Bilateral congenital agenesis of lacrimal glands in a Tunisian family

Agénésie bilatérale de la glande lacrymale dans une famille Tunisienne

Ahmed Chebil, Yosra Falfoul ,Rym Limaiem, Leila El Matri

Department B of ophthalmology, Hedi Rais Institute of Ophthalmology /Faculty of medicine of Tunis, university of El Manar, Tunis

RÉSUMÉ

Prérequis : Les ulcères cornéens chroniques nécessite une enquête étiologique etposent souvent des problèmes thérapeutiques. Elles sont graves et engagent souvent le pronostic fonctionnel.

But: Nous rapportons l'observation d'un patient présentant un ulcère cornéenbilatérale, chronique révélant une forme familiale rare d'agénésie bilatérale de la glande lacrymale.

Observation: Un homme de 39 ans adressé pour ulcère cornéen bilatérale, stérile et chronique. A l'examen, le temps de rupture du film lacrymal était inférieur à une seconde et le test de Schirmer a montré l'absence de sécrétion lacrymale. A l'interrogatoire, il a rapporté l'absence de larmoiement même quand il pleurait. Ce fut le cas aussi de son frère et sa sœur. L'échographie orbitaire complétée par l'imagerie par résonance magnétique ont montré l'absence des deux glandes lacrymales chez les deux frères et la sœur. Nos patients ont été traités par des larmes artificielles topiques associées à une occlusion permanente des points lacrymaux inférieurs avec une bonne évolution clinique.

Conclusion : Nous rapportons, à notre connaissance, le premier cas d'une famille Tunisienne présentant une agénésie bilatérale de la glande lacrymale et les premiers cas documentés de forme familiale diagnostiqués à l'âge adulte.

Mots-clés

Agénésie, glande lacrymale, famille Tunisienne

SUMMARY

Background: Chronic corneal ulcers still pose etiological and therapeutic challenge. They are serious complications and often associated with poor functional prognosis.

Aim: We report the case of a patient with bilateral and chronic corneal ulcer revealing a rare familial form of bilateral agenesis of the lacrimal gland.

Cases report: A 39-year-old man was referred to our department for bilateralchronic and sterile ulcer. The tear break-up time was less than one second and the schirmer test detected no wetting in 5 minutes. He mentioned that lacrimation had been totally absent even when crying as was the case of his brother and his sister. Orbital echography showed absence of lacrimal gland. Orbital magnetic resonance imaging revealed absence of both lacrimal glands. Our patients were treated with permanent topical artificial tears. We performed also permanent occlusion of lower lachrymal poncti to preserve basic tear flow.

Conclusion: Congenital lacrimal gland agenesis is rare. We report, to ourknowledge, the first case of Tunisian family with three patients suffering from bilateral lacrimal gland agenesis and the first documented familiarly cases diagnosed in adulthood.

Key-words

Agenesis; lacrimal glands; Tunisian family

Chronic corneal ulcers still pose etiological and therapeutic challenge. Lacrimal gland agenesis (LGA) is a rare congenital abnormality and which it is usually associating with salivary gland agenesis or abnormalities of the lacrimal puncta. The transmission is autosomal dominant. Isolated cases of LGA are extremely rare [1]. Only a few cases are reported in the literature [1-4]. We report the case of a patient with bilateral and chronic corneal ulcer revealing a rare familial form of bilateral agenesis of the lacrimal gland.

CASE REPORTS

Case 1: A 39-year-old man, offspring of consanguineous marriage, was referred to our department for bilateral chromic and sterile ulcer. The patient complained about photophobia since his young age. At the first examination, his best-corrected visual acuity was 0.125 in the right eye (RE) and 0.1 in the left eye (LE). Slit lamp examination disclosed bilateral conjunctival hyperemia, right large corneal ulcer without stromal infiltration (figure 1a) and left corneal central scar opacity (figure 1b). The tear break-up time was less than 1 second and the Schirmer test detected no wetting in 5 minutes (figure 1c and 1d). Fluorescein stains revealed large ulcer with punctuate epithelial erosions (figure 1e and 1 f). Lacrimal ducts were open.

