

Corneal Dystrophies

A PRIMER ON CORNEAL DYSTROPHIES



Diagnosis and management depend on the location of the dystrophy and other factors.

BY TRENNDA L. RITTENBACH, OD

Corneal dystrophies are a result of abnormal cellular metabolism. This group of noninflammatory inherited diseases can affect the transparency of the cornea. As primary eye care providers, optometrists are uniquely positioned to diagnose and manage patients with corneal dystrophies. This article presents some tips for diagnosis and management of these conditions, based on my own experience with them.

ANTERIOR CORNEAL DYSTROPHY

The most common hereditary anterior corneal dystrophy is epithelial basement membrane dystrophy (EBMD), also known as *map-dot-fingerprint dystrophy* or *anterior basement membrane dystrophy*. EBMD is caused by dysfunction in the basal epithelial cells, resulting in a secretion of abnormal basement membrane that extends into the epithelium and an accumulation of fibrillogranular material between

Bowman layer and the basement membrane and within the epithelium.¹ EBMD tends to affect patients of all ethnicities and both sexes equally.^{2,3}

Diagnosis

The main symptoms of EBMD are typically ocular irritation, blurred vision, glare, photophobia, and pain. Recurrent corneal erosion (RCE) in EBMD can cause sharp pain, usually in the morning upon waking. RCE usually occurs after a traumatic abrasion in patients with or without EBMD. The pain can range from mild and lasting only a few minutes to severe and lasting hours or days or until treatment is initiated. Surface irregularity and rapid tear breakup time at the area of slightly elevated epithelium overlying the map-like changes, fingerprint lines, and/or microcysts in EBMD may reduce visual acuity. Retroillumination while the pupil is dilated is one of the easiest ways to view EBMD (Figure 1).³

Treatment

Treatment consists of the following³⁻⁵:

- topical lubricants;
- hypertonic sodium chloride (NaCl) drops during the day;
- lubricating ointment or NaCl ointment at bedtime;

Photos courtesy of Jacob Lang, OD, FAAO

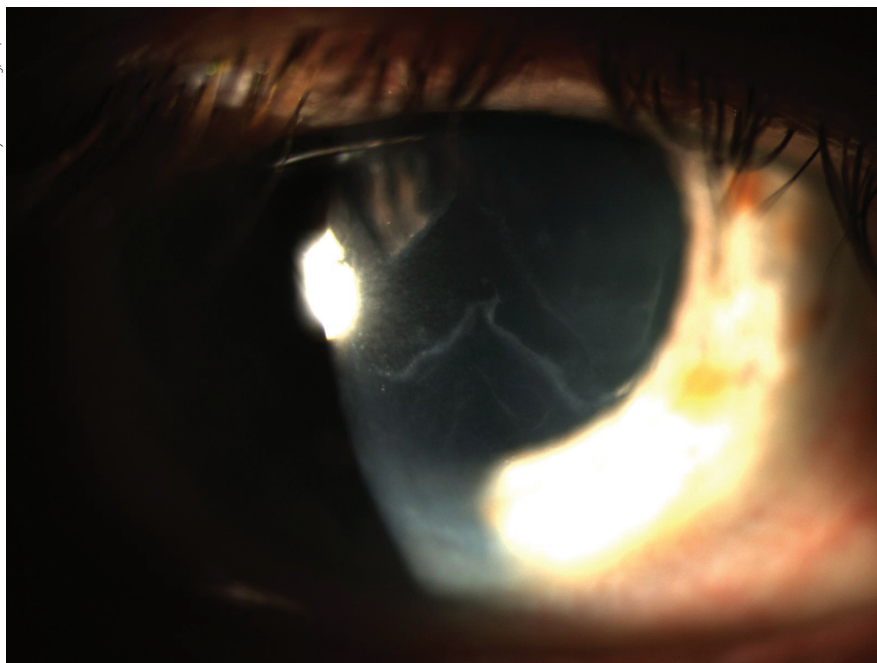


Figure 1. EBDM viewed using retroillumination.

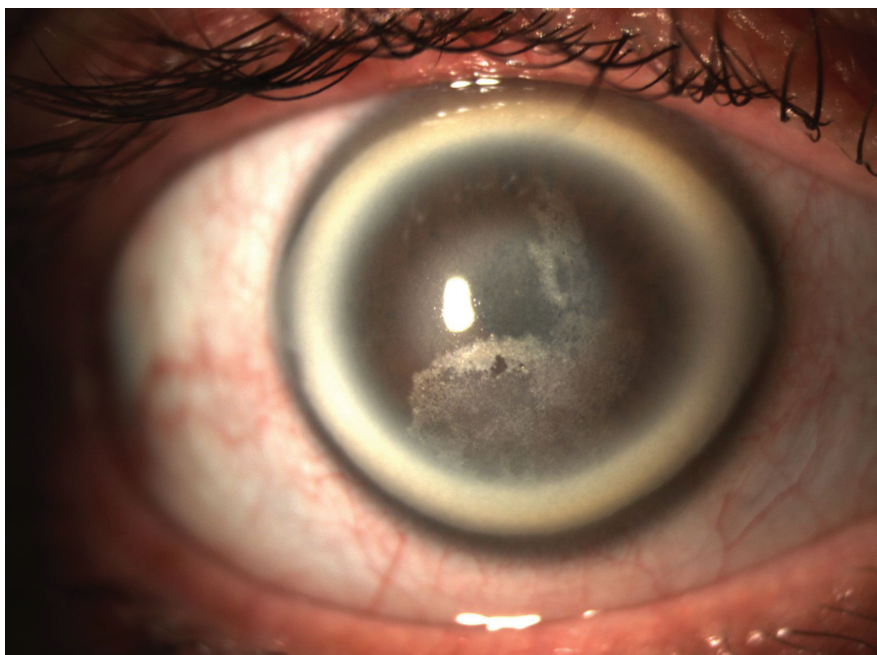


Figure 2. Crystal deposits seen in an eye with SCD.

