Overview of Corneal Dystrophies

Brad Sutton, O.D., F.A.A.O.
Indianapolis Eye Care Center
IU School of Optometry
brsutton@indiana.edu

- No financial disclosures

Corneal Dystrophies

- In general, corneal dystrophies are…….
  - Bilateral and relatively symmetric
  - Progressive
  - Non-inflammatory
  - They often present in the first two decades of life
  - Most gene loci have been identified precisely

Major corneal dystrophies

- Epithelial: EBMD, Meeseman's, Reis-Buckler
- Stromal: Granular, Macular, Lattice, (and Avellino)
- Endothelial: Fuch's, PPD, CHED

- Top three for each layer

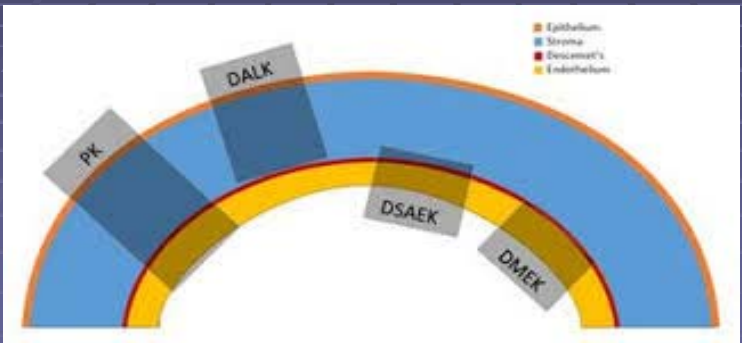
Avagen genetic testing from Avellino

- In office collection kit via cheek swab
- Cost of about $300, billed to doctor, who then collects from the patient. May sometimes qualify for insurance filing
- Detects the following based up TGFBI analysis……..
- Stratisfies keratoconus risk as L, M, H, all others below as Y/N
- EBMD
- Granular
- Avellino
- Lattice I & Lattice IIIA
- Reis-Buckler's
- Schnyder's

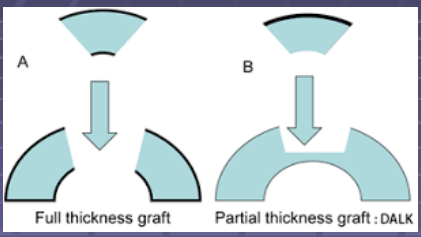
Types of corneal transplants

- DALK
- Full thickness PKP
- DSAEK
- DMEK

Types of corneal transplants (helio.com)



