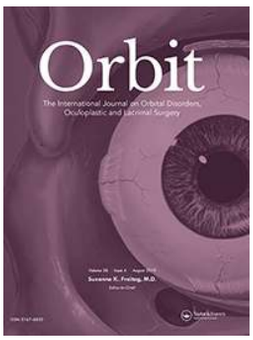

Orbit

The International Journal on Orbital Disorders, Oculoplastic and Lacrimal Surgery

ISSN: 0167-6830 (Print) 1744-5108 (Online) Journal homepage: https://www.tandfonline.com/loi/iorb20

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To cite this article: Jaee Milind Naik, Milind N Naik & Mohammad Javed Ali (2019) Lacrimal drainage anomalies in Rubinstein–Taybi syndrome: case report and review of literature, Orbit, 38:4, 335-337, DOI: 10.1080/01676830.2018.1515961

To link to this article: https://doi.org/10.1080/01676830.2018.1515961

ORBIT
2019, VOL. 38, NO. 4, 335–337
https://doi.org/10.1080/01676830.2018.1515961

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CASE REPORT

Lacrimal drainage anomalies in Rubinstein–Taybi syndrome: case report and review of literature

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ABSTRACT

Rubinstein–Taybi syndrome is a rare multisystem disorder characterized by broad thumbs and first toes, short stature, microcephaly, delayed milestones, beak nose, and hypertelorism. Lacrimal drainage anomalies are not uncommon in this syndrome. We present a patient with Rubinstein–Taybi syndrome with bilateral congenital nasolacrimal duct obstruction and left-sided grossly dilated nasolacrimal duct.

ARTICLE HISTORY
Received 13 July 2018
Accepted 15 August 2018

KEYWORDS
Rubinstein–Taybi syndrome; lacrimal; CNLDO; nasolacrimal duct; ocular

Introduction

Rubinstein–Taybi syndrome (RTS) or broad thumb-hallux syndrome is a rare autosomal dominant disorder characterized by broad thumbs and first toes, microcephaly, mental retardation, short stature, and facial features including high arched eyebrows, anti-mongoloid slants, hypertelorism, and ear anomalies.[1–7] Strict criteria for diagnosis of RTS are not defined, and typical clinical features are commonly relied upon. The prevalence of congenital lacrimal anomalies in RTS patients range from 10–37% in two large systemic series.[1,2] Anomalies recognized include congenital nasolacrimal duct obstruction, nasolacrimal duct stenosis, punctal inversion, punctal malposition, common canalicular obstruction, lacrimal fistula, and pediatric acute dacryocystitis.[1–10] We present a case of RTS with bilateral congenital nasolacrimal duct obstruction (CNLDO) and left-sided grossly dilated nasolacrimal duct.

Case report

A male infant of 7 month age presented with bilateral epiphora since birth associated with discharge in the left eye (Figure 1A). The patient was a known case of RTS with typical features of broad thumbs, broad hallux, microcephaly, and characteristic facial features of high arched eyebrows and palate, beak nose, and ear anomalies (Figure 1B). The patient was under the care of cornea service for a left dense corneal opacity. Examination of the lacrimal drainage system revealed bilateral raised tear meniscus with left-sided matting of lashes and periocular dried discharge (Figure 1A). The puncta and canaliculi were normal. Irrigation was suggestive of bilateral CNLDO. Nasal endoscopic evaluation showed a normal right inferior meatus; however, the left inferior meatus showed a gross dilatation of the nasolacrimal duct (Figure 1C). Probing was performed under endoscopic guidance and the nasolacrimal duct could be successfully recanalized. The retained discharge in the left nasolacrimal duct could be evacuated (Figure 1D). Postoperatively there was resolution of epiphora and discharge and a corneal intervention was planned for the left eye.

Discussion

Syndromic CNLDOs are not very common, and RTS accounts for an uncommon cause even amongst the syndromes.[4] RTS is linked to a mutated CREBBP gene on chromosome 16 in most cases and to mutations in the EP300 gene in few cases, while in others the cause is not defined.[2,7] Affected individuals are more susceptible to tumors, especially leukemias and lymphomas.

While there are constellations of numerous systemic and ocular features, the current discussion would focus on the congenital lacrimal drainage anomalies in RTS. Rubinstein[1] reviewed a total of 571 cases from 40 countries who were reported between 1957 to 1988. Of these, 80 patients were reported to have nasolacrimal duct obstruction (NLDO), reflecting an association in 37% of the cases. However, further details as regards