Parents mentioned that lacrimation had been totally absent; even when crying as was the case of his brother and his sister. He had no signs of dysphagia or salivary glands abnormalities. Both brother and sister were exanimated.

Case 2: The brother 31-year old had best-corrected visual acuity 0.4 in the RE and 0.3 in the LE. Slit lamp examination disclosed conjunctival hyperemia and corneal diffuse punctuate epithelial erosions (figure 2). Tear break-up time was less than 1 second and the Schirmer test detected 1 mm of wetting in 5 minutes in each eye. Lacrimal ducts were permeable.

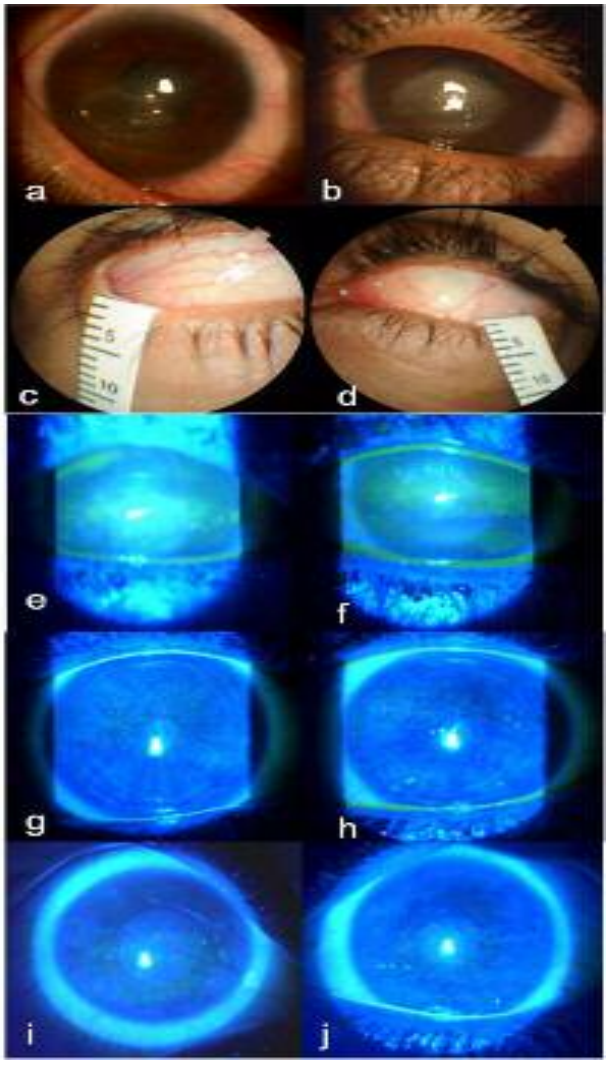
Case 3: The sister 35-year old had best-corrected visual acuity 0.9 in the RE and 0.4 in the LE. Slit lamp examination showed diffuse corneal punctuate erosions (figure 3). Tear break-up time was less than 1 second and the Schirmer test detected 2 mm of wetting in RE and 1 mm LE in 5 min. Lacrimal ducts were open.

The slit lamp examination of the parents showed normal cornea and their Schirmer test detected 6mm of wetting in 5 minutes. Lacrimal ducts were open.

In case 1, a rheumatology workup was performed, and all laboratory tests including Rheumatoid factor, anti-SSA, anti-SSB, antinuclear antibodies, C-reactive protein and serum angiotensin-converting enzyme titer were negative. Salivary glands biopsia was also normal. Orbital echography showed the absence of lacrimal gland, but it was insufficient to confirm the diagnosis (figure 2). Orbital magnetic resonance imaging (MRI) was performed in case 1 and revealed absence of both lacrimal glands (figure 3).

