



Albinism

An Ophthalmologist's Perspective

James Elder

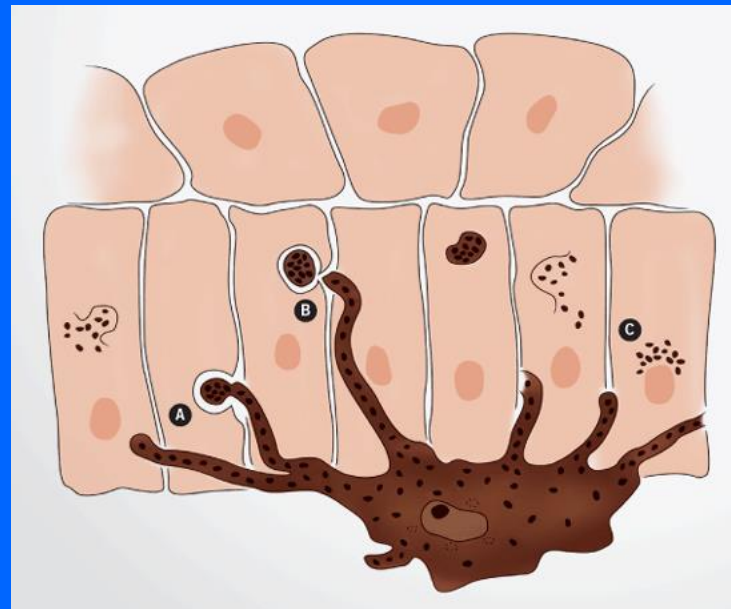
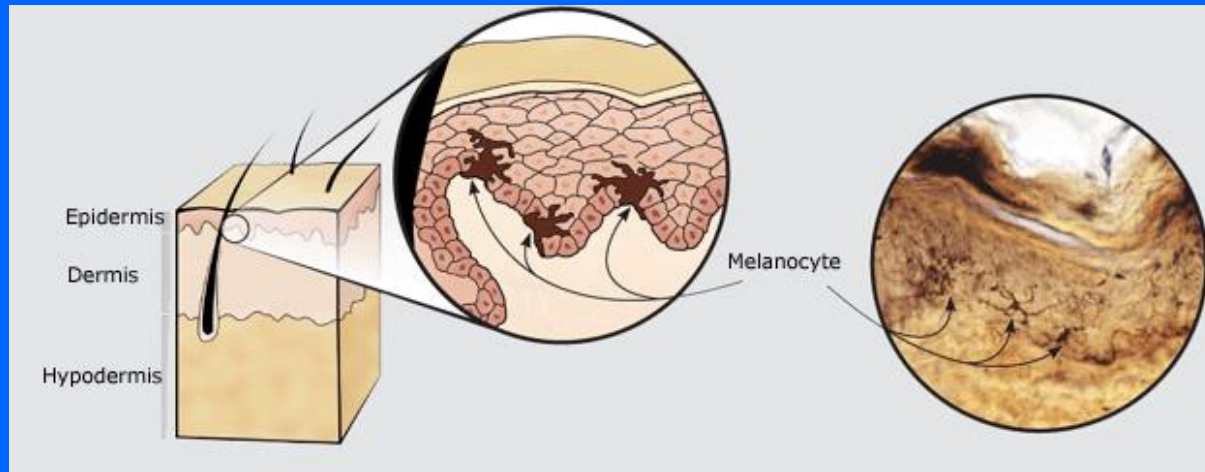
Department of Ophthalmology
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The Children's
Excellence in
clinical care,
research and
education