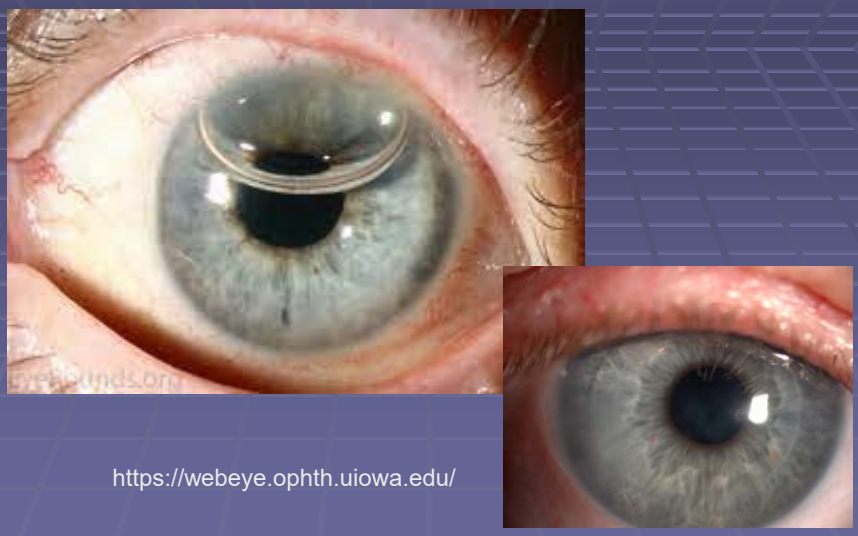
DALK and PKP



Ocvermont.com

Jirehdesign.com

DSAEK and DMEK



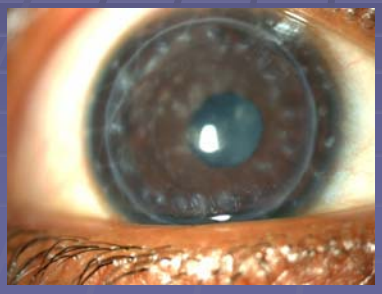
https://webeye.ophth.uiowa.edu/

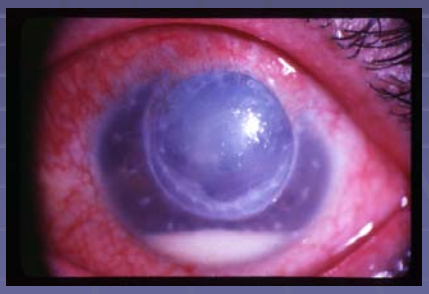
Types of corneal transplants

- In general, partial thickness grafts, compared to full PKP,……..
- Have shorter recovery time
- Have more predictable / better refractive outcomes
- Have lower rates of rejection

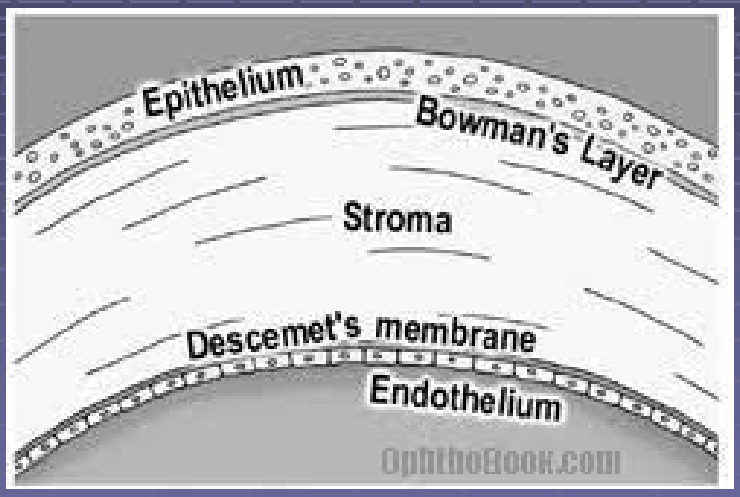
Corneal transplants

- DSAEK / DMEK can be performed on failed PKP
- Graft failure
- Graft rejection





Epithelial Dystrophies



EBMD

- Also known as map-dot-finger print, ABMD, and others
- May have AD inheritance pattern but unclear
- Relatively non-progressive
- Visible as early as second decade: prominent in middle age

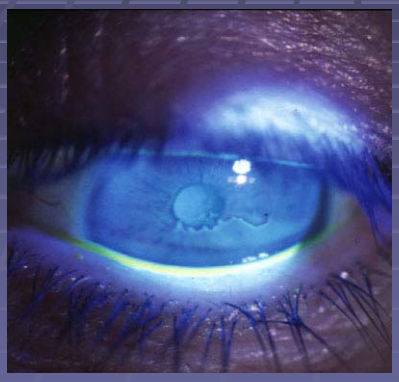
EBMD

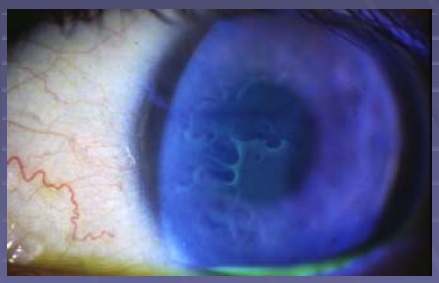
- Most common anterior dystrophy
- Dots, whorls, fingerprints, and “maps”
- Often asymptomatic, may cause ghost imaging and RCE’S
- Appearance constantly changes
  - Manage with NACL solutions or artificial tears (but not at the same time), bandage CL

