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Case Report

Clinicopathological Correlation in Reis-Bücklers's Dystrophy

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Abstract

Reis-Bückler's Corneal Dystrophy (RBCD) is a rare corneal dystrophy which mainly affects the Bowman's layer leading to its disintegration and eventually corneal opacification. Various investigative modalities are needed to establish the diagnosis of RBCD. We report a patient in whom various clinical and pathological tests were performed to establish the diagnosis of RBCD. We will also describe the clinical course of the same patient. Treatment modality for the same patient is described.

Key words: Reis-Bückler's Corneal Dystrophy (RBCD), Anterior Segment OCT (AS OCT), Phototherapeutic Keratectomy (PTK)

INTRODUCTION

- Reis-Bückler's corneal dystrophy is an autosomal dominant progressive dystrophy initially described by Reis in 19171 and later by Bücklers2 in 1949.
- The transforming growth factor beta induced gene (TGFBI, OMIM 601692), located on chromosome 5q31 has been linked with RBCD3. Thus, genetic analysis helps in confirmation of the diagnosis.
- Anterior segment OCT can help localizing the exact depth of involvement of corneal layers and can help in planning further management.
- Pathological examination of corneal button can help confirm the diagnosis by showing disintegration of Boman's membrane, subepithelial and anterior stromal eosinophilic deposits that stain positively with Masson's trichome.

CASE REPORT

- A 35-year-old male presented to cornea outpatient department with chief complaints of progressive diminution of vision in both eyes and photophobia since childhood. Visual acuity was finger counting close to face in both eyes.
- On clinical examination, cornea had white rod and granule-shaped deposits involving the anterior layers of cornea sparing the peripheral cornea and limbus [Figure 1]. Clinically, Reis-Bücklers dystrophy, granular dystrophy, and Thiel-Behnke dystrophy were suspected differential diagnosis. The patient had no other significant ocular or systemic abnormality.
- The patient was advised corneal topography, pachymetry, anterior segment-optical coherence tomography (AS-OCT), and B-scan of both eyes. AS-OCT revealed homogeneous, hyper-reflective deposits at the level of Bowman's membrane, and anterior stroma [Figure 2]. B-scan revealed early cataractous changes in both eyes.
- Phototherapeutic keratectomy (PTK) was done in the right eye 1 month later, but no visual improvement was seen. Vision subsequently improved to 20/60 at the 3rd month follow-up after cataract extraction. Six months later, PTK was performed in the left eye and BCVA improved to 20/100.
- Two years later, the patient presented with worsening of vision to hand movements in both eyes due to recurrence of dystrophy and was planned for deep anterior lamellar keratoplasty in the left eye.
- Corneal button sent for histopathological examination revealed discontinuity of bowman's membrane along with abnormal subepithelial and anterior stromal eosinophilic deposits that stained positively with Masson's trichome but did not stain with Alcian Blue or Congo Red, thus confirming the diagnosis [Figure 3].
- Postoperatively, vision of the patient improved to 20/100 at 3 months follow-up in the left eye.

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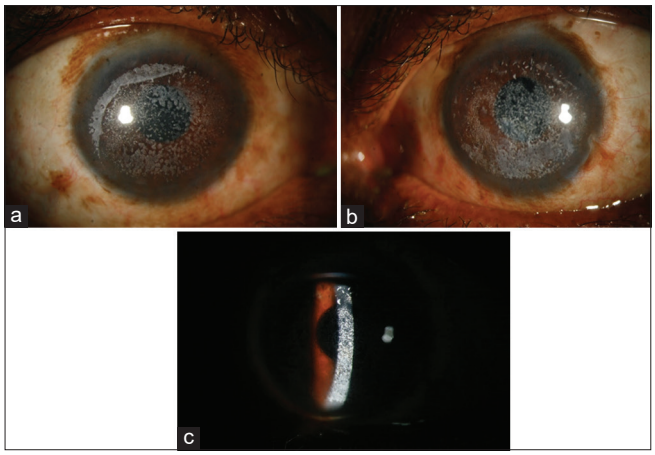

