



# Albinism

## An Ophthalmologist's Perspective

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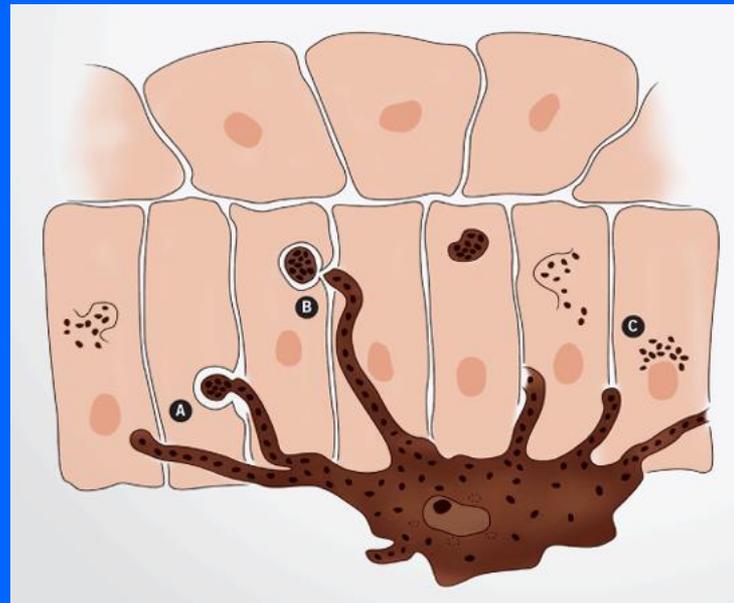
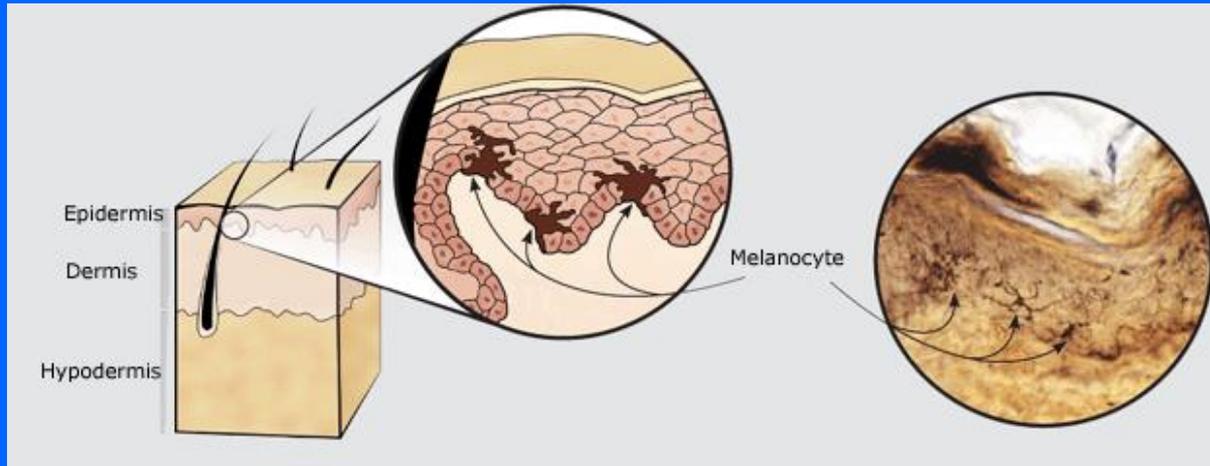
The Children's