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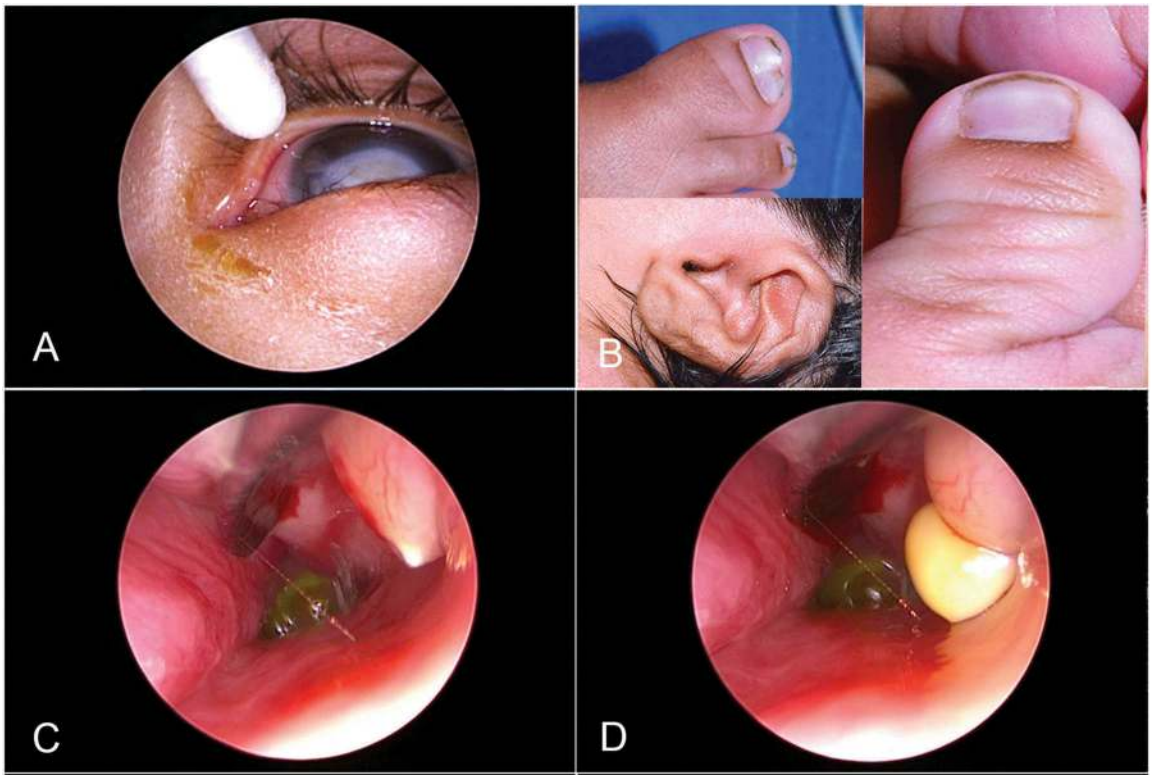

Figure 1. Clinical image of the left eye showing the matting of lashes, dried discharge, and a normal upper punctum (A). Clinical image showing a broad hallux (B, left upper), ear anomaly (B, left lower), and broad thumb (B, right). Endoscopic image of the left inferior meatus demonstrating a gross dilated nasolacrimal duct. Also note the tip of the probe within the nasolacrimal duct (C). Endoscopic image showing evacuation of retained discharge from the dilated nasolacrimal duct following a successful recanalization (D).

to their specific presentations and management were missing. Van Genderen et al.[2], a decade later, published their review of 207 patients of which 10% (21/207) showed NLDO. Amongst patients with ocular abnormalities (117/207), 18% (21/117) showed NLDO. About two-thirds of their own patients had bilateral presentation. Using the national parents support group, Stevens et al.[3] surveyed 50 patients of RTS using a questionnaire and found 15 of these patients to have NLDO. Since all these were systemic reviews, details of these NLDOs were not given. The current case reported endoscopic details of biltateral CNLDO, especially a grossly dilated nasolacrimal duct, which could be secondary to increased retention and stasis of secretions.

Filippi[5] reported two cases of CNLDO amongst the seven they studied. One of them presented with bilateral acute dacryocystitis and had undergone a probing and later a bilateral dacryocystotomy at the age of 9 years. Roy et al.[6] presented a 27-month-old child with several past episodes of acute dacryocystitis which was managed by conservative treatment and probing. The patient in addition had punctal inversion and common canalicular obstruction. The other patient also had bilateral CNLDO with left corneal leukoma, just like the present case, but the child died before any intervention. Marabotti et al.[7] reported one case of RTS in a 9-year-old girl who presented with bilateral acute dacryocystitis. The patient had multiple such attacks in the last 5 years. Dacryocystography revealed a grape-bunch like appearance of the lacrimal sacs. Although the patient was successfully treated with a bilateral external dacryocystorhinostomy (DCR), the surgeon needs to be aware of the thicker bones during osteotomy and brittle lacrimal sacs with potential adhesions to periosteum. The current case could be managed with probing alone in spite of a grossly dilated nasolacrimal duct and did not require a DCR. We believe a good endoscopic guidance during probing in an RTS patient can help address associated issues and most children may not need a DCR.

In summary we describe a neonatal presentation of a patient with RTS with bilateral CNDLO and a grossly dilated left nasolacrimal duct. The patient was successfully treated with an endoscopy-guided probing. Any encounter with a RTS patient should alert the clinician to search for associated congenital lacrimal anomalies and initiate appropriate management.

Disclosure statement

Dr Ali receives royalties from Springer for his treatise "Principles and Practice of Lacrimal Surgery" and "Atlas of Lacrimal Drainage Disorders". Other authors have none.

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