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Case Report

Isolated bilateral congenital lacrimal gland agenesis – Report of two cases

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Abstract

Congenital lacrimal gland agenesis, also called congenital alacrima, is a rare cause of dry eye and is characterized by aplasia or hypoplasia of lacrimal glands. We present two 5-year old children with congenital lacrimal gland agenesis. The two cases had the final diagnosis of isolated bilateral congenital lacrimal gland agenesis and we document the clinical aspects, treatment and present a literature review related to this rare condition.

Keywords: Alacrima, Lacrimal gland, Lacrimal gland agenesis, Punctal plugs

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Introduction

The lacrimal gland arises in human embryos of 22–24 mm crown-rump length from the ectoderm of the superior conjunctival fornix at the end of the second month of intrauterine life.[1] Interaction between epithelial and mesenchymal cells is necessary for proper lacrimal gland development and also governs salivary gland development. The orbital and palpebral aspects are formed in the fifth month of life but full differentiation occurs three to four years after birth. Therefore any structural alterations in very early intrauterine life may cause lacrimal gland agenesis.[1,2]

Congenital lacrimal gland agenesis is a rare cause of dry eye in childhood and is characterized by aplasia or hypoplasia of the principal lacrimal gland.[2,3]

Lacrimal gland agenesis can occur as an isolated condition or in association with salivary gland agenesis, lacrimal drainage system atresiaas well as systemic comorbidities.[4,5]

We present two cases of isolated bilateral congenital lacrimal gland agenesis attended in 2016 at King Khaled Eye Specialist Hospital - Saudi Arabia, including a description of the clinical aspects, treatment and a review of the relevant literature.

Case report

Case 1

A 5-year-old male presented for an evaluation because the parents noted the absence of tears when the child cried, since birth. The patient was otherwise healthy and denied any pain, redness, photophobia or foreign body sensation. No symptoms of dry mouth were reported. There was no positive family history or symptoms of alacrima. On examination, visual acuity was 20/20 in both eyes (OU). Tear breakup time (BUT) was less than 2 s OU. Conjunctival hyperemia with staining and corneal punctate epithelial keratitis were present. The patient had normal corneal sensation OU. The dilated fundus examination was unremarkable. The lacrimal excretory system was normal with no other abnormalities.

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Magnetic resonance imaging (MRI) confirmed agenesis of both lacrimal glands and normal salivary and submandibular glands (Fig. 1). The patient was diagnosed with isolated congenital alacrima and received bilateral punctal plugs in the upper and lower lids. Currently she is being treated with lubricants resulting in an healthy corneal surface.

Case 2

A 5-year-old healthy female presented to evaluation because the parents noticed the absence of tears when the child cried since birth. There was no pain, redness, photophobia or foreign body sensation and no positive family history of symptoms of alacrima. On examination vision was 20/20 OU. BUT was less than 1 s OU. There was conjunctival hyperemia with staining and diffuse corneal punctate epithelial erosions. The corneal sensation was normal OU and the dilated fundus examination was unremarkable. The lacrimal excretory system was normal with no agenesis or anomalies. Computed tomography (CT) scan revealed agenesis of both lacrimal glands with the presence of normal salivary and submandibular glands (Fig. 2). The patient was diagnosed with isolated congenital alacrima. She was treated with lubricants and bilateral punctal plugs in the upper and lower lids. The parents reported that she is more comfortable after treatment.

Discussion

We present a very rare condition, bilateral isolated alacrima, diagnosed in two children with similar presentations. Both children present at 5 years of age. Other case reports have been presented on patients who were 6 years old at presentation.[3,6,7] The majority of previous cases reported alacrima occurring in male children.[3,6,7] In the current report, we present a boy and a girl with the condition.

Interestingly in both cases the absence of tears when crying led to presentation to the hospital. There were no other signs or symptoms of dryness, besides punctate keratitis at presentation in both cases. Other cases of alacrima presented with filamentary keratitis as a sign.[3,6,7]

The lacrimal film is fundamental for the maintenance of a healthy ocular surface. The lipid layer of the tear film in infants is thicker than in adults, slowing evaporation and increasing tear film stability, and thereby preventing the ocular surface from drying. In addition, less ocular surface is exposed in infants.[8] The combination of these factors (i.e. a more stable tear film and a smaller ocular surface area) may explain the relatively fewer symptoms in children even in the absence of the principal lacrimal gland.

The principal lacrimal gland produces tears on crying and the accessory lacrimal glands, distributed in the eye surface, are responsible for the production of tears in normal conditions. Hence, evaluation of the accessory lacrimal glands is also important in similar cases.

However in the current cases, we did not evaluate the accessory glands or the amount of tears on the ocular surface as examination in children is significantly more difficult than adults. If the examination is performed under sedation the lacrimal film may be affected due to the anesthetics or sedatives.[9]

Although we have not done Schirmer 2 test to evaluate the accessory lacrimal glands we believe the tear production was normal because the ocular surface was almost normal, except for mild punctate keratitis.