Excellence in  
clinical care,  
research and  
education

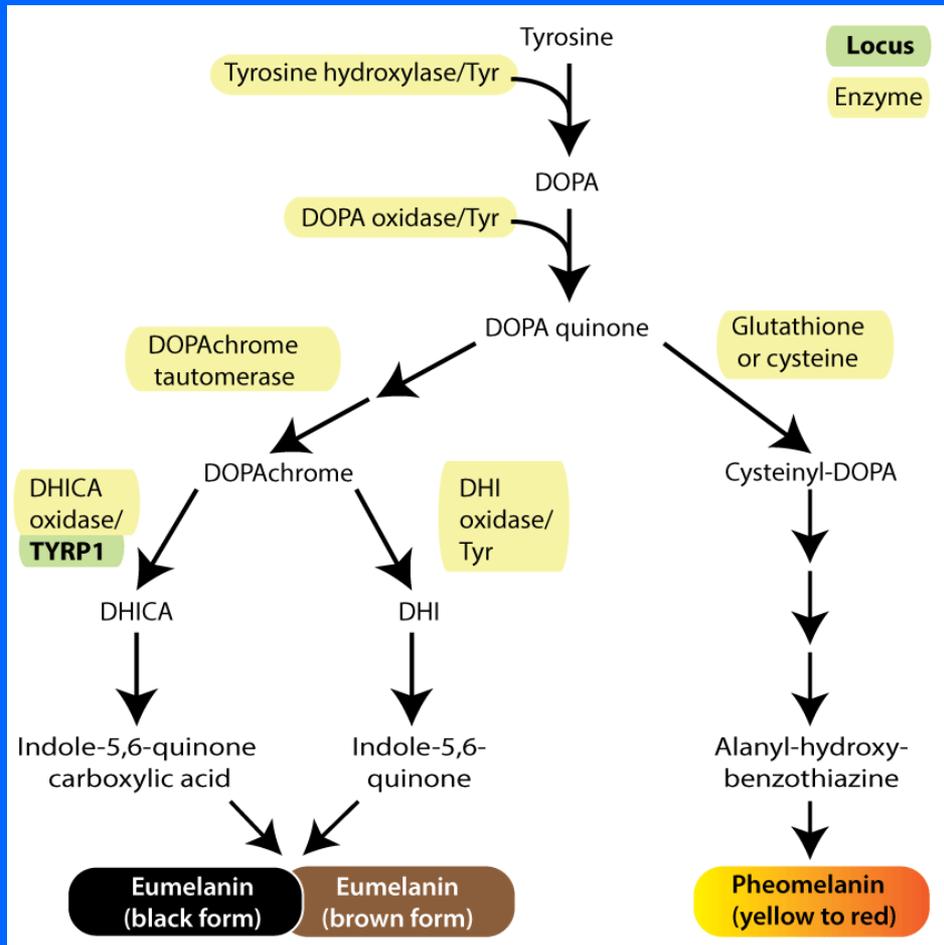




# Pigmentation & Melanin



# Melanin Production

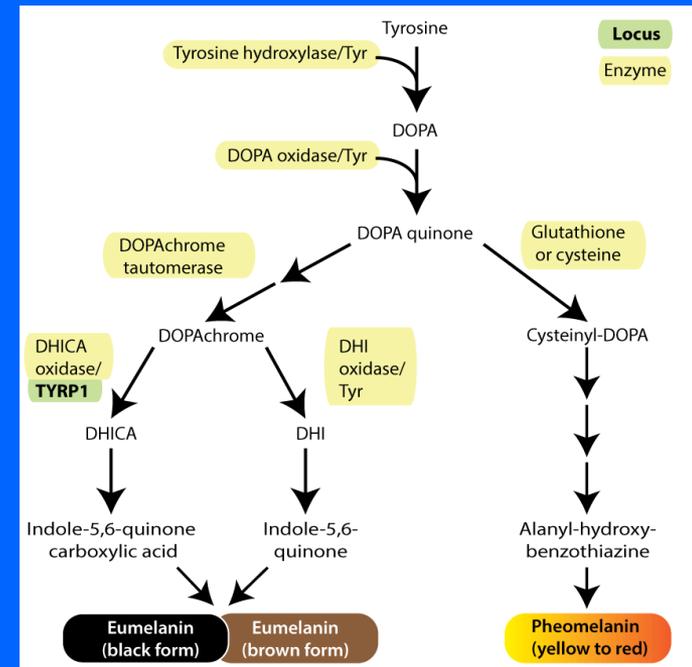


# Types of Albinism

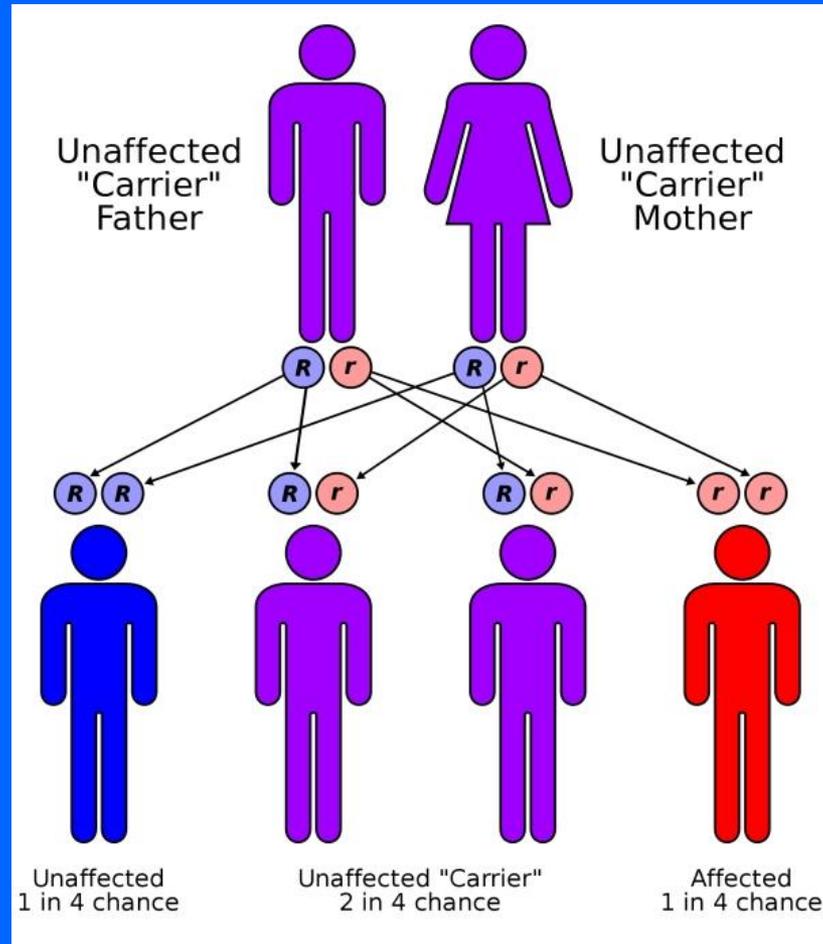
- Oculocutaneous
- Ocular
- Albinism with systemic features

# 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



