Corneal Endothelial Dystrophies

Arun Joseph, MD
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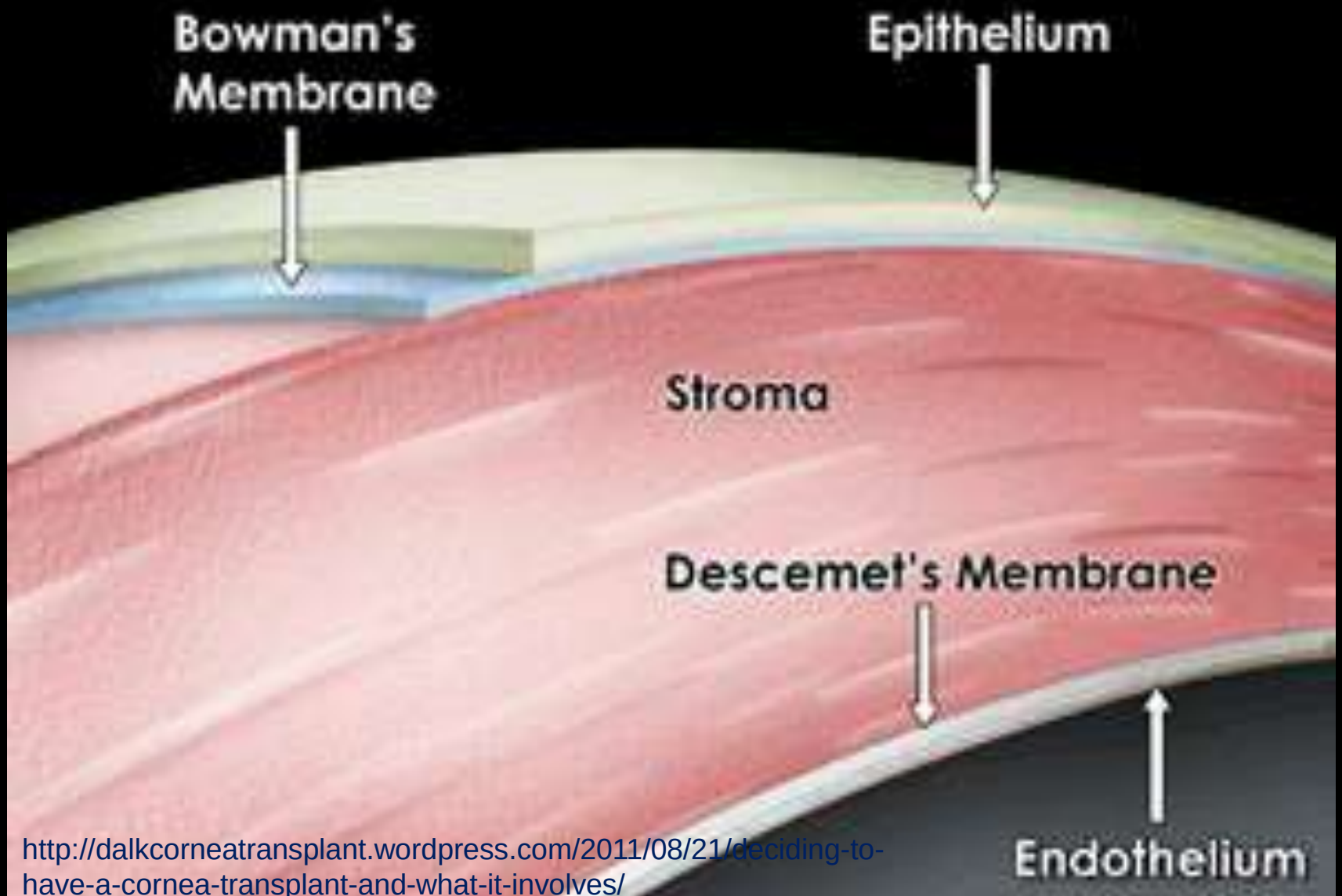
Corneal endothelium

- Single layer of mitochondria rich cells on inner surface of cornea
- Embryologically derived from neural crest
- Attached to the other layers of cornea through an acellular layer of collagen, Descemet's membrane
- Governs fluid and solute transport across posterior surface of cornea
  - Maintains cornea in the dehydrated state required for optical transparency

Cornea



http://dalkcorneatransplant.wordpress.com/2011/08/21/deciding-to-have-a-cornea-transplant-and-what-it-involves/

Endothelial dystrophies

- Fuchs' endothelial dystrophy
- Posterior polymorphous dystrophy
- Congenital hereditary endothelial dystrophy