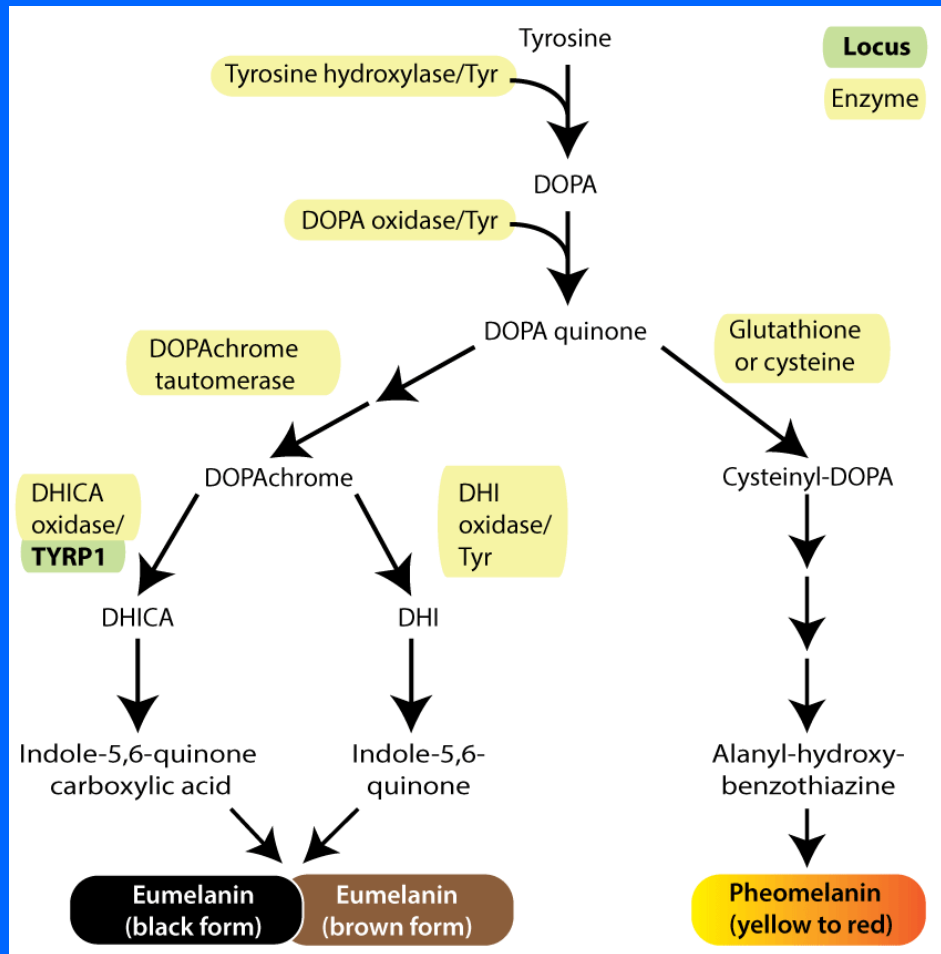




Pigmentation & Melanin



Melanin Production

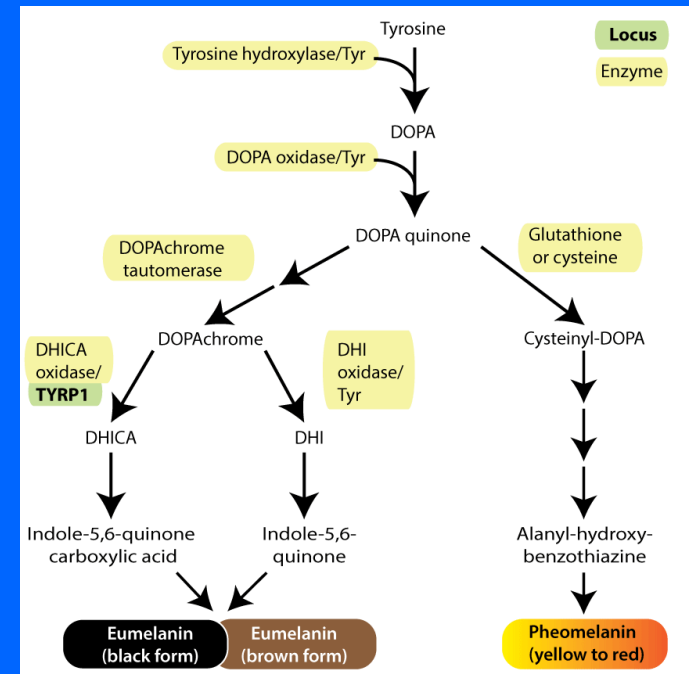


Types of Albinism

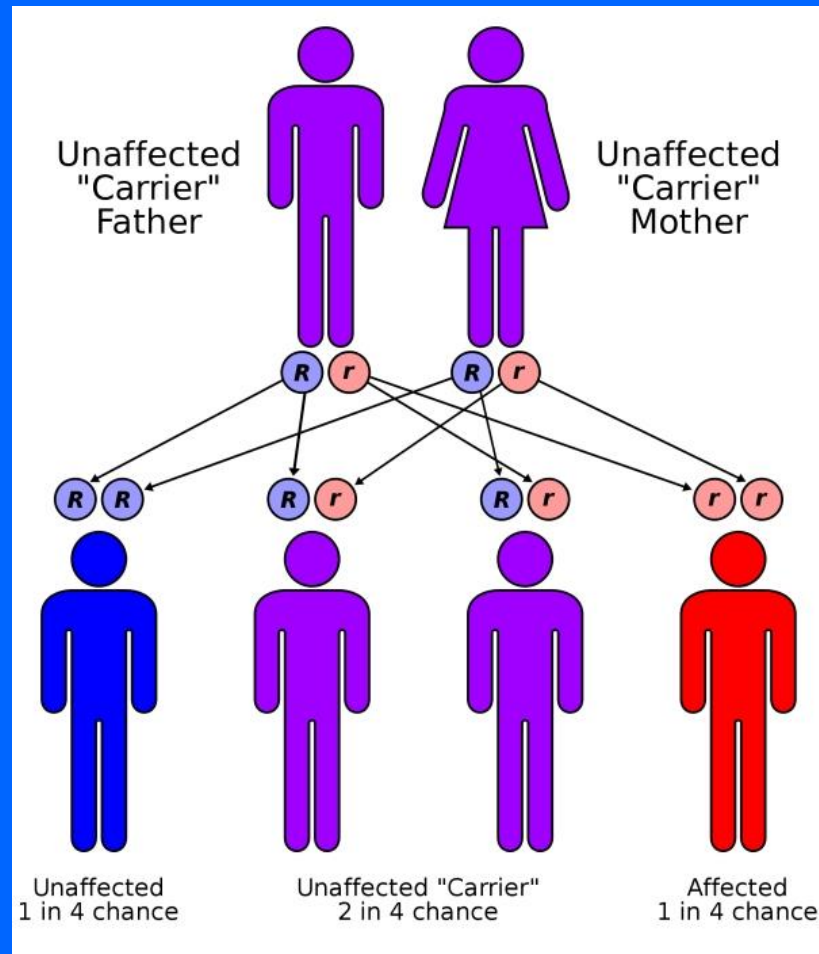
- Oculocutaneous
- Ocular
- Albinism with systemic features

Oculocutaneous (OCA)

- Types 1 – 7 (& counting) - Recessive
- OCA1
 - Tyrosinase absent
 - Worst vision
- OCA2
 - Due to mutation in *P* gene
 - Stops tyrosine entering melanosome
 - Often better vision



