Case Report
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Familial lacrimal punctal agenesis in who are members of three generations

Abstract

Lacrimal punctal agenesis is a rare cause of congenital epiphora and may occur in isolation or in combination with other lacrimal drainage anomalies such as pathologies of the canaliculus, lacrimal sac, and nasolacrimal duct. It rarely occurs in a familial manner. If diagnosed earlier, management of this condition may be as simple as a sharp opening or may necessitate more complex interventions. Here, we report the familial absence of lacrimal punctal in four patients who are members of three generations.

Keywords: lacrimal punctal agenesis, familial, lacrimal drainage system anomalies

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Introduction

Epiphora in the presence of lacrimal punctal agenesis is a rare condition. There are few reports on the congenital absence of lacrimal punctum in the current literature. It may occur in an isolated form or more frequently in association with other genetic or developmental anomalies, which may help to recognize signs of serious childhood problems.[1] Symptomatic children mostly present with recurrent epiphora and chronic conjunctivitis. In particular, patients with a single absent punctum may be asymptomatic and remain undiagnosed.[2] We present a case of a patient with epiphora in both eyes whose family members belonging to three generations showed a history of punctal agenesis.

Case report

A 28-year-old male was admitted with epiphora in both eyes since birth. On examination, four of the punctal were found to be atretic. His 62-year-old mother was complaining of watering in both eyes and had absent right inferior punctum. The lacrimal irrigation test revealed obstruction on both sides. The patients two children, an 8-year-old girl and a 9-year-old boy, also had congenital epiphora, and all punctal were atretic in both children (Figure 1) (Figure 2). No coexisting oculofacial anomalies or systemic conditions were detected. In all the patients, there were mild depressions at the localizations of punctal, which were opened using a hypodermic needle and punctum dilator. Lacrimal irrigation test revealed patent nasolacrimal passage in the index patient. His mother received bilateral external dacryocystorhinostomy (DCR) with silicone tube intubation. Epiphora was resolved after the bilateral operation. Punctal openings were successfully performed in the two children, which revealed intact canaliculi. Lacrimal irrigation with fluorescein revealed patency on both sides in the boy and unilateral patency in the girl. During the follow-up, watering of the eyes resolved at the patent sides. The girl received external DCR, and the complaints regressed postoperatively. There were no recurrences at the end of the 3 years of follow-up.

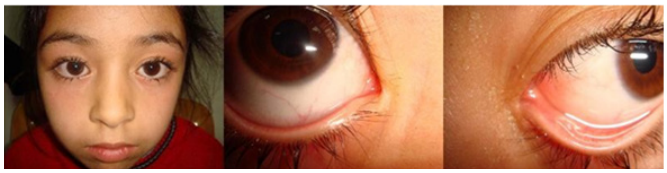

Figure 1 Bilateral punctal agenesis in the 8-year-old girl.

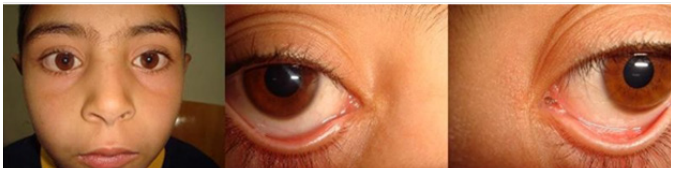

Figure 2 Bilateral imperforated lacrimal punctal in the 9-year-old boy.

Discussion

Absence of lacrimal punctum is a rare condition. It may be from a defect during the embryonic development of the nasolacrimal system. Agenesis of the punctum can develop unilaterally or bilaterally. It can be an isolated finding or combined with other congenital oculofacial and systemic abnormalities. Punctal agenesis is usually associated with the absence of canalicular tissue or lacrimal ducts and predisposes the patient to dilatation of nasolacrimal duct or lacrimal sac cyst.[3] The origin of lacrimal glands is surface ectoderm and it is oriented in the super temporal region of the orbit. The lacrimal drainage system, including canaliculi, lacrimal sac, and nasolacrimal duct, is also derived from the surface ectoderm. Though lacrimal canaliculus becomes patent at the fourth month after birth, punctal development does not occur until the seventh month of gestation.[4]

Lacrimal gland agenesis may be associated with abnormalities derived from first and second branchial arches, aplasia or hypoplasia of the lacrimal punctum, and also from agenesis of the salivary glands. There may also be various facial anomalies such as hemi facial microsomia, mandibulofacial dysostosis, and lacrimo-auriculodento-digital syndrome.[5] The combined absence of lacrimal canaliculi and

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puncta was reported in patients with CHARGE syndrome.[6] Boerner et al.[1] reported about two cases with isolated punctal agenesis and five cases with combined punctal and systemic abnormalities. Cahill et al.[7] described about a patient with punctal and canalicular agenesis. Lai et al.[8] reported lacrimal fistula, skin tag, and staphylococcal disease as ocular abnormalities related to punctal agenesis and cleft lip and cleft palate, secretory otitis media, hydrocephalus, and low birth weight as related general physical abnormalities. Saltzmann and Lissner described an uncommon case of familial punctal atresia with an obvious genetic linkage to bilateral preauricular sinuses, without any such co-morbid syndromic features.[9]

In the light of the above data, agenesis of puncta seems to be frequent than expected. Asymptomatic patients tend to be discovered by chance, and hence presentations are limited to only case reports or case series in the current literature. Patients with punctal agenesis should be precisely examined before the operations for possible coexisting lacrimal system abnormalities. Punctal stenosis and presence of a membrane formation should be excluded. Probing or external DCR may be the treatment option in advanced cases. Punctal, canalicular agenesis, and nasolacrimal duct obstruction may be found together or in isolated conditions. In 86% of patients with absence of both puncta, no canalicular tissue was detected intraoperatively.[10] In partial occlusion of puncta, treatment could be done easily using Jones'one-snip method.

Conclusion

We presented a family with punctal agenesis and nasolacrimal duct obstruction without any other ocular or nonocular abnormalities. The puncta should be evaluated in cases of congenital epiphora, through which a simple intervention could restore the normal lacrimal drainage. A good history taking should also be included in the evaluation of such patients in order to detect and treat undiagnosed family members.

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None.

Conflicts of interest

The authors declare there is no conflict of interests.

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