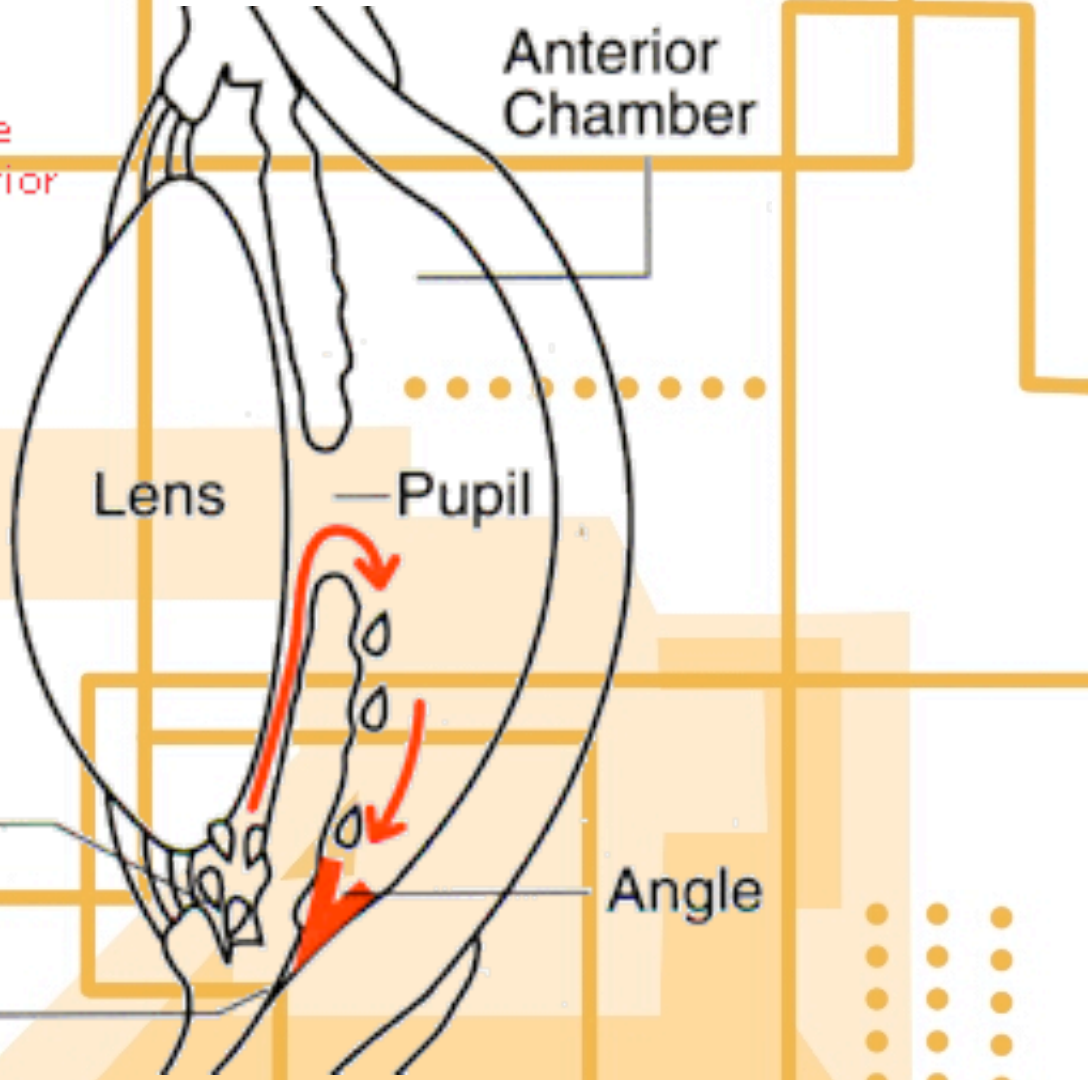
Glaucoma

By Annelise Mah

What is glaucoma?

- Abnormally high intraocular pressure (most cases)
- Damage of the optic nerve
- Different types:
  - Open-angle: flow of aqueous humor unusual
  - Closed-angle: drainage channel of aqueous humor blocked
  - Congenital: from birth
  - Juvenile: recognized in late childhood/early adulthood
  - Adult onset: after 50's
- If not treated will lead to loss of vision, eventual blindness
- Can be in one eye or in both

Diagram of the eye showing how pressure can build in the anterior chamber (behind the lens) in open angle glaucoma. [Image credit: The National Eye Institute, NIH.]