LASIK issues

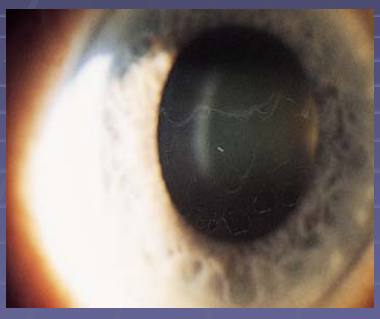
- Amazon.com for NACL

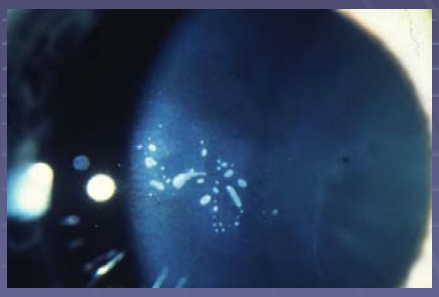
EBMD





EBMD





Meeseman's

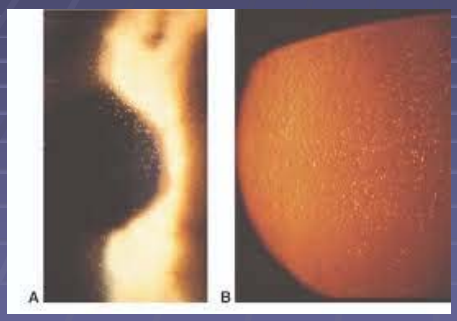
- AD
- Symmetrical: presents in first two years
- Often asymptomatic for several years

Meeseman's

- Tiny intraepithelial cysts
- Uniform size, greater concentration centrally
- Main problem is RCE'S: manage like EBMD

Meeseman's

- Often best seen with retro-illumination



Aao.org

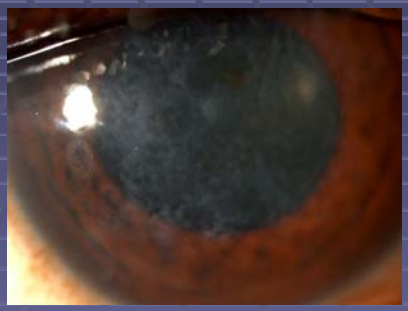
Reis-Buckler

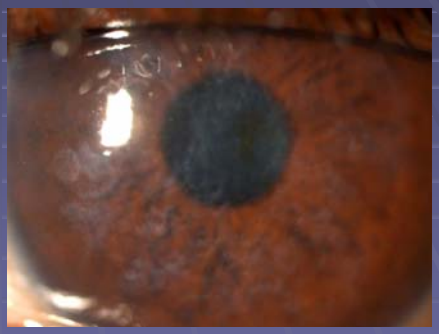
- AD
- Symmetrical: onset first few years
- Rare!
- Honeycomb shaped opacities in Bowman's layer
- More dense centrally

Reis-Buckler

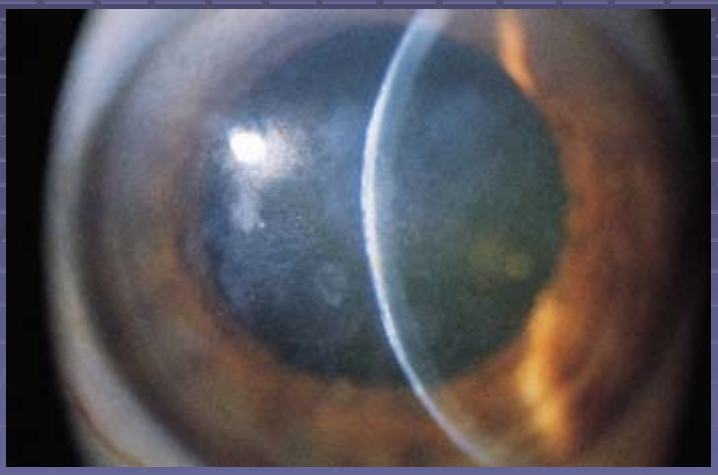
- RCE and scarring related vision loss common by second-third decade
- Only epithelial dystrophy that commonly requires some type of corneal transplant for management