Our patients (cases 1, 2 and 3) were treated with permanent topical artificial tears. That was insufficient for case 1, so we performed permanent occlusion of lower poncti to preserve basic tear flow. Three months later, corneal ulcer healed. The Schirmer test detected 1mm of wetting in 5 minutes.

Figure 1 : clinical findings
a: right large corneal ulcer without stromal infiltration, b: left corneal central scar opacity
c: wetting 0 mm in 5 minutes in RE d : wetting 0 mm in 5 minutes in LE
e: Fluorescein stains revealed large ulcer with punctuate epithelial erosions in RE
f: punctuate erosions in LE
g: corneal diffuse punctuate epithelial erosions in case 2 in RE
h: corneal diffuse punctuate epithelial erosions in case 2 in LE
i: diffuse corneal punctuate erosions in case 3 in RE
j: diffuse corneal punctuate erosions in case 3 in LE



DISCUSSION

Congenital lacrimal gland agenesis is rare but should be considered in young patients with long history of dry eye and chronic aseptic ulcer. The Schirmer test must be done in all cases of chronic corneal ulcers and completed by orbital magnetic resonance imaging if necessary. To our knowledge, this is the first report of

Tunisian family with bilateral congenital agenesis of lacrimal glands. Lacrimal gland has a double origin; the glandular parenchyma derives from the epithelium and the surrounding mesenchymal stroma. Full differentiation occurs in 3 to 4 years after birth. Accessory lacrimal glands produce the basic tear secretion while main lacrimal glands give reflex tears; it explains the various presentations in our patients.

Isolated congenital lacrimal gland agenesis is extremely rare and only few cases are reported [1,2,4]. A previous report presented 2 cases of lacrimal gland agenesis in the same family in which a 6-year old boy and his mother complained about severe punctuates epithelial erosions and mucous filaments [1]. Most cases of LGA occur in children with early presentation. Only one case of hypoplasia presenting in adulthood has been reported; but acquired causes were not ruled out [3]. This affection is probably genetically determined. The prevalence and the causing genes are until now unknown. In our cases, transmission is autosomal recessive, but rare autosomal dominant cases have also been described [1]. More investigations are necessary to identify the genetic mechanisms of the disease.

Figure 2 : orbital echography mention absence of lacrimal glands, a: case1 , b: case2, c: case 3, d: normal orbital echography of a comparative normal case

