

Ductal cyst of lacrimal gland in patient with congenital ichthyosiform erythroderma

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Adriana Ribeiro de Almeida¹, Eduardo Damous Feijó²

ABSTRACT

Lamellar ichthyosis is a congenital disease autosomal recessive which is characterized clinically by peeling of all the body surface due hyperkeratosis mucocutaneous that can cause ocular involvement. We reported a case of ductal cyst of the lacrimal gland in patient with lamellar ichthyosis attended in the Ophthalmological Hospital of Anápolis.

Keywords: *Ichthyosiform erythroderma, congenital; Ichthyosis, lamellar; Genodermatosis; Cysts/pathology; Lacrimal apparatus diseases/pathology; Case reports*

RESUMO

A ictiose lamelar é uma doença congênita de herança autossômica recessiva que se caracteriza clinicamente por descamação de toda a superfície corporal devido hiperqueratose mucocutânea que pode levar a comprometimento oftalmológico. Neste estudo relatamos um caso de cisto ductal de glândula lacrimal em paciente portador de ictiose lamelar atendido no Hospital Oftalmológico de Anápolis.

Descritores: Eritrodermia ictiosiforme congênita; Ictiose lamelar; Genodermatose; Cistos/patologia; Doenças do aparelho lacrimal/patologia; Relatos de casos

¹Residency Program in Ophthalmology, Hospital Oftalmológico de Anápolis, Anápolis, GO, Brazil.

²Department of Ocular Plastic Surgery, Hospital Oftalmológico de Anápolis, Anápolis, GO, Brazil.

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INTRODUCTION

The congenital ichthyosiform erythroderma is a variant of recessive congenital ichthyosis, a lamellar ichthyosis also known as nonbullous congenital ichthyosiform erythroderma, not characterized by cutaneous manifestations resulting from a sharp hyperkeratosis whose main clinical manifestation is the peeling of the skin which can lead, among other changes, to severe eye involvement⁽¹⁾.

Lamellar ichthyosis presents an incidence of 1:200,000-300,000 births, with no difference of distribution between genders^(1,2). It is characterized by proliferative hyperkeratosis, with increase in the number of mitoses and time of passage of keratinocytes through the epidermis, leading to structural changes in these cells⁽¹⁾.

These changes result from a mutation in the TGM1 gene on chromosome 14, and may also involve other genes like Ichthyin, ALOX3/12B, FLJ39501 and ABCA12, SICI and BSI (23, 24), SLC27A4 (9q34.11) and IPS (25)⁽²⁻⁴⁾.

It is a rare congenital defect that, due to its mucocutaneous hyperkeratosis, can lead to catastrophic and recurrent ocular and dermatological conditions. At birth, the disease is manifested with diffuse erythema that develops into thick, sometimes dark scales, which distribute in a mosaic pattern throughout the body surface, predominating in flexor areas. However, in many cases, death occurs early, since genodermatosis can predispose to complications such as dehydration, hypothermia, infections and sepsis^(1,3).

Other systemic alterations may include eclubium, rudimentary and glued ears, scarce, dry, ringy and brittle hair, alopecia, hypohidrosis in varying degrees, and palmoplantar keratoderma⁽³⁾.

Ophthalmologic involvement can occur through the appearance of cicatricial ectropion, lagophthalmos, corneal ulcers, blepharitis, dry keratoconjunctivitis, madarosis, corneal edema, and open-angle juvenile glaucoma⁽⁵⁻⁷⁾.

The aim of this paper is to describe a case of ductal cyst of the lacrimal gland in a patient with lamellar Ichthyosis attended at Hospital Oftalmológico de Anápolis.

Case Report

Female 57-year-old patient diagnosed with congenital ichthyosiform erythroderma with ambulatory follow-up for 5 years in this service. In her previous history, she had a skin graft on the four eyelids due to severe cicatricial ectropion with lagophthalmos and corneal ulcer in both eyes four years ago, evolving with bilateral ulcer healing, but with leukomatous sequelae in both corneas. In the present assessment, she complained of burning, ocular hyperemia and tumor in the supero-lateral quadrant of the left ocular globe with onset about 6 months before, and with rapid growth and no history of trauma, phlogistic signs or pain. The ophthalmologic examination showed visual acuity with correction of 20/50 in both eyes, intense desquamation of the skin of the periocular region, madarosis of eyelashes and eyelashes, ectropion recurrence of the upper and lower eyelid of both eyes, conjunctival keratinization, moderate meibomitis, superficial leukomas in both corneas, intense ocular hyperemia (Figure 1A), and presence of apparently cystic, mobile, painless, fibroelastic consistency tumor located in the topography of the left lacrimal gland (Figure 1B). No external ocular motility changes, proptosis or dystopia. The diagnostic hypotheses were ductal cyst of the lacrimal gland, conjunctival cyst, dermoid cyst, pleomorphic adenoma, lymphoproliferative