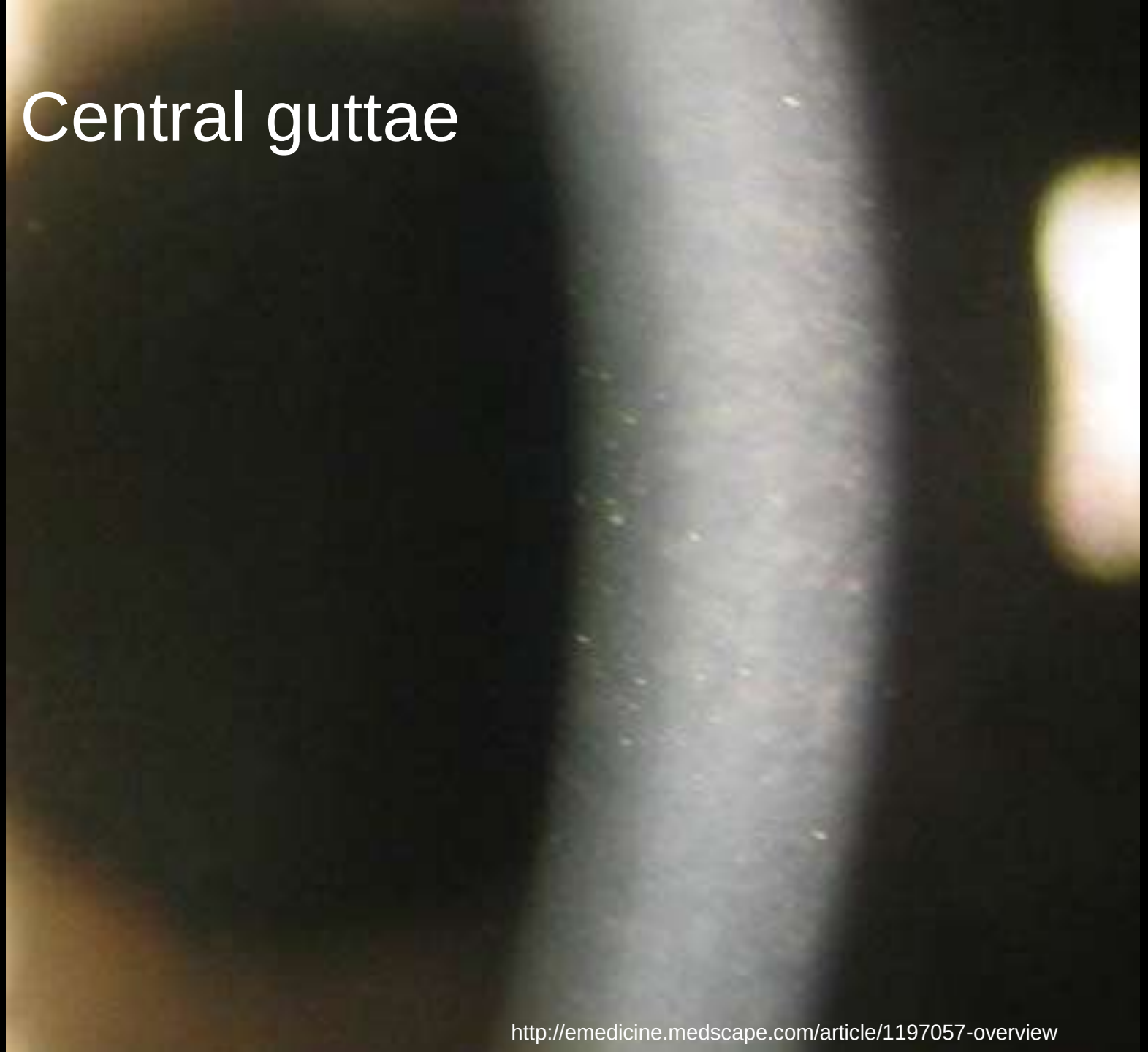
Endothelial dysfunction
➔ Corneal edema
➔ Visual compromise and pain