- bandage soft contact lenses for RCE with an antibiotic drop administered over the lens;
- oral matrix metalloproteinase inhibitors;
- autologous serum drops;
- amniotic membrane for RCE;
- anterior stromal puncture;
- mechanical debridement of the

- loose epithelium for RCE, with or without diamond-dusted burr polishing; and/or
- phototherapeutic keratectomy (PTK) ablation.

STROMAL DYSTROPHIES

Dystrophies affecting the stroma include lattice, granular, macular, and

Schnyder corneal dystrophies. These conditions affect patients of all ethnicities and both sexes equally.

Lattice corneal dystrophy is characterized by a network of multiple branching refractile lines or spindle-like corneal opacities from amyloid deposition in the anterior and deep stromal layers. Granular corneal dystrophy is made up of hyaline depositions, and it is characterized by multiple snowflake-like, distinct, granular, white opacities in the central anterior stromal layer. Both lattice and granular corneal dystrophies result from mutations in the transforming growth factor beta-induced (*TGFB1*) gene.⁶

Macular corneal dystrophy is caused by mutations in the corneal carbohydrate sulfotransferase 6 (*CHST6*) gene. Clinically, macular corneal dystrophy is characterized by diffuse stromal clouding from an accumulation of glycosaminoglycans and central corneal thinning.⁷

Schnyder corneal dystrophy (SCD) is a rare, progressive corneal disorder caused by abnormal accumulations of lipid and cholesterol. The condition results in corneal opacification and premature or early arcus. Patients with SCD may also have subepithelial crystal deposition (Figure 2). Familial hypercholesterolemia is the most common lipoprotein abnormality in patients with SCD, with some reports suggesting that up to two-thirds of SCD patients have the disorder.⁸ The causative gene for SCD is *UBIAD*.^{8,9}

Diagnosis

Patients with stromal dystrophies may be asymptomatic, or they may present with symptoms including decreased VA, photophobia, and ocular irritation or pain from corneal erosions. Patients with SCD generally experience decreases in visual acuity and corneal sensation and increased corneal clouding. Arcus and mid-peripheral haze typically develop as these patients age.⁹

AT A GLANCE

- EBMD, also known as map-dot-fingerprint dystrophy or anterior basement membrane dystrophy, is the most common hereditary anterior corneal dystrophy.
- Stromal dystrophies include lattice, granular, macular, and Schnyder corneal dystrophies.
- Fuchs endothelial corneal dystrophy is the most common posterior corneal dystrophy, followed by posterior polymorphous dystrophy.

Anterior segment OCT and corneal topography can be helpful for diagnosing and monitoring stromal corneal dystrophies.

Treatment

Treatment for stromal dystrophies is dictated by visual impairment. PTK can remove opacities from the anterior corneal stroma, but it cannot remove stromal haze.¹⁰ Penetrating keratoplasty has largely been replaced by other surgical options. For example, deep anterior lamellar keratoplasty removes the affected corneal layers but leaves the patient's native endothelium intact, significantly reducing the rate of graft rejection.

ENDOTHELIAL DYSTROPHIES

Fuchs endothelial corneal dystrophy is the most common posterior corneal dystrophy, followed by posterior polymorphous dystrophy (PPMD).

Fuchs dystrophy is characterized by corneal guttae (excrescences on or within Descemet membrane) and a reduction in endothelial cell density, with pleomorphism and polymegathism best seen on specular microscopy. The condition is more common in women than men, and it usually manifests in the fourth decade of life. Disease progression is characterized by an increase in the number and size of the guttae. As the disease advances, endothelial cell function decreases, leading to corneal thickening and

edema. Epithelial bullae may appear and rupture due to stromal edema, leading to severe pain.¹¹

PPMD is a bilateral but often asymmetric, nonprogressive dystrophy that is caused when the corneal endothelium produces an aberrant basement membrane. Clinically, PPMD is characterized by a Swiss cheese- or moon-like pattern. The eye may exhibit a peripheral ring, a focal wedge, or a band of parallel lesions with horizontal or vertical vesicular changes.

Diagnosis

Most patients with early Fuchs dystrophy are asymptomatic, but they may develop symptoms as the disease advances. They will typically experience blurry vision upon waking, decreased contrast sensitivity, ocular discomfort, and/or photophobia.

Patients with PPMD are usually asymptomatic. The condition is rarely severe enough for patients to develop corneal edema, corectopia, or angle-closure glaucoma, caused by the migration of epithelialized PPMD endothelial cells across the anterior chamber angle.¹²

Treatment

Management of patients with Fuchs dystrophy and PPMD depends on their symptoms and objective findings. Hypertonic saline drops or ointment may improve symptoms. Surgical intervention may be

warranted in severe Fuchs dystrophy. Penetrating keratoplasty has largely been replaced by partial-thickness corneal transplantation procedures such as Descemet stripping automated endothelial keratoplasty (DSAEK) and Descemet membrane endothelial keratoplasty (DMEK). DSAEK and DMEK are similar procedures. DSAEK involves the transplantation of donor stromal tissue, Descemet membrane, and endothelial cells. In DMEK, only donor Descemet membrane and endothelial cells are transplanted.

CONCLUSION

Corneal dystrophies will present themselves in an optometric examination, so optometrists should be able to identify and manage these conditions. It's also a good idea to establish a relationship with a cornea specialist, if you haven't already, and to refer patients with suspected or confirmed corneal dystrophies to him or her when they are in need of surgery. ■

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