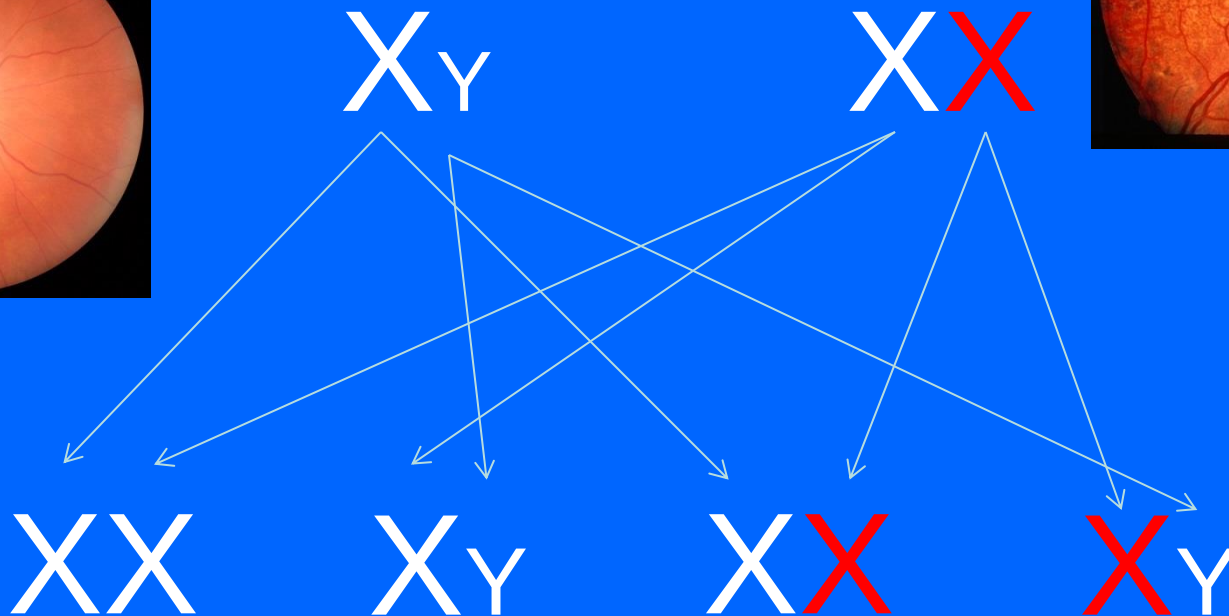
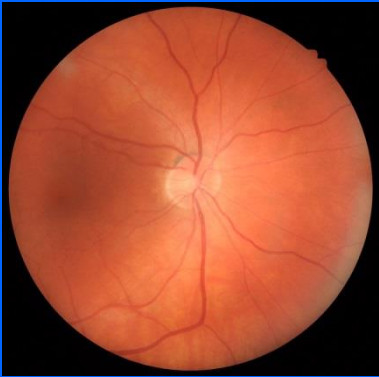
Recessive Inheritance



Ocular Albinism

- ↓ eye pigment
- Due to mutation in *GPR143* gene
- Melanin accumulates in melanosome (creating macromelanosomes)
- X-linked

X-linked Inheritance



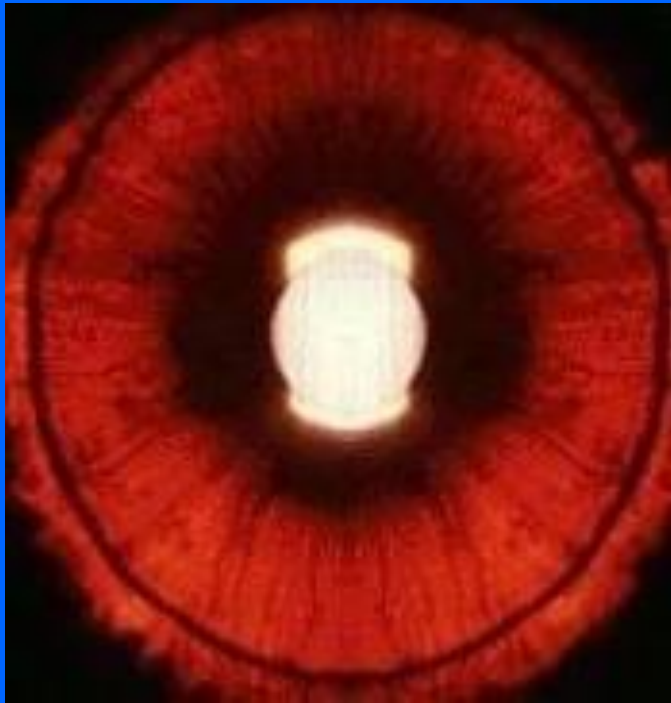
Albinism with systemic features

- Hermansky-Pudlak
 - OCA
 - Bleeding disorder
- Chediak-Higashi
 - OCA
 - White cell disorder