Fuchs’ Endothelial Dystrophy

Fuchs' dystrophy

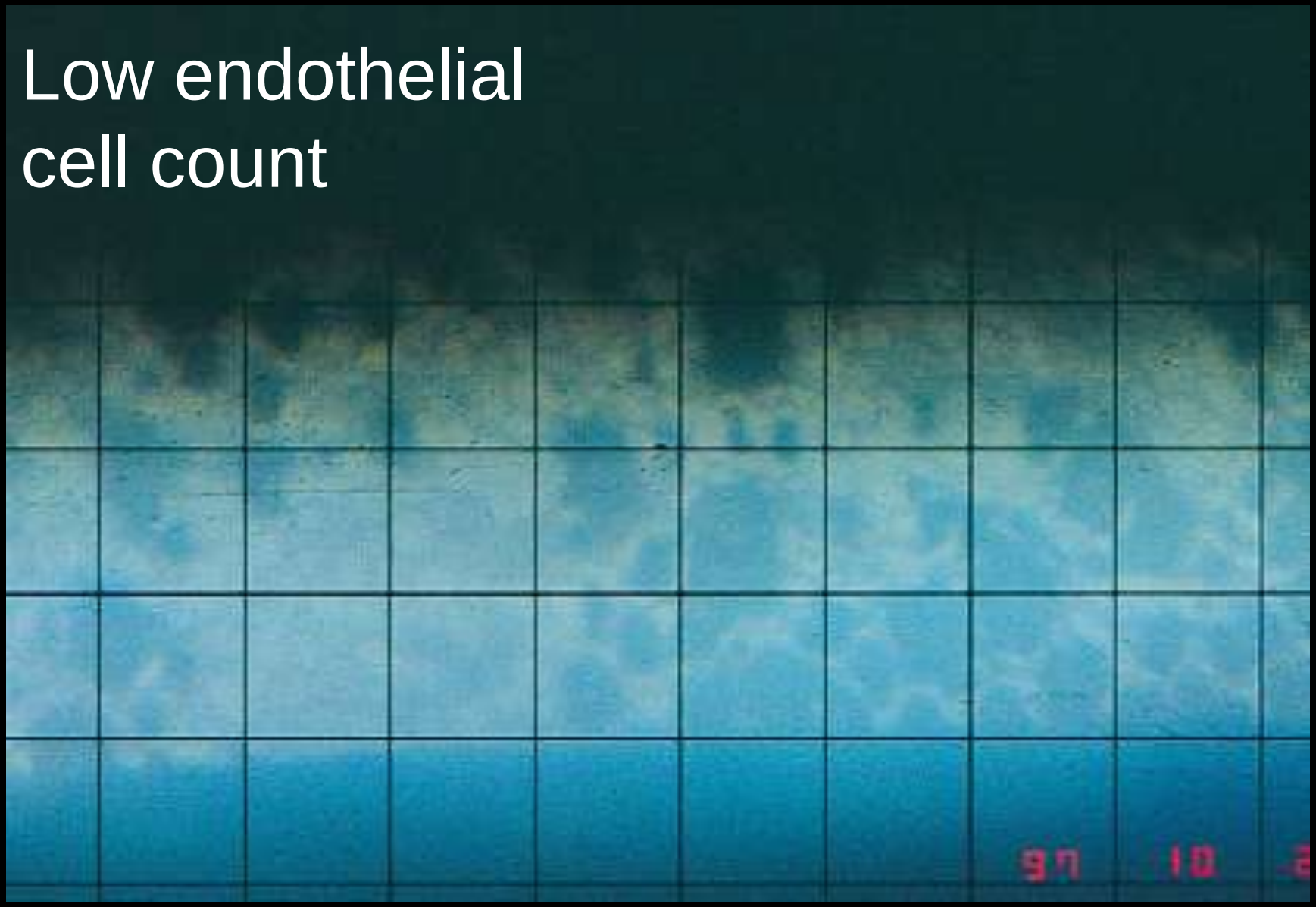
- Bilateral, noninflammatory, degenerative disease of the endothelium with reduced Na/K pump activity leading to accumulation of focal outgrowths called guttae, corneal edema, and loss of vision
- Autosomal dominant
- Onset in 5-6th decade of life
- F >> M
- Corneal findings: central guttae, stromal thickening, Descemet's folds
- Associated with narrow angles and glaucoma

