**Category 1**

**PATHOLOGY** Light microscopy shows subepithelial and stromal amyloid deposits. Disruption of epithelial tight junctions leads to abnormally high epithelial permeability. Confocal microscopy shows irregular, elongated epithelial cells with large accumulations of brightly reflective material noted within or beneath the epithelium and within the anterior stroma. Amyloid deposition is noted in the basal epithelial layer on transmission electron microscopy. See also Chapter 8 for a more complete discussion of amyloidosis.

**CLINICAL PRESENTATION** Onset occurs in the first to second decade of life with groups of multiple small nodules (mulberry configuration) or with subepithelial lesions that may appear similar to those of band keratopathy (Fig 7-5A, B). The lesions are visible on fluorescein staining. There is a significant decrease in vision, with photophobia, irritation, and tearing, as well as progression of protruding subepithelial lesions. Superficial vascularization is often seen. Stromal opacification or larger nodular lesions (kumquat-like lesions) may develop (Fig 7-5C).

**MANAGEMENT** The lesions recur within a few years following superficial keratectomy, lamellar keratoplasty (LK), or penetrating keratoplasty (PK). Soft contact lenses are effective in reducing the abnormal epithelial permeability in an effort to decrease recurrence.


![Figure 7-5](reproduced_with_permission_from_weiss_js_meller_hu_aldave_aj_et_al_ic3d_classification_of_corneal_dystrophiesEdition_2_Cornea_2015_34(2)_130)
Epithelial–Stromal TGFBI Dystrophies

Reis-Bücklers corneal dystrophy (RBCD)

Alternative names  Corneal dystrophy of Bowman layer type 1 (CDB1), atypical granular corneal dystrophy

Inheritance  AD

Category  1

**PATHOLOGY**  On light microscopy, the Bowman layer is disrupted or absent and replaced by a sheetlike connective tissue layer with granular deposits that stain red with Masson trichrome stain. Transmission electron microscopy shows subepithelial electron-dense, rod-shaped bodies, which are immunopositive for the TGFBI protein, keratoepithelin. Electron microscopy is needed to histologically distinguish RBCD from Thiel-Behnke corneal dystrophy (TBCD), which has curly fibers (see the next section). On confocal microscopy, distinct deposits are found in the epithelium and Bowman layer. The basal epithelial cell layer shows high reflectivity associated with small granular deposits without any shadows (Fig 7-6A), which would be typical of TBCD. The Bowman layer is replaced with highly reflective irregular material. Greater hyperreflectivity is seen at the Bowman layer in RBCD than in TBCD.

![Figure 7-6](image)