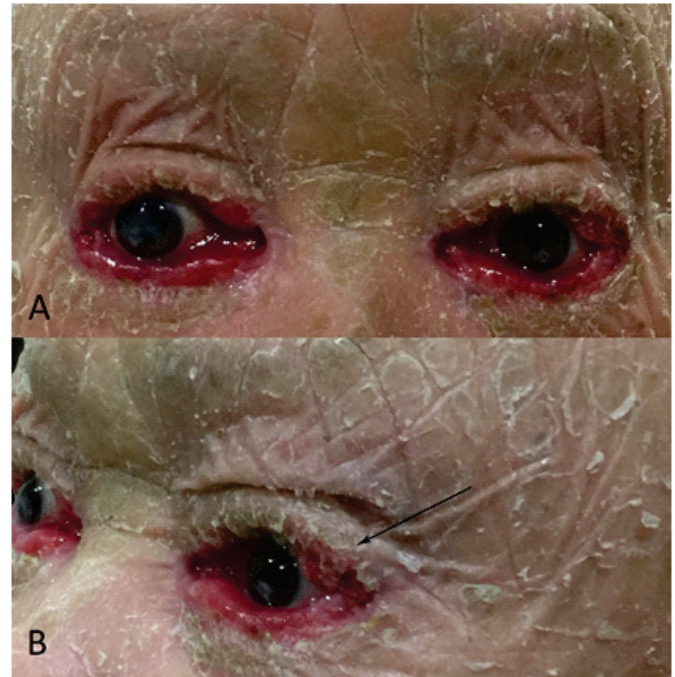


Figure 1. A. Periocular madarose, superior and inferior cicatricial bilateral ectropion, leukomas in both corneas and conjunctival keratinization. B. Small apparent lesion in a super-lateral quadrant of the left ocular globe (arrow)

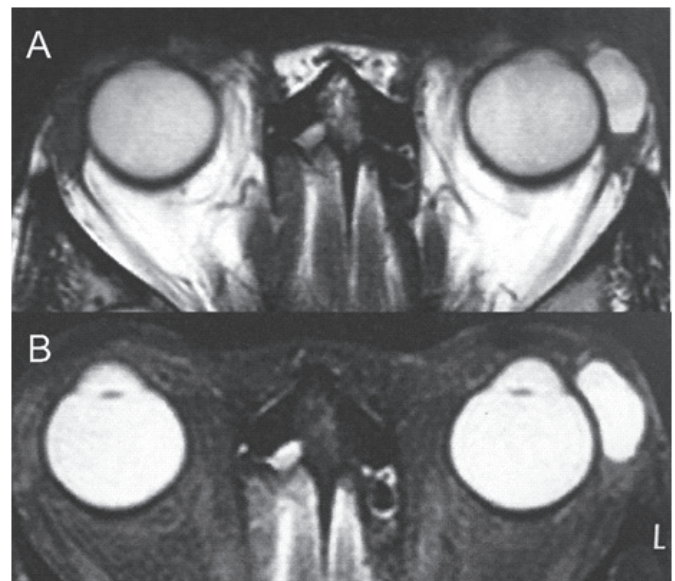


Figure 2. A and B. Magnetic nuclear resonance in T1 and T2, demonstrating cystic lesion and well delimited in the left lacrimal gland.

disease and cystic adenoid carcinoma.

Magnetic Nuclear Resonance (NMR) was requested for diagnostic elucidation, which showed a cystic formation in the left lacrimal gland, with no evidence of septa, vegetation or anomalous impregnation by contrast (Figure 2A and B).

The patient underwent surgery to excision of the cystic tumor by transconjunctival via with subsequent anatomopathological examination. At surgery, the lesion appeared as a cystic tumor of approximately 1.5 x 1.5 x 1 cm in size, easily dissectable from the conjunctiva and in continuity with the lacrimal gland.