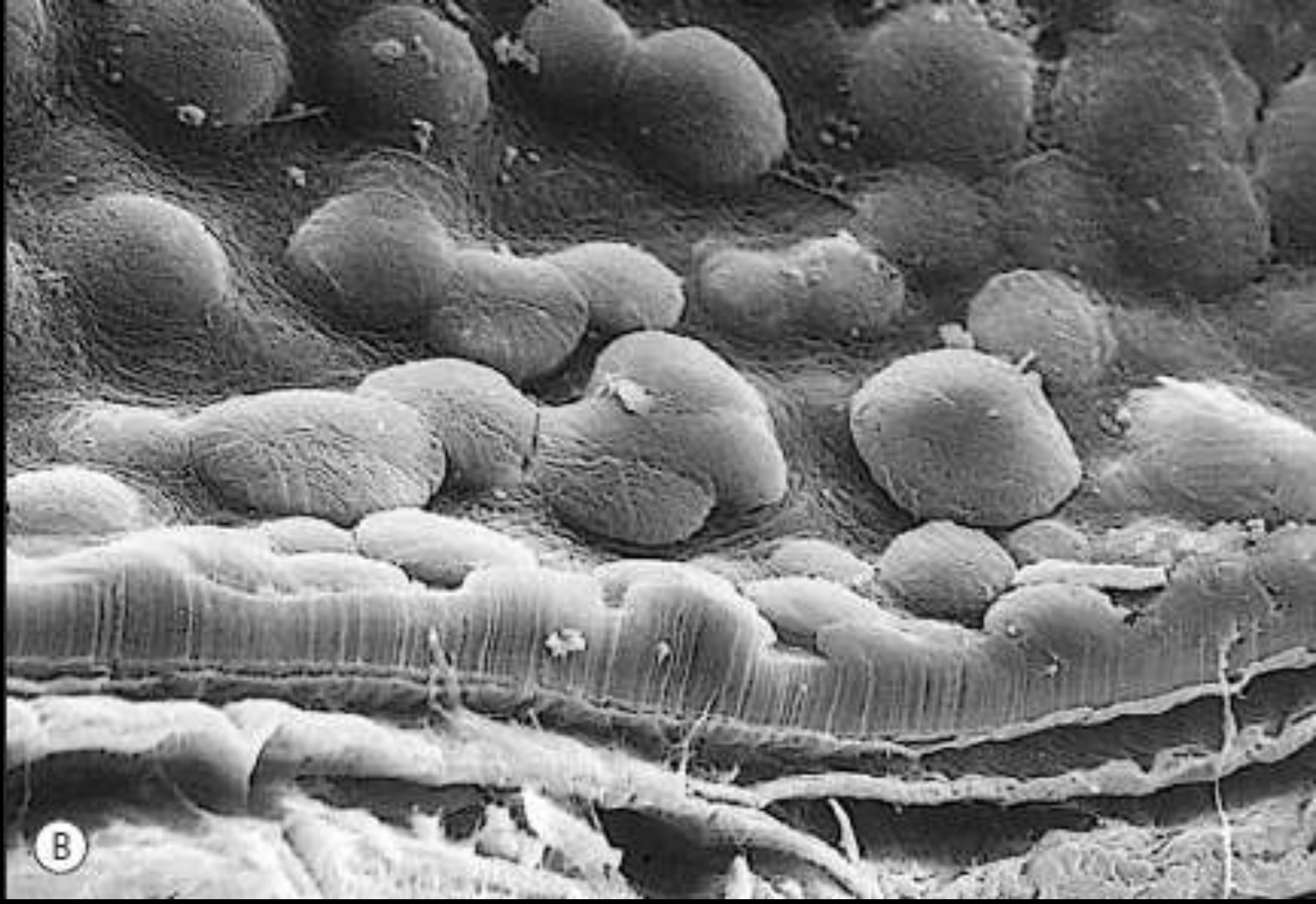
Central guttae



http://emedicine.medscape.com/article/1197057-overview

Low endothelial cell count

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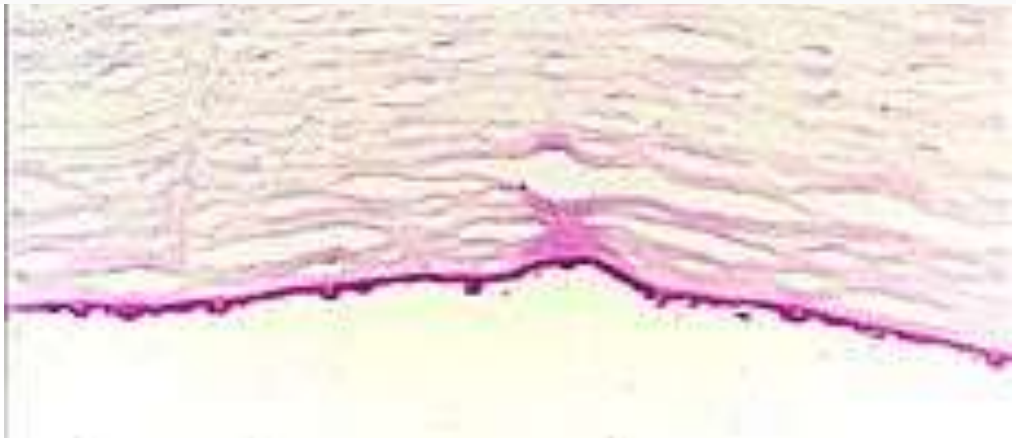