Figure 1: Slit-lamp photograph (a and b) (Diffuse illumination) – Deposits are seen involving the central and paracentral zones of cornea in both the eyes, sparing the peripheral cornea and limbus. Intervening clear spaces between the deposits are seen in central cornea but deposits are seen coalescing in superotemporal region and inferior portions of paracentral right and left cornea, respectively (c) Slit illumination showing characteristic rod and granule-shaped deposits in the region of anterior stroma sparing the posterior layers

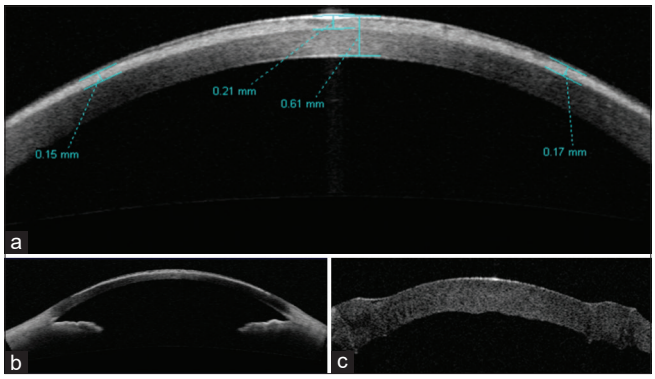

Figure 2: AS-OCT demonstrating (a) Hyper-reflective deposits affecting up to depths of 210 micron and 170 micron in central and mid-peripheral cornea, respectively. (b) Hyper-reflective and homogeneous deposits at the level of Bowman's membrane and anterior stroma sparing the peripheral cornea (c) Post-PRK decrease in hyper-reflective deposits and thinning of central cornea in the left eye

DISCUSSION

- Reis-Bücklers corneal dystrophy is an autosomal dominant progressive dystrophy initially described by Reis in 1917[1] and later by Bücklers[2] in 1949.
- The transforming growth factor beta-induced gene (TGFBI, OMIM 601692), located on chromosome 5q31, has been linked with RBCD.[3] Thus, genetic analysis helps in confirmation of the diagnosis.
- RBCD is clinically characterized by confluent geographic opacities at the level of Bowman's layer and histopathologically by band-shaped granular Masson-positive subepithelial deposits.[4]

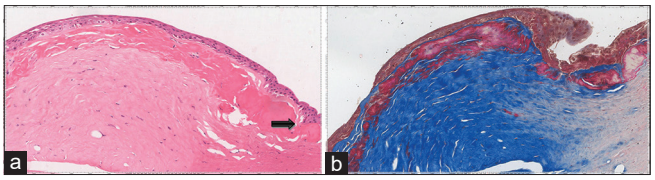

Figure 3: Histopathological findings of RBCD (a) Photomicrograph shows irregularly thickened squamous epithelium with focal saw tooth appearance (black arrow). Bowman's membrane is fragmented and subepithelial and anterior stroma shows amorphous eosinophil hyaline material deposits. Rest of stroma is densely fibrosed; H&E stain (10×) (b) Photomicrograph of Masson's trichome stains demonstrate diffuse Bowman's membrane loss, subepithelial band of fibrosis along with brilliant red deposit in subepithelial and superficial stroma. Rest of stroma is densely; fibrosed. Masson's Trichrome stain (10×)

- On light microscopy, characteristics of RBCD include-
  - Epithelium – degeneration, thinning, edema
  - Bowman's layer – absent and is replaced by band-shaped granular eosinophilic deposits which stain positive with Masson's trichome.
- On electron microscopy, crystalloid, rod-shaped bodies at the level of Bowman's membrane are seen.
- On confocal microscopy, highly reflective amorphous deposits on epithelial basal layer and anterior stroma can be noted.[5]
- Anterior OCT can help localize the precise level of deposits and help us judge the reflectivity and type of dystrophy. Thus, AS-OCT is useful modality for the diagnosis and management of RBCD.[6]
- Significant visual loss can occur, and recurrences are common after keratectomy and keratoplasty. Hence, multiple interventions may be needed.

CONCLUSION

AS-OCT can help localize the precise level of deposits in RBCD. Histopathology and genetic analysis remain the gold standard for diagnosis.

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