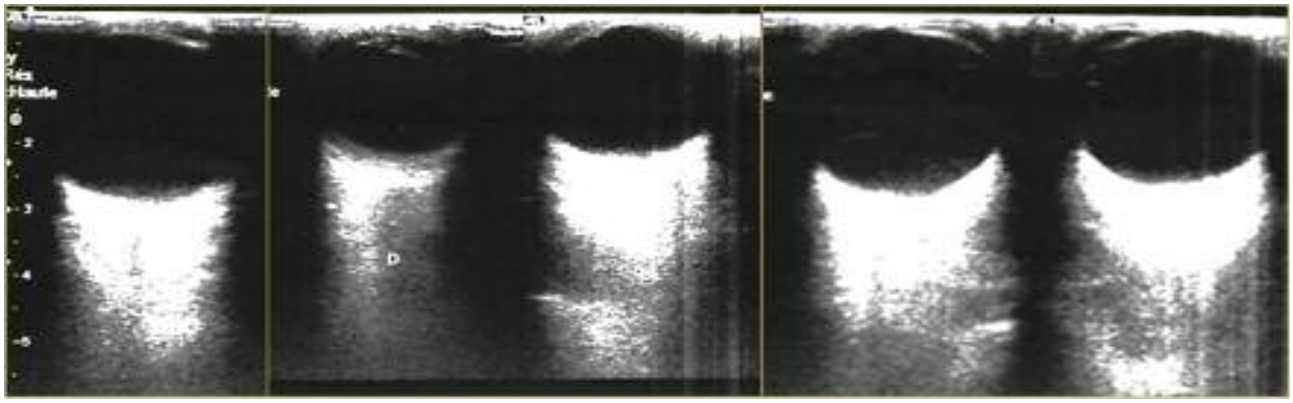
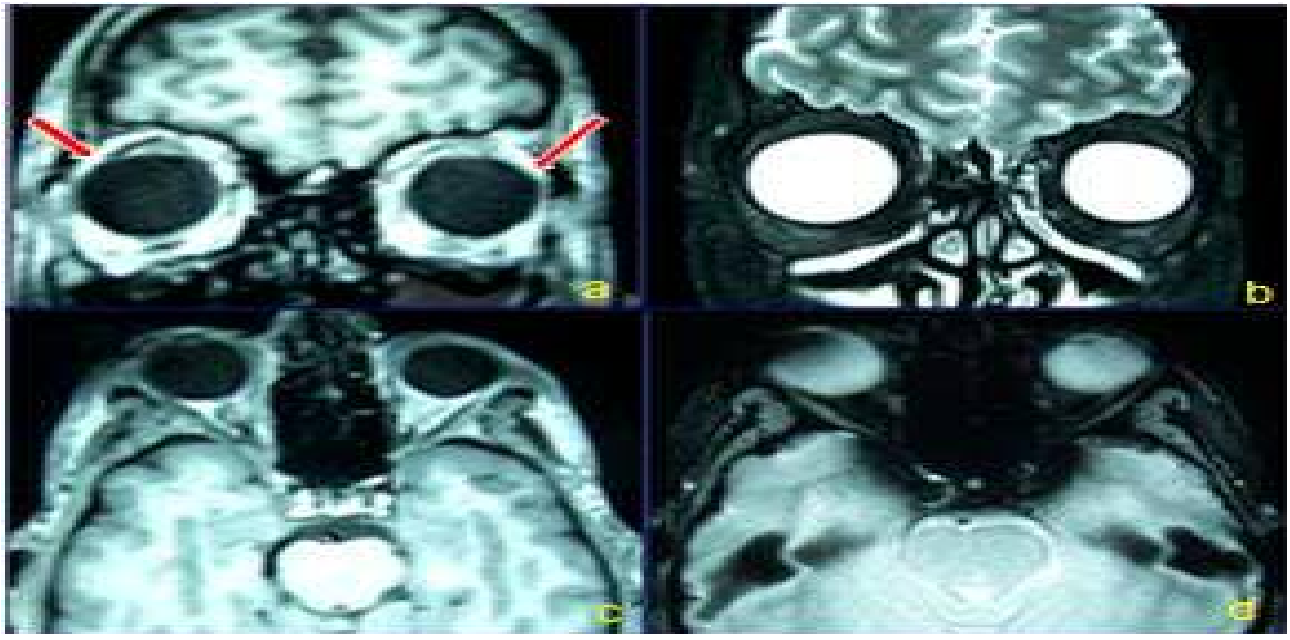


Figure 3 : MRI (a) Coronal T1-weighted, non-contrast image (b) Coronal T2-weighted, contrast image; (c) Axial T1-weighted, non-contrast image; (d) Axial T2-weighted, contrast image. The arrows showed absence of both lacrimal glands.



MRI scan confirms the diagnosis; treatment is based on artificial tears instillation. In case of permeability of lacrimal ducts obliteration of the puncti may improve basic tear flow [1]. Recently, Hirayama and al have performed regeneration of lacrimal gland by bioengineered organ germ method in the hope that will be the ultimate therapeutic model for dry eye diseases [5].

CONCLUSION

Congenital lacrimal gland agenesis is rare but should be considered in young patients with long history of dry eye and chronic aseptic ulcer. We report, to our knowledge, the first case of Tunisian family with three patients suffering from bilateral lacrimal gland agenesis and the first documented familiarly cases diagnosed in adulthood.

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