Surgical considerations with Fuchs'

- Visually significant cataract and borderline corneal function
  - CCT < 620 and no evidence of stromal edema ➔ cataract surgery alone recommended
  - CCT > 620, frank stromal edema, or 10% difference in corneal thickness in morning compared to evening ➔ combined cataract surgery with PK/DSEK

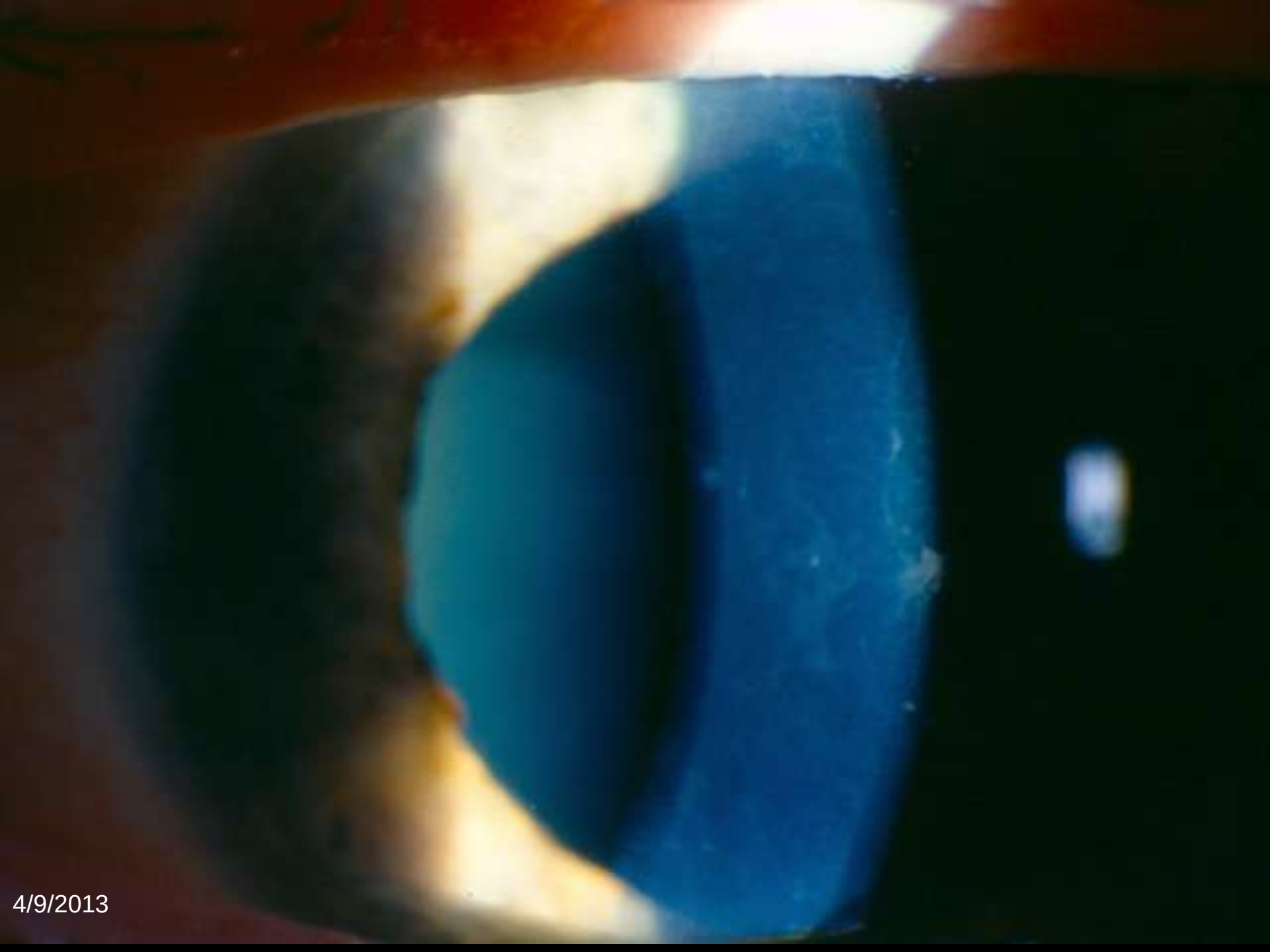
Surgical considerations with Fuchs'