Eye features of Albinism

- Variable
- Some very obvious
- Others less obvious

Reduced pigment



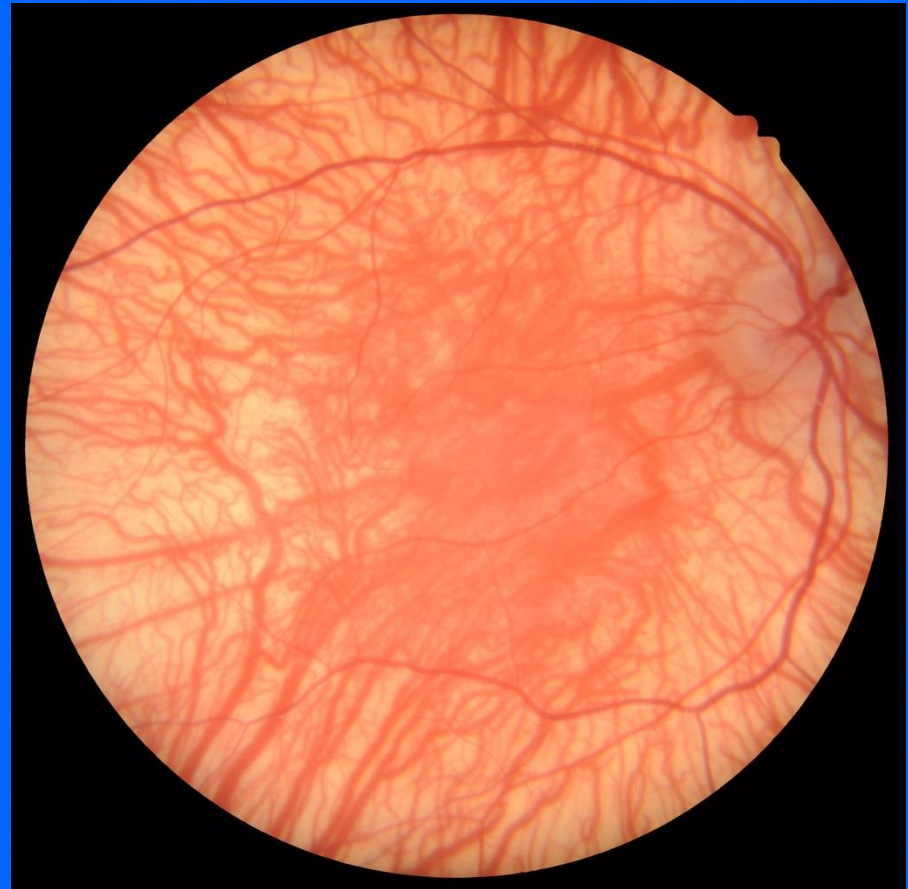
Eye features of Albinism

- Effects quality of vision
- Effects eye movements

Quality of vision

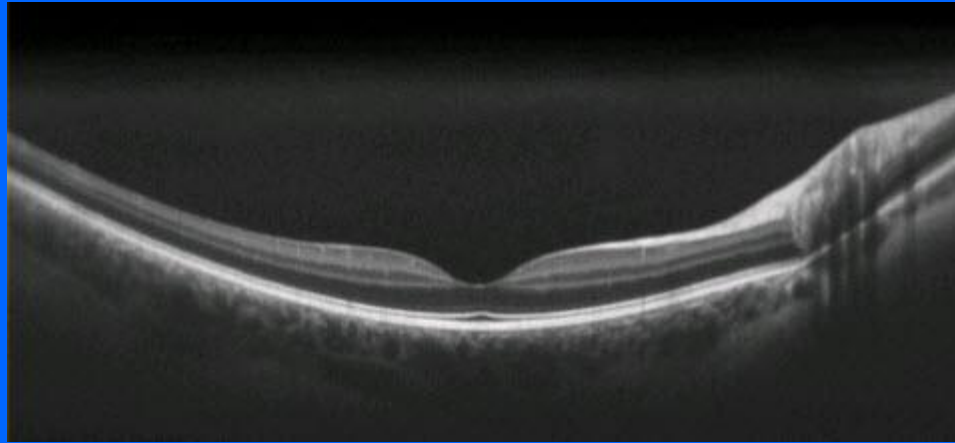
- Abnormality in eye
- Abnormal co-ordination of eyes
- Glasses problems