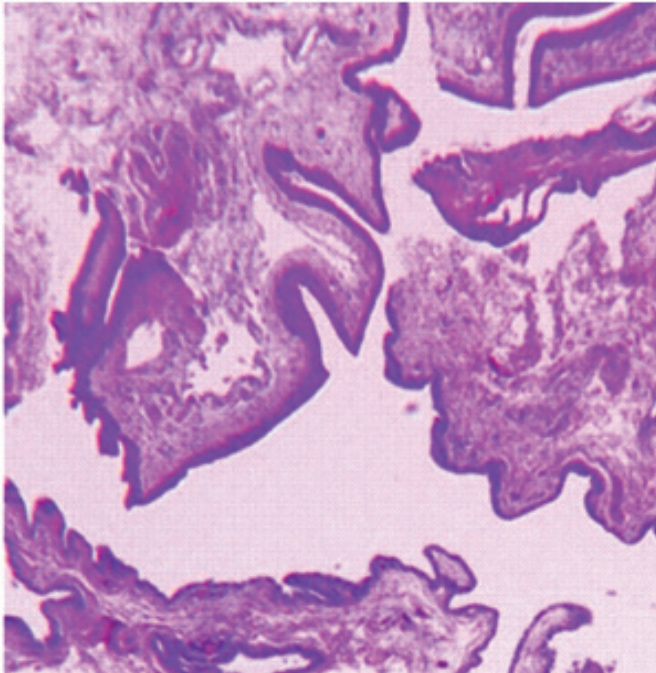


Figure 3. Anatomopathological examination demonstrating cystic spaces surrounded by typical columnar epithelium of the lacrimal gland

The anatomic-pathological medical report revealed cystic spaces delimited by columnar epithelium, corresponding to ductal cyst of the lacrimal gland (Figure 3).

The postoperative period was uneventful, and there was no recurrence after 6 months of follow-up.

DISCUSSION

Lamellar ichthyosis is a congenital disease of autosomal recessive inheritance clinically characterized by extensive, thin and dark scales throughout the body surface, which in turn can result in severe ophthalmological changes, in addition to dermatological changes^(1,8).

In the case reported, the patient is eligible for the diagnosis of lamellar ichthyosis with ocular involvement, highlighting cicatricial ectropion, meibomitis, conjunctival keratinization, cicatrized corneal ulcers and ductal cyst of the lacrimal gland (dacriopus). Thus, it is essential to have specialized follow-up for the amelioration or even correction of comorbidities that may interfere in the development of the patient's vision and quality of life, since it is a disease that can lead to functional limitations, as well as aesthetic and psychological disorders.

Mucocutaneous changes of lamellar ichthyosis may lead to dysfunction of the meibomius glands that can lead to recurrent blepharitis conditions, as well as dry keratoconjunctivitis^(1,6,7). These changes can make the skin a hostile place for the growth of eyelashes and eyebrows leading to madarose^(6,7).

The ectropion occurs due to the significant dryness of the skin that causes retraction of the anterior lamella of the eyelid, both superior and inferior, leading to cicatricial ectropion with a tendency to recurrence, as in the case reported⁽⁹⁾.

The main lacrimal gland cysts are rare entities and are caused by obstruction of their secretory ducts. History of chronic conjunctival inflammations, traumas, and congenital abnormalities

of the gland may predispose this condition. The pathophysiology of the presence of ductal cysts in lamellar ichthyosis is unclear, since both desquamation of the skin and the keratinized conjunctiva and chronic inflammation can lead to glandular hypersecretion and obstruction of the exit holes of the lacrimal gland ducts, leading to the formation of dacriopus. They usually present as cystic tumors, with clear fluid inside and visible in the upper temporal conjunctiva, as in the case reported^(5,6,9).

The differentiation between the diagnoses can be made by imaging tests such as orbit computed tomography or nuclear magnetic resonance - the dermoid cyst will present as a well circumscribed heterogeneous lesion, the pleomorphic adenoma is well delimited, homogeneous and spherical, but the dacriopus is presented as a cystic formation with well defined limits. Therefore, the importance of imaging and anatomical-pathological examinations to confirm the diagnosis⁽¹⁰⁾.

Angmo et al.⁽⁷⁾ reported a case of open-angle juvenile glaucoma in a patient with lamellar ichthyosis, showing that an association between juvenile glaucoma and lamellar ichthyosis cannot be made, but highlighting the importance of a detailed ophthalmologic examination to cover any changes that the patient may present.

The treatment of ichthyosis patients require a team with multidisciplinary approach consisting of neonatologists, dermatologists and ophthalmologists. Among the therapeutic options for ocular damage, extensive lubrication of the ocular surface can be achieved using artificial tears, immunosuppressants such as cyclosporine, tacrolimus, and N-acetylcysteine⁽¹¹⁾.

Dacriopus are rare entities, as well as lamellar ichthyosis, but their association may be frequent and underdiagnosed, since ichthyosis generates ophthalmological alterations favourable to its onset. Case reports like this are important to keep alive the knowledge of pathologies that, being rare, end up surprising specialists in the day-to-day clinical care, especially ophthalmologists, who may face systemic diseases, which can lead to severe and recurrent ocular damage, as described.

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Corresponding author:

Eduardo Damous Feijó

Setor de Plástica Ocular do Hospital Oftalmológico de Anápolis, Anápolis, Goiás, Brazil. Av Faiad Hanna, 235. Cidade Jardim Anápolis-GO Cep: 75080-410 Email: eduardodff@yahoo.com.br