- Corneal edema requiring transplantation with mild-moderate cataract
  - Retrospective studies indicate that most patient undergoing corneal transplant eventually require cataract surgery
  - Combined transplantation and cataract surgery recommended to avoid increased costs and delay in visual rehabilitation

Posterior Polymorphous Dystrophy

PPMD

- Bilateral, nonprogressive disease thought to be 2/2 focal metaplasia of endothelial cells into a population of aberrant keratinized epithelial-like cells
- Autosomal dominant
- Onset in 2-3rd decade of life
- Corneal findings: vesicular changes, endothelial band lesions, irregular placoid opacities of the posterior corneal surface and multilayered endothelium (irregular scalloped edges)
- Associated with PAS, iris atrophy, corectopia, and glaucoma (overlap with ICE syndrome)
- Most patients asymptomatic



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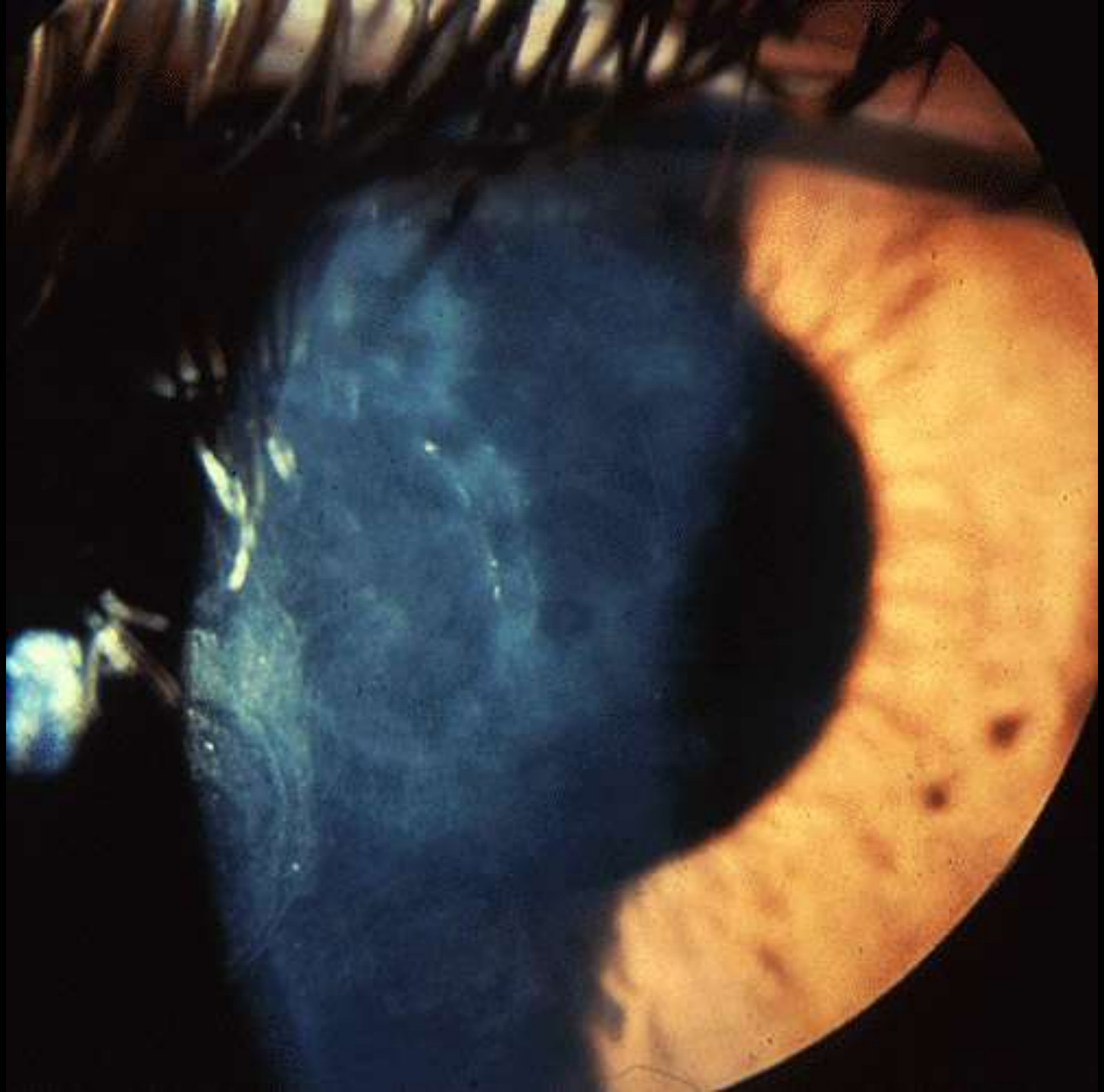