Retina in albinism

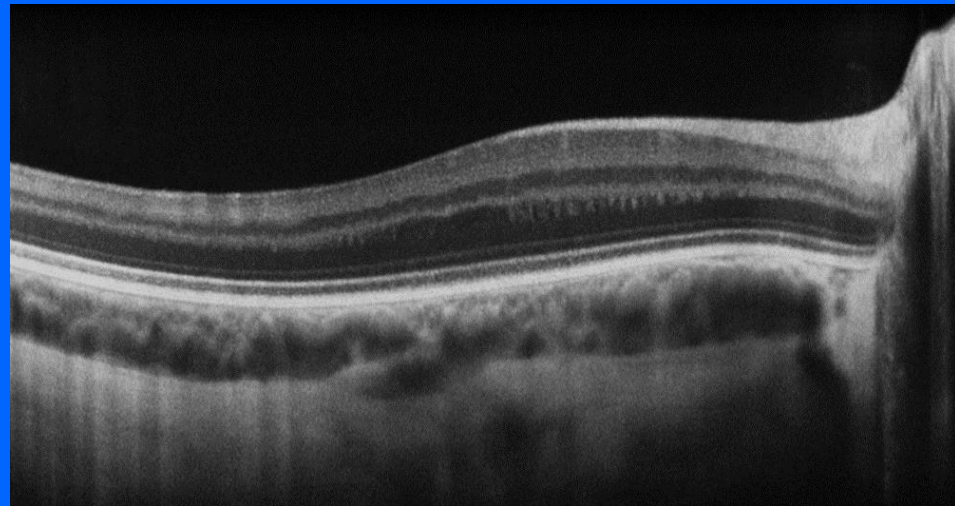


Retina in albinism

Normal



Albinism

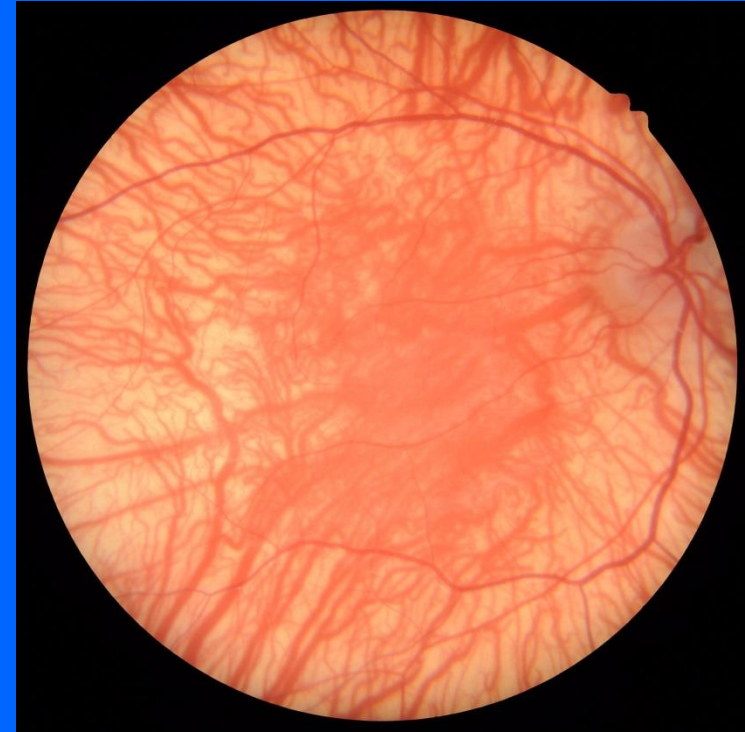