The ophthalmic evaluation indicated both patients had no other abnormalities of the eyelids or the lacrimal excretory system. The ophthalmic examination in such cases should

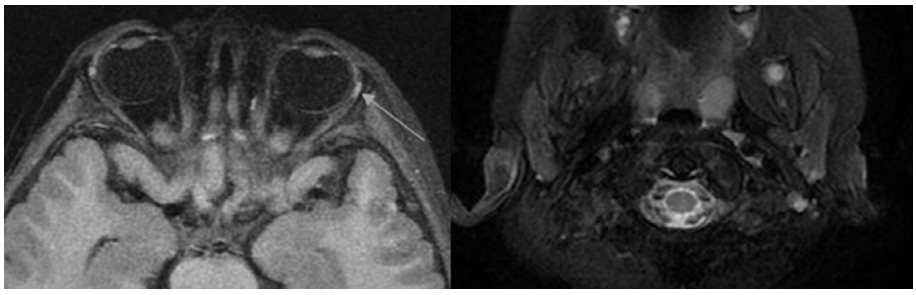

Figure 1. Case 1 - MRI showing non-contrast axial Tl WI and Axial T2 WI with fat suppression with the absence of both lacrimal glands with empty lacrimal fossa (white arrow) and normal appearing parotid gland.

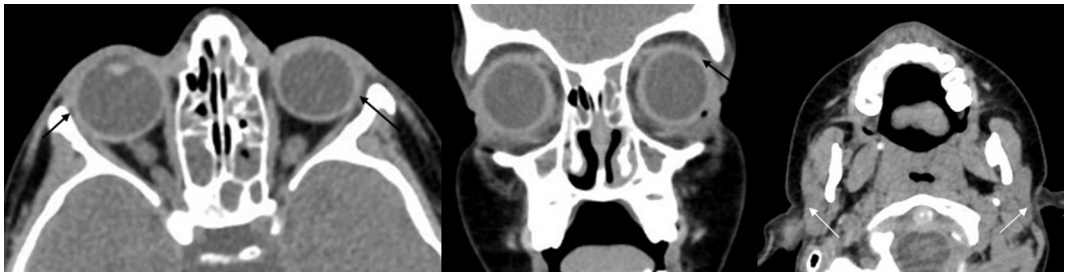

Figure 2. Case 2 - Non contrast axial coronal CT scan image showing the absence of both lacrimal glands with empty lacrimal fossa (black arrow) with normal appearing parotid (white arrows) glands.

exclude the absence of puncta which can be associated with alacrima.[2,4,5,10]

We performed additional tests such as imaging studies because even with the absence of the palpebral lobe of the lacrimal gland, the orbital portion can be in place. Although patient 2 had the lacrimal gland screened using CTscan, MRI is the best choice for lacrimal gland evaluation. Hence, image examinations aided information in the diagnosis of bilateral absence of the lacrimal glands and also confirmed the presence of normal salivary glands characterizing both cases as isolated alacrima.

The parents of both patients denied other similar cases in the family. This information is important as alacrima can have a hereditary background and is linked to familial dysautonomia or other conditions such as hereditary congenital alacrima.[11] Mutations of the fibroblast growth factor 10 (FGF10) gene, which is necessary for the development of both the lacrimal and the salivary glands in mice, have been found in association with aplasia of the lacrimal glands and salivary gland syndrome (ALSG syndrome) explaining the link between lacrimal and salivary gland aplasia.[11]

Both patients in the current report underwent extensive ophthalmic and systemic examination. However, all the examinations were negative. Systemic evaluations are important because ocular surface dryness can be associated with systemic disorders such as Sjogren's, Riley-Day or ectodermal dysplasia syndrome, hypovitaminosis A secondary to small bowel atresia,[12]and possibility of graft-versus-host disease after bone marrow transplant, with abnormalities of the ear, teeth and digits in lacrimo-auriculo-dento-digital syndrome[11] or with blepharophimosis syndrome.[13] Other differential diagnosis of congenital alacrima includes persistence of neonatal alacrima and achalasia-addisonianism-alacrima syndrome and the absence or hypoplasia of the lacrimal gland.[14] Additionally, rheumatological disease should be considered as part of the workup for children with dry eye and also alacrima can be an early symptom of Allgrove syndrome, the AAA syndrome (achalasia, alacrima, and adrenocorticotropin (ACTH) insensitive adrenal insufficiency).[15] Both patients were negative for all these conditions.

Success rates of treating these patients with punctal occlusion suggest that basal tearing from the accessory lacrimal glands is intact.[11] Patients with both the absence of lacrimal glands and puncta might present with less severe keratoconjunctivitis sicca than those with the absenceof lacrimal gland alone reinforcing the fact punctal plugs or other forms of permanent occlusion of the excretory lacrimal system as cauterization or surgical procedures can be good solutions when the puncta are present.

Hence, punctal occlusion with plugs in addition to topical lubricants appears to be successful for treating congenital alacrima.

Isolated congenital alacrima or lacrimal gland agenesis is a rare condition which should be considered in the differential diagnosis of childhood dry eye syndrome. In children with dry eye syndrome without any systemic findings, it should be emphasized that noninvasive measures such as imaging studies can be undertaken primarily for confirming the diagnosis of isolated lacrimal gland agenesis. The use of punctal plugs associated with lubricants can be successful to treat these cases.

Conflict of interest

The authors declared that there is no conflict of interest.

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