Symptoms

- Slow loss of sight (often unnoticeable)
- Edema
- Opacification of cornea with rupture of Descemet's membrane
- Photophobia- sensitivity to light
- Blepharospasm- spasms of eyelid muscle
- Hyperlacrination- excessive tearing

Classic Diagnosis

- Congenital glaucoma diagnosed at birth
- Perimetry: testing field of vision
- Observation: increased corneal diameter and expanded globe, abnormal appearance of optic disc
- Applanation tonometry- tests the intraocular pressure by pushing the eyeball with a small tool
  - Normal IOP 10-21 mmHg
  - Glaucoma >21 mmHg

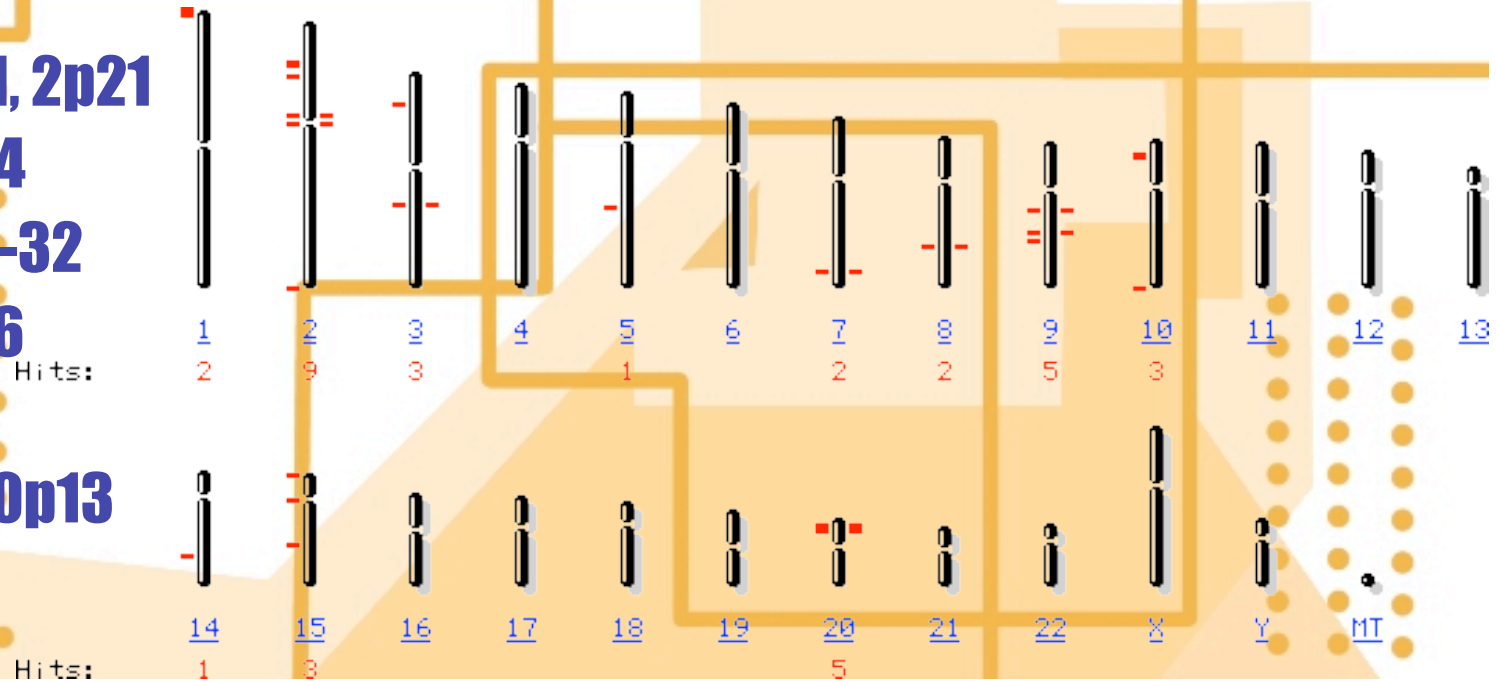
Classic Treatment

- Mild cases that are corrected early normally regain normal vision
- Surgery to open up drainage channel
  - Laser trabeculoplasty
  - trabeculectomy
- Drainage implants
- Medicine to control intraocular pressure

Genetics

- Congenital: autosomal recessive
- Juvenile and adult-onset: autosomal dominant
- Multifactorial inheritance

- Known genetic causes:
- GLC1A: MYOC, 1q24-25
- GLC3B: 1p36
- GLC3A: CYP1B1, 2p21
- GLC1C: 3q21-24
- GLC1M: 5q22.1-32
- GLC1F: 7q35-36
- GLC1J: 9q22
- GLC1E: OPTN, 10p13
- GLC1K: 20p12



Speculation...

- According to case studies, primary open-angle glaucoma could also be caused by...
  - Abnormal amounts of cortisol metabolism enzymes. An intermediate stage builds up at the open angle.
  - Immune system abnormalities– too many free radicals and unusual amounts of certain lymphocytes

Genetic Diagnosis & Treatment

- Sequence and mutation analysis tests sparsely available
- Can forewarn individual and family so the disease can be watched for and stopped earlier
- Gene therapy not currently studied