Masked ocular amyloidosis simulating xanthelasma and follicular conjunctivitis: case reports

Amiloidose ocular mascarada simulando xantelasma e conjuntivite folicular: relatos de casos

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ABSTRACT

Amyloidosis is a rare disease in which ocular involvement may occur as an isolated event or associated with a systemic disease. This paper describes two clinical cases of ophthalmologic manifestations of amyloidosis: a bilateral eyelid lesion similar to xanthelasma and tarsal conjunctival disease similar to follicular conjunctivitis. The anatomopathological analysis confirmed the diagnosis.

RESUMO

A amiloidose é uma doença rara que pode ter o acometimento ocular como forma isolada ou estar associada a doenças sistêmicas. Neste relato, são descritos dois casos clínicos de alterações oftalmológicas da amiloidose: um que se manifestou por meio de lesão palpebral bilateral de aspecto similar ao xantelasma. O outro era uma afecção em conjuntiva tarsal, semelhante à conjuntivite folicular. O estudo anatomopatológico confirmou o diagnóstico.

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INTRODUCTION

Amyloidosis is a systemic condition that belongs to a group of protein metabolism disorders.[1,2] Amyloid substance is characterized as a protein that is deposited as insoluble fibrils, especially in the intercellular space of tissues, causing cytotoxic effects with harm to the tissue structure and consequent alterations in the function of the affected organ.[2]

Amyloidosis can be classified as systemic or localized, depending on the location of the deposit site. Systemic amyloidosis is classified as primary and secondary, the latter of which is related to other diseases. Ophthalmological manifestations are a type of localized amyloidosis described as a rare event.[1] In most cases, amyloidosis with conjunctival and eyelid involvement is classified as primary and localized. In cases of the involvement of the cornea, choroid, retina, vitreous and orbital structure, however, systemic amyloidosis is the predominant form. The prevalence is 65.1% in the female sex, with an average age of presentation of 54.9 years.[1]

Eyelid/conjunctival involvement is manifested in different forms depending on the size and location of the masses and can cause pain, diplopia, mechanical ptosis and altered ocular mobility.[3-5] Clinically, amyloid protein deposits may be symptomatic or asymptomatic. The most common clinical eyelid signs include a visible or palpable periocular mass and mechanical ptosis (95.8% and 54%, respectively). When involving the conjunctiva, periocular pain and discomfort, and recurrent subcutaneous hemorrhage constitute the most frequent conditions (25% and 12.5%, respectively).[1,6] When associated with orbital structures, proptosis and altered ocular mobility may be found. In most cases, however, orbital involvement is asymptomatic and painless, which helps in the differential diagnosis of amyloidosis as an idiopathic inflammatory pseudotumor.[7]

Regarding the diagnosis, polarized microscopy with red stain is currently considered the gold standard to identify amyloid in tissues. Extracellular hyaline deposits are seen with hematoxylin-eosin and staining with Congo red, which shows characteristic birefringence to green due to the fibrillar nature.[1,8]

The objective of this case report was to describe two clinical cases of ophthalmological manifestations of amyloidosis: bilateral eyelid lesion similar to xanthelasma and tarsal conjunctival disease similar to follicular conjunctivitis.

This study was approved by the Research Ethics Committee of the *Faculdade de Medicina de São Jose do Rio Preto- FAMERP – SP* - CAAE 64705922.7.0000.5415.

CASE REPORT

Case 1

A 59-year-old white male, hypertensive, with multiple myeloma sought the Redentora Eye Hospital of São José do Rio Preto (SP, Brazil), with a lesion on the upper eyelid of both eyes with slow, progressive growth. The patient also reported the excision of similar lesions in the oral cavity (Figure 1).