Reis-Buckler



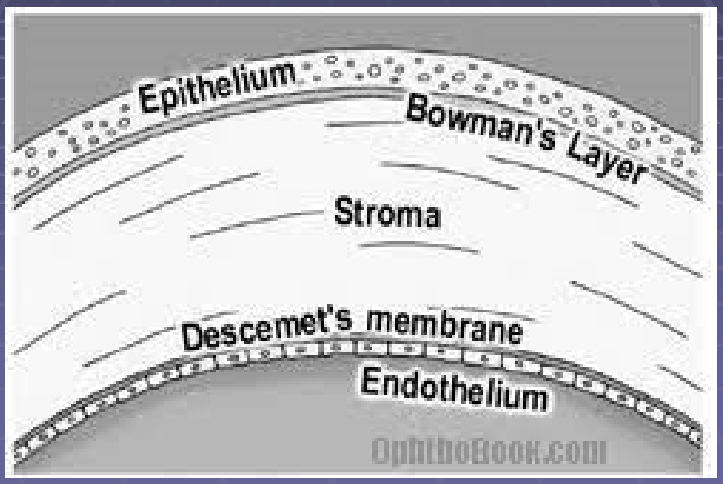


Reis-Buckler



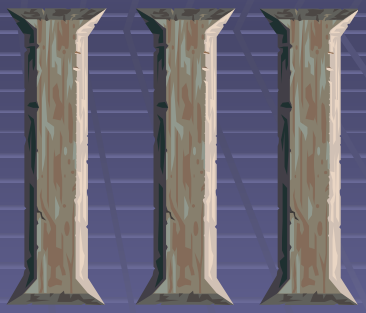
Link.springer.com

Stromal Dystrophies



Lattice Dystrophy

- Three types:
- Lattice Type 1
- Lattice Type 2 (Meretoja syndrome)
- Lattice Type 3 / 3A



Lattice Type 1

- AD inheritance pattern
- Onset in first decade of life
- RCE's common early on
- Branching lattice pattern forms and spreads outward from the center
- Amyloid deposits: stain with Congo Red
- Decreased vision can lead to corneal transplant :reoccurrence in graft relatively common (highest amongst all corneal dystrophies)

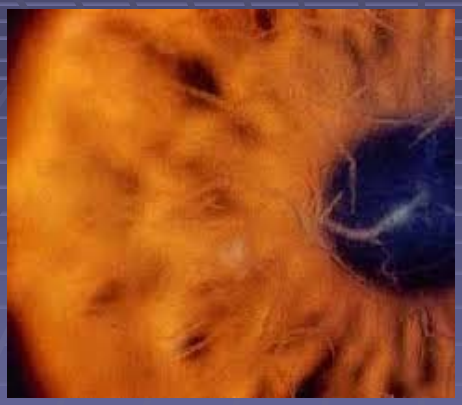
Lattice Type 2 (Meretoja)

- Less severe ocular presentation but concurrent systemic manifestations
- Onset around the fifth decade
- Same ocular signs but less prominent
- Progressive facial palsy

Lattice Type 3 / 3A

- Onset in 4th to 6th decade
- Prominent branching lines in stroma but very little haze
- Slowly progressive, often asymmetric

Lattice Dystrophy



Emedicine.medscape.com

Granular Dystrophy

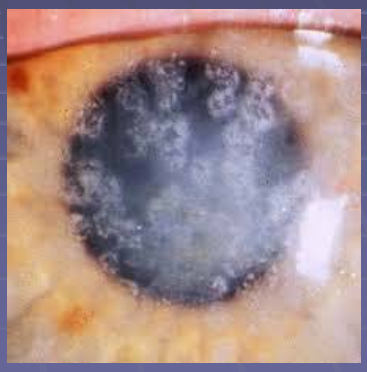
- AD inheritance with onset in first decade
- RCE's (quite rare), glare, decreased vision
- Abnormal keratocytes produce hyaline deposits that stain bright red with Masson trichrome
- Discrete, white opacities begin in the anterior, central stroma and spread. They do not reach the limbus

Granular Dystrophy

- Lesions often resemble crumbs or snowflakes

Granular dystrophy

Wikipedia.org



Macular Dystrophy

- Onset in first decade
- AR inheritance: more severe
- Least common of the stromal dystrophies
- Dense gray-white lesions that are poorly defined
- Mucopolysaccharide: stains with Alcian blue
- Eventually involves multiple corneal layers and extends to the limbus

Macular Dystrophy

- Stromal haze noted very early on (much earlier than granular dystrophy)
- Early transplant

