesia because of the latency stage of the disease. According to conventional technique that consisted in “bird’s tail” shape exeresis of redundant skin³ leaving 10 mm of both upper and lower lid margin intact (► **Fig. 3**). A small portion of middle orbital fat on each side was also removed. The incision was sutured continuously with 6–0 Mononylon.

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Fig. 1 Preoperative frontal view.

Cheiloplasty of double lip was performed on upper lip by horizontal elliptical incision from the commissure of one side to the other preserving labial frenulum removing mucosa and submucosa without muscle, performing incision closure by separate points with 4-0 absorbable suture.

Postoperative progressed without problems with suture removal after 5 days to prevent eyelid visible scar.

Anatomopathological study of the excised lip sample showed epithelial stratified squamous slightly keratinizing tissue with microulceration and acantopapillomatosis, which sits on fibrocollagenous well-vascularized stroma. Chronic

Fig. 2 Preoperative smile.

Fig. 3 Design of the palpebral incision.

inflammatory infiltration, with occasional transepithelial polymorphonuclear migration and glandular mucous acini surrounded by edema (►Fig. 4).

At 12 months postoperative follow-up, good result, no recurrence, facial symmetry, smile, and esthetics correction were observed (►Figs. 5 and 6).

Discussion

The clinical case presented corresponds to an early manifestation of Ascher syndrome. This syndrome has unknown etiology, although authors have suggested autosomal dominant transmission⁴ manifesting in adolescence generally younger than 20 years.⁵ Few articles have been published with the presentation of patients who develop symptoms at the age of 6 years.

In most cases, double lip formation occurs only in the upper lip. Lower lip or both together are rarely affected. The mechanism of formation is because of a persistence of the horizontal sulcus located between the outer cutaneous area (pars glabrosa) and inner mucosal area (pars villosa) suffering hypertrophy.⁶

Surgery is the usual treatment indicated when the lip defect interferes with chewing, speech or produce esthetic changes.⁷ Several techniques have been suggested as

Fig. 4 Lip biopsy.

Fig. 5 12 months postoperative frontal view.

W-plasty,⁸ triangular incision, among others; being the most suitable the horizontal incision.⁹

The blepharochalasis is formed by a repeated angioedema with decreased in dermal elastin and adherence to the elevator fascia, which is related to weakness of the orbital septum and orbital fat protrusion, forming redundant folds falling on the eyelid margin that may cause ulceration of the eyes by deflecting eyelashes. Only 10% of blepharochalasis are

Fig. 6 12 months postoperative smile.

associated with double lip in young people. It is characterized by the following three stages: (1) intermittently painless swelling of the lids; (2) ptosis because of dehiscence of the levator aponeurosis or lax and thin skin falling over lid margin; and (3) ptosis adiposa with medial fat pad atrophy, prolapse of orbital fat, and lacrimal gland.¹⁰ Surgery should be delayed for up to 1 year after the last episode of edema. The most common technique is the usual cosmetic blepharoplasty, in this case by choosing the “bird’s tail” incision design to compensate for the tissue excess at the lateral area of the lids.³

Regarding thyroid manifestations, they are only found in 10 to 50% of cases, usually after the age of 20 years.¹¹ This enlargement courses without symptoms, so expectant behavior is recommended. In this patient, no thyroid alteration was found.

The differential diagnosis includes cardiac or renal diseases, and some syndromes such as Enlher Danlos VII⁴ can also occur in association with other syndromes.¹² Double lip and blepharochalasis can also occur as isolated forms.

Conclusion

Early diagnosis in Ascher syndrome prevents unnecessary treatments and therapeutic delays. Although this syndrome does not bring major functional alterations, the psychological impact that this produce is important. A suitable cosmetic treatment at the correct stage of evolution of the disease is usually sufficient to return the patient to normal.

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