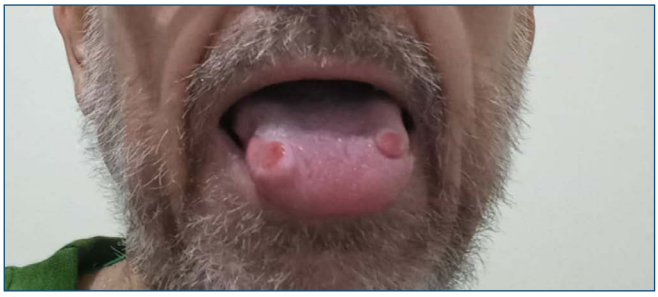

Figure 1. Intraoral photograph showing waxy nodules involving the lateral side of the tongue.

Ophthalmological ectoscopy revealed raised, vascularized lesions of heterogeneous yellowish coloration in the medial region of the upper eyelid of both eyes (symmetrical), measuring approximately 2x2cm in diameter (Figure 2). The rest of the ophthalmological exam was normal.

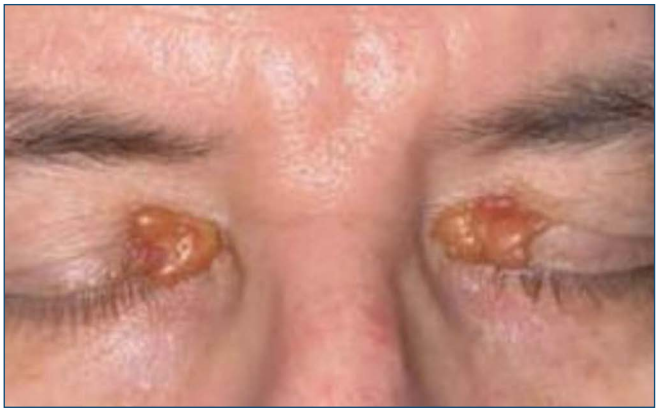

Figure 2. Amyloidosis affecting medial corner of the upper eyelid of both eyes (symmetrical), measuring approximately 2x2cm in diameter.

Excisional biopsy of the lesions was performed, followed by palpebral reconstruction during the same surgical event. The excised material was sent for anatomopathological analysis, and the results revealed extracellular deposits of an amorphous, eosinophilic material stained positive with Congo red. The patient is currently in follow-up with no complications.

Case 2

A 46-year-old male, hypertensive, in dermatological follow-up for vitiligo, sought the Redentora Eye Hospital of São José do Rio Preto (SP, Brazil), with the complaint of a "foreign body sensation" in the right eye with a 30-day history and no improvement with the use of lubricating eyedrops.

The biomicroscopic examination of the right eye revealed a follicular appearance, with yellowish coloration, prominent vascularization around the follicles in the temporal tarsal superior conjunctiva, impairment of the entire fundus of the sac (Figure 3), and punctate keratitis of the upper third.

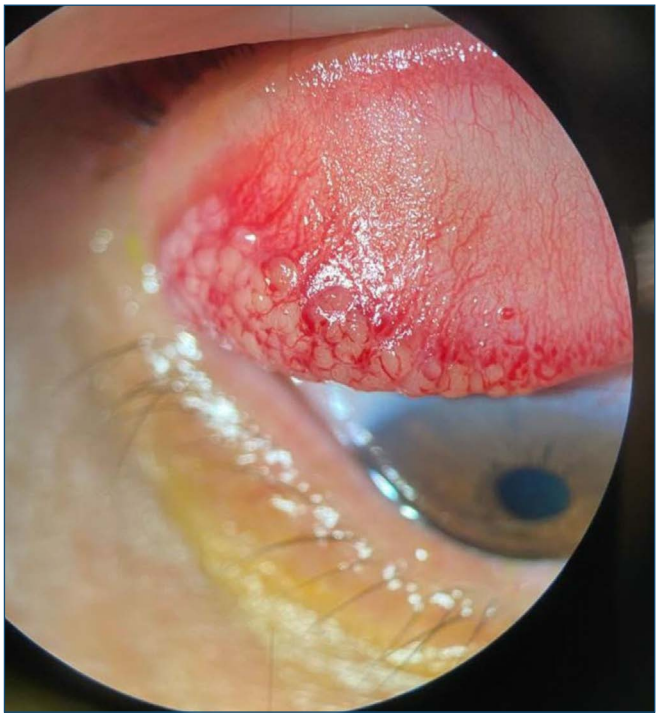

Figure 3. Lesion in right eye characterized by a follicular appearance, yellowish coloration, with prominent vascularization around the follicles in the temporal tarsal superior conjunctiva.