Macular dystrophy

Link.springer.com

Avellino Corneal Dystrophy

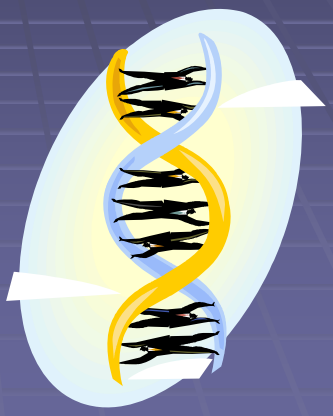
- Avellino is an autosomal dominant corneal stromal dystrophy that shares characteristics of both Lattice and Granular Dystrophies

Avellino Dystrophy

- Originally described in an extended family of patients from Avellino Italy, a small town in central Italy near Naples
- Since reported in families from many other countries

Avellino Dystrophy

- Genetic mutation was localized in 1996 to human chromosome 5 (long arm; 5q31); codon 124
- Arginine to Histadine substitution



Avellino Dystrophy

- Mutation leads to production of an abnormal transforming growth factor Beta protein that deposits in the cornea
- Amyloid in nature : related to brain deposits in Alzheimer's patients
- Standard Lattice and Granular Dystrophies also have their origin in mutations to the same region of chromosome 5 as does Reis-Buckler dystrophy

Avellino Dystrophy

- There are many phenotypic variants (range of clinical manifestations) and vision / symptoms vary widely
- Most common scenario is to have standard Granular type deposits develop in the anterior stroma early in life
- Lattice like lesions then develop in the middle to deep stroma by middle-age followed by stromal haze

Avellino Dystrophy

- Symptoms include:
  - Glare
  - Decreased vision
  - Discomfort / pain from RCE (relatively rare)

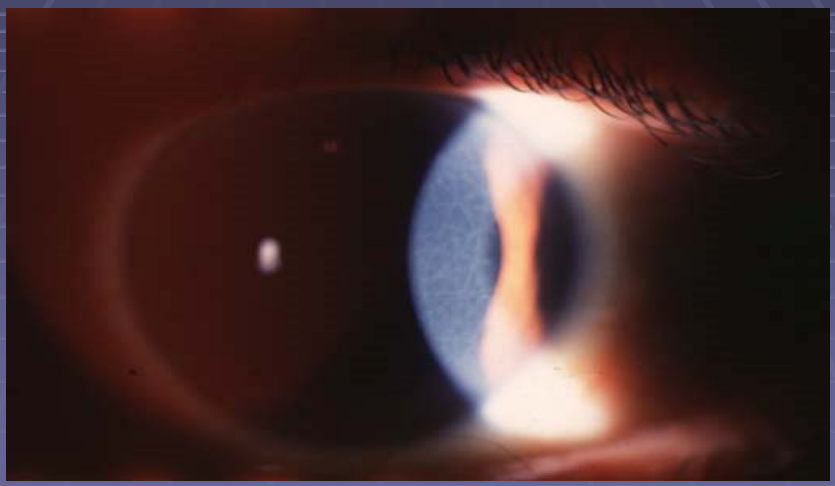
Avellino Dystrophy

- Management includes:
  - Artificial tears / lubricants
  - NaCl solutions
  - Measures for acute RCE when applicable (bandage CL, topical drops, etc.)
  - Corneal transplant in severe cases