Foveal hypoplasia

- Common in albinism
- Results in reduced vision

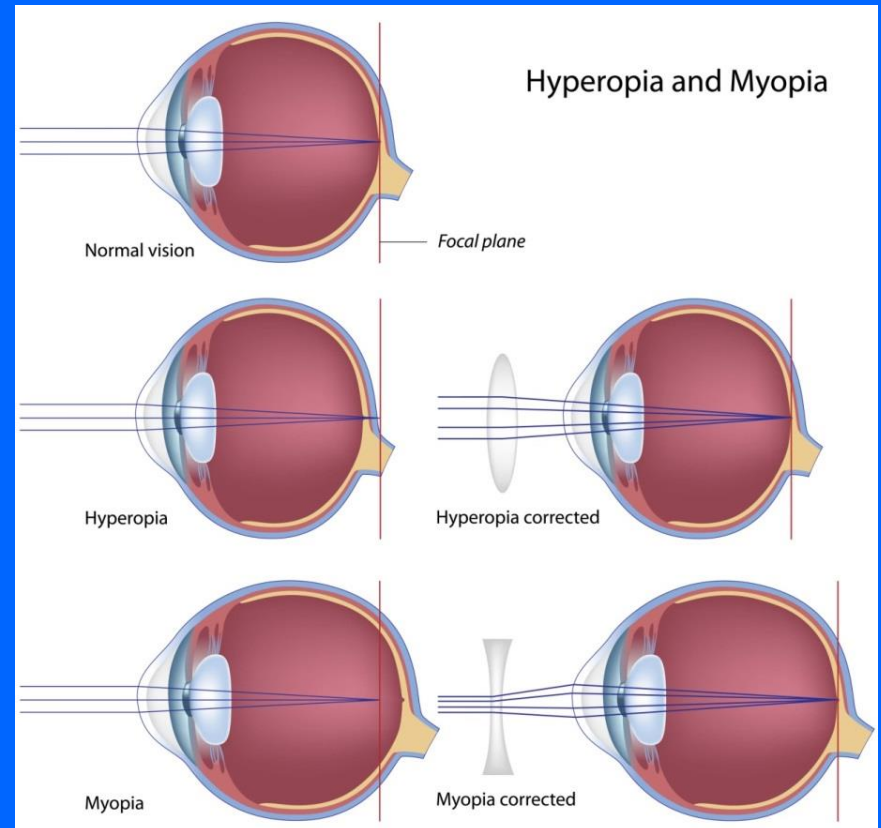
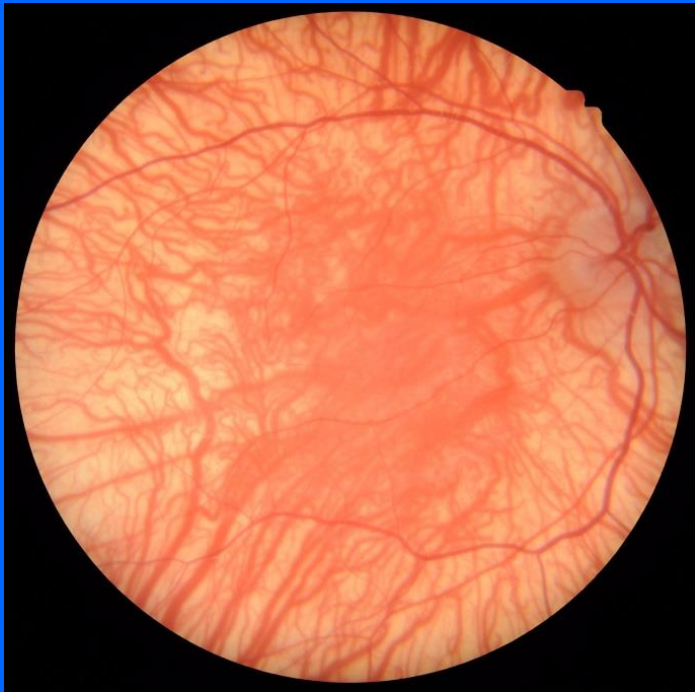
GLARE!