The initial diagnostic hypothesis was chronic allergic and/or infectious follicular conjunctivitis, for which azithromycin (1g) was prescribed orally in a single dose combined with antibiotics and corticoid drops (moxifloxacin 0.5% and dexamethasone sodium phosphate 0.1%) every 6 hours for seven days. In the absence of a clinical response, the hypothesis of conjunctival tumor manifesting as a masked syndrome was raised. Incisional biopsy of the lesion was performed and sent for anatomopathological analysis.

After confirmation of ocular amyloidosis (Figure 4), the patient was referred to a hematologist for systemic evaluation and further management. The negative results confirmed the diagnosis of primary ocular amyloidosis. Follow-up evaluations occurred on the tenth and 13th days, during which excellent healing and corneal re-epithelialization were observed.

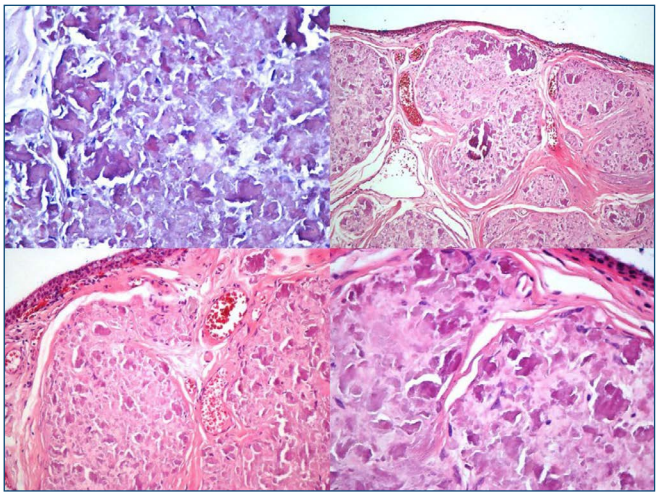

Figure 4. Photomicrograph showing tissue with finely granulose surface, brownish color, firm-elastic consistency, and Congo red staining positive for amyloid deposits. Conclusion: tumoral amyloidosis. Magnification: 100x, 200x and 400x.

DISCUSSION

This paper reports two cases of ocular amyloidosis manifesting as a masked syndrome – one simulating xanthelasma and the other simulating follicular conjunctivitis. Considering the scarcity of publications, the literature describes the rarity and obscurity of ophthalmological manifestations of amyloidosis, enabling a possible differential diagnosis as atypical conditions or not responsive to traditional treatment. The diagnosis of amyloidosis is performed by biopsy and anatomopathological analysis with green birefringence following staining with Congo red.[1,8]

The variability of presentation was also demonstrated in terms of location and systemic association. The first case corresponded to the ophthalmological manifestation of systemic amyloidosis (impairment of other organs), as systemic amyloidosis occurs in approximately 10% of patients with multiple myeloma and related malignancies[9] and secondary to other diseases. The second case corresponded to the ophthalmological manifestation of localized primary amyloidosis.

In both cases, there was complete excision of the lesions on the eyelids and conjunctiva. Only

histopathological analyses can confirm the diagnosis in an objective manner. The short-term follow-up of the patients has been satisfactory, demonstrating the effectiveness of the treatment performed.

There is no consensus on treatment. In the literature, the predominant treatment is surgical, with the excision of the lesions. The average recurrence rate is 15% in the conjunctiva and 29% in the orbit. Cryotherapy and radiotherapy have been used as complementary treatment in the postoperative period of partially removed lesions or in cases of recurrence.[10]

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