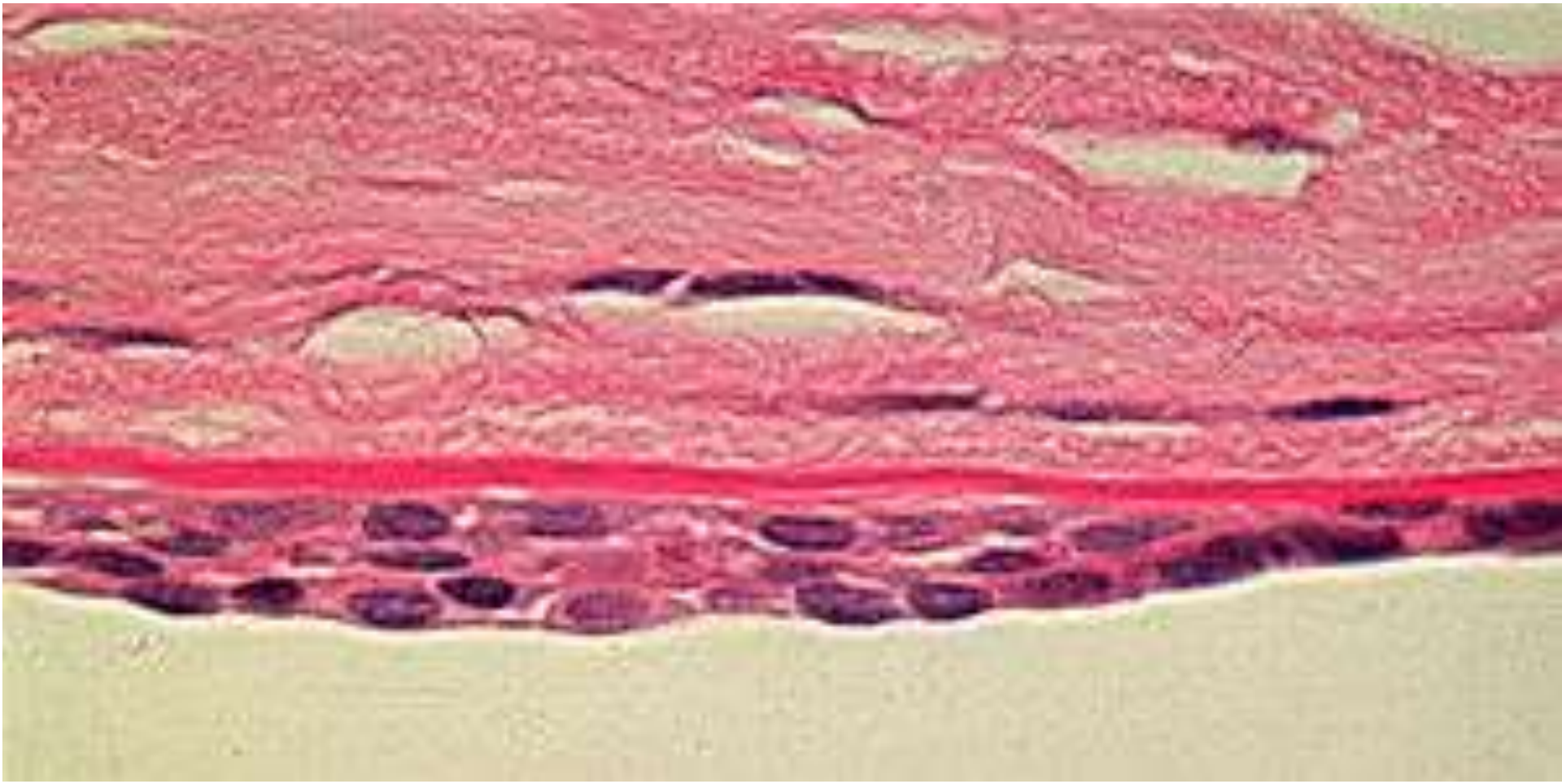

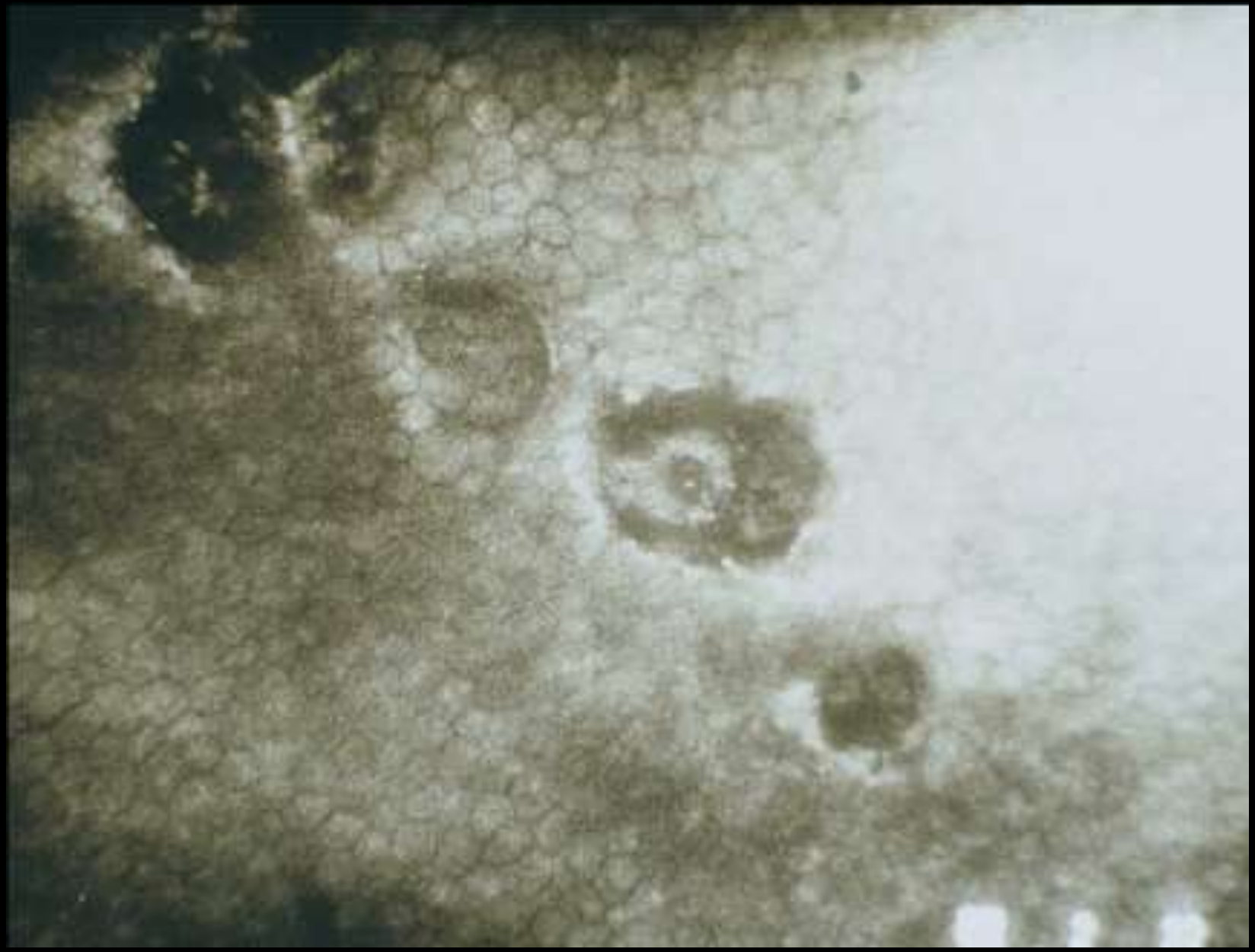

Congenital Hereditary Endothelial Dystrophy

CHED1

- Autosomal dominant
- Diffuse thickening and lamination of Descemet's membrane with sparse, atrophic endothelial cells
- Occurs congenitally or during 1-2nd year of life
- Diffuse corneal clouding and thickening that slowly progresses over 1-10 years

CHED2

- Autosomal recessive
- Diffuse thickening and lamination of Descemet's membrane with sparse, atrophic endothelial cells
- Congenital nonprogressive disease, but more severe than CHED1
- Diffuse corneal clouding and thickening often associated with nystagmus

Treatment

- Fuchs'
  - Early: conservative, hypertonic ointment, IOP lowering
  - Advanced: transplantation (fairly good prognosis)
- PPMD: Most patients asymptomatic.
  - Early symptomatic: conservative
  - Advanced: transplantation (prognosis related to presence of PAS and glaucoma, higher chance of recurrence)
- CHED
  - CHED1: PK in advanced cases
  - CHED2: PK usually required given severity

References

- Yanoff, et al.
- BCSC