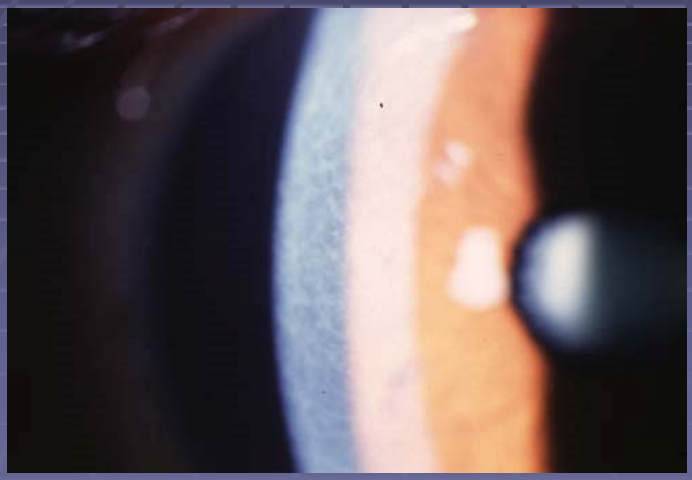
Granular – Lattice Dystrophy

- A better name may be Granular-Lattice Dystrophy

Avellino Dystrophy



Avellino Dystrophy

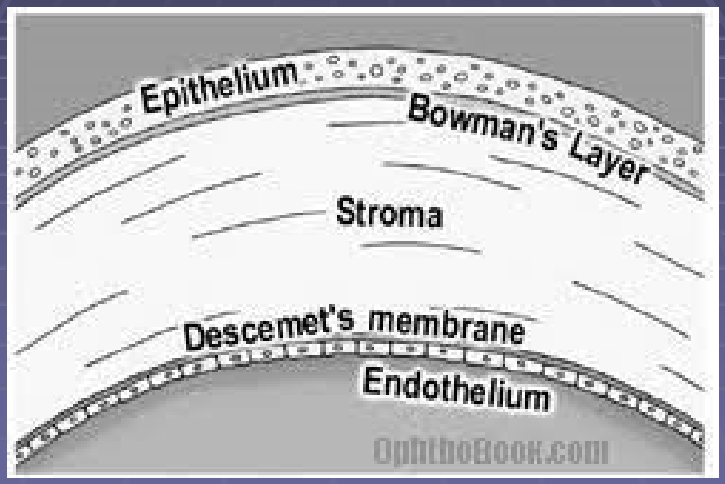


Avellino Dystrophy

Schnyder's Dystrophy

- Rare, AD
- Cholesterol / lipid deposition

Endothelium Dystrophies



Fuch's Dystrophy

- Most commonly encountered endothelial dystrophy. Some AD, many unclear
- One of the few corneal dystrophies with sexual predilection: F 4x M
- Appears between 40-60 years of age
- May be relatively asymmetric compared to other corneal dystrophies
- Often worsened by anterior segment surgery

Fuch's Dystrophy

- Increase in central guttata that then progresses peripherally
- "beaten metal" : seen well in retro
- As endothelial pump fails, stromal edema increases

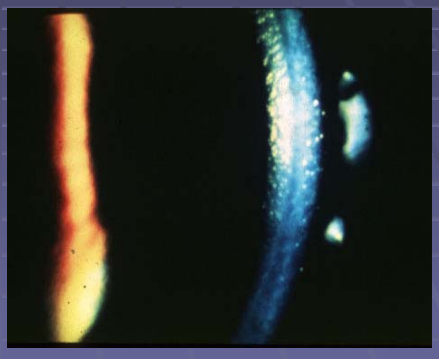
Fuch's

- Stromal edema eventually leads to epithelial disruption: erosions and bullae
- Early on worse in the morning
- Poor vision common

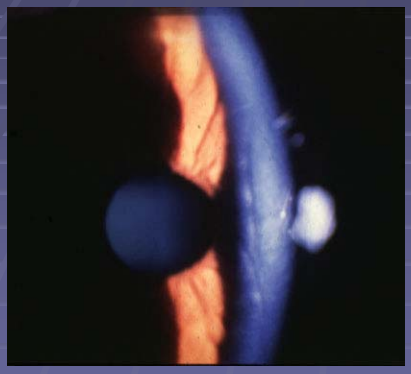
- Nacl solutions to manage epithelial edema: fairly ineffective against stromal edema

Fuch's

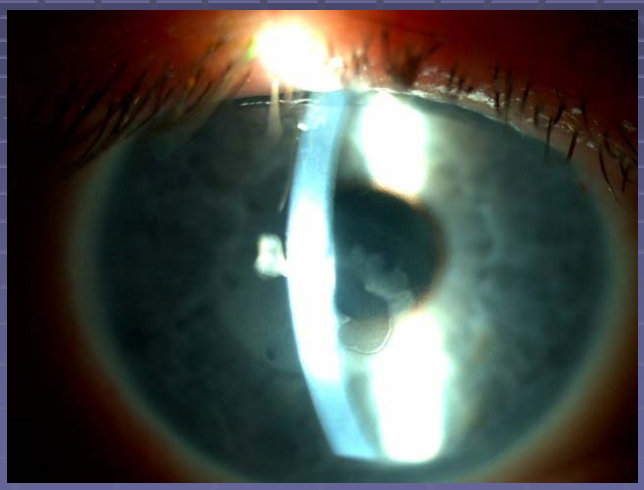
- Bandage CL may be helpful
- Steroids?
- Eventually DSAEK or DMEK