- Common
- Due to ↓ pigment



Glasses = Refractive Error

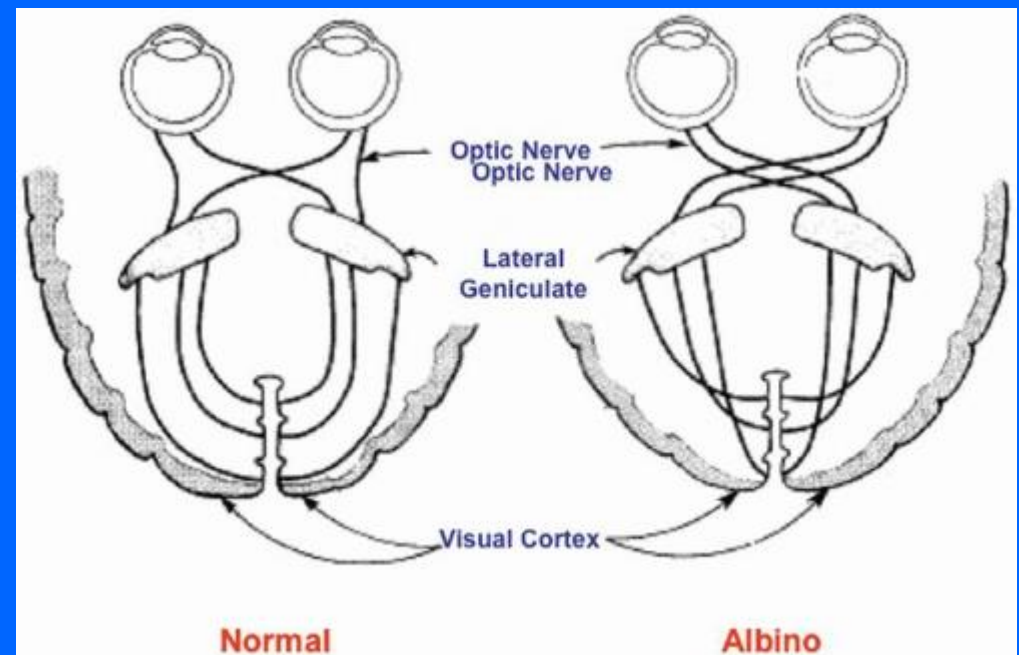
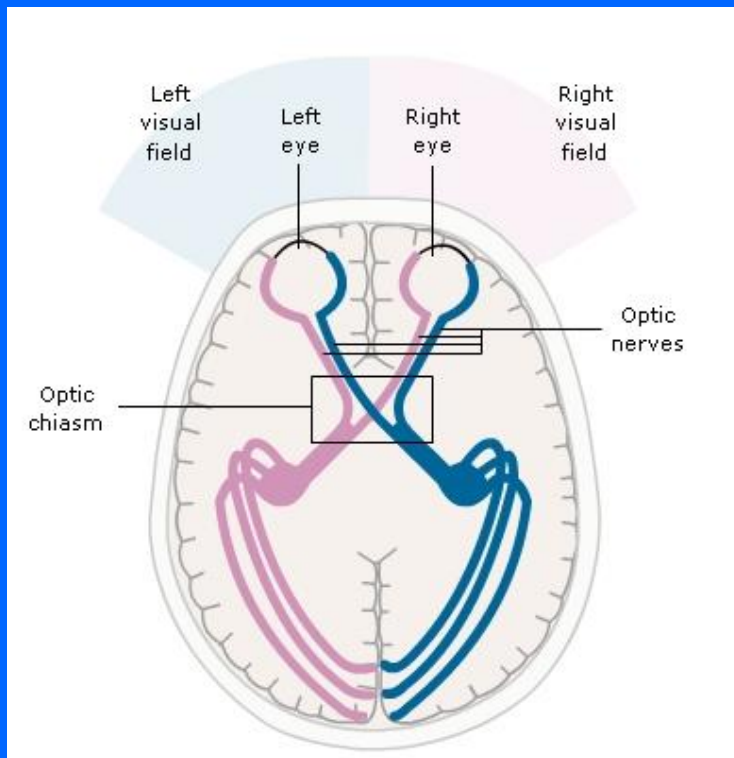
- Simply wearing glasses $\neq$ better vision



Eye movements

- Turned eyes = strabismus
- Wobbly eyes = nystagmus

Albinism & the brain



Turned eyes



- ↓ binocular potential
- ↑ long sightedness

Wobbly eyes



Wobbly eyes

- Largely caused by reduced vision
- Severity $\neq$ level of vision
- Vision related to time eyes are NOT moving
- Stopping movement will not always improve vision

What can we do!

- Support services
 - Referral is 1st step!
 - Pre-school
 - Educational
 - O & M and low vision aides
 - Employment

What can we do!

- Glasses
 - Sunglasses
 - Correct refractive error

What can we do!

- Strabismus (turned eyes)
 - Glasses if longsighted
 - Operations if bad enough
 - Results variable

What can we do!

- Nystagmus
 - Consider surgery
 - Results variable

Thank you
&
Questions