Fuch's



Fuch's with Bullous K



Fuch's with bullous K

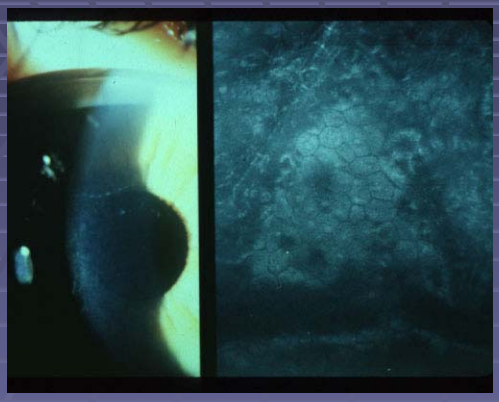
Posterior Polymorphous Dystrophy (PPD)

- AD
- Onset shortly after birth
- Relatively rare
- Most often asymptomatic and identified by chance
- Polymorphous changes in endothelium and deep stroma resembling cysts, lines, nodules, or vesicles

PPD

- Variable appearance
- Can be mistaken for tears in Descemet's membrane
- RCE's can occur but are uncommon
- Need for transplant very rare

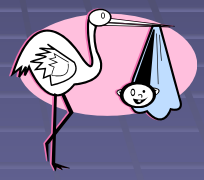
PPD



https://webeye.ophth.uiowa.edu/

Forcep trauma

- Can be mistaken for PPD
- “Railroad track” double line appearance on endothelium that is roughly horizontal

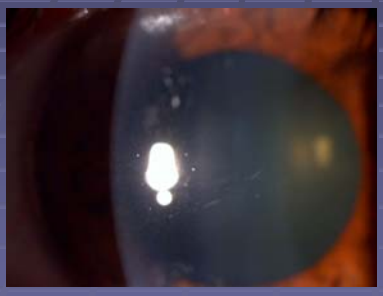


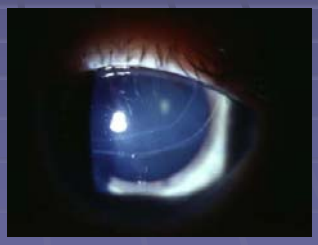
Forcep Trauma

- Forceps squeeze the globe and rupture Descemet’s membrane. The stroma is uninvolved for the most part
- As the eye grows, the edges of the scar separate leading to classic double line
- Asymptomatic, noted during slit lamp exam

Forcep trauma

- No treatment needed





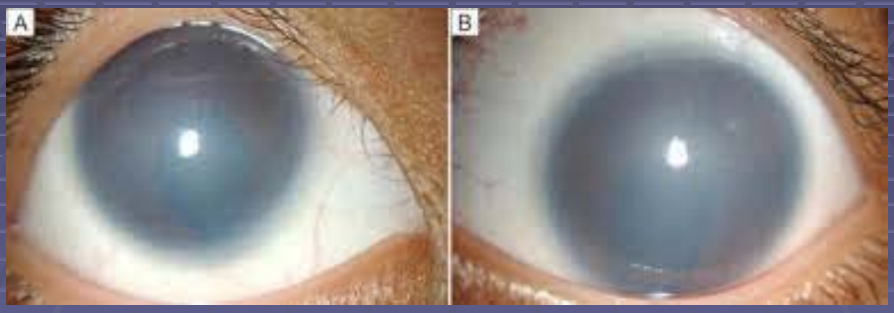
CHED

- Congenital Hereditary Endothelial Dystrophy
- Rare!
- CHED 1 : AD , less severe, presents in first two years of life
- CHED 2 : AR, more severe, cloudy cornea at birth

CHED

- Endothelium fails to develop
- Diffuse corneal edema and scarring
- Epithelial disruption is rare, so pain is uncommon
- Vision can be very poor
- Corneal transplant needed: success variable (deprivation amblyopia, etc.)
- Appearance can mimic congenital glaucoma

CHED



Jaapos.org

Congenital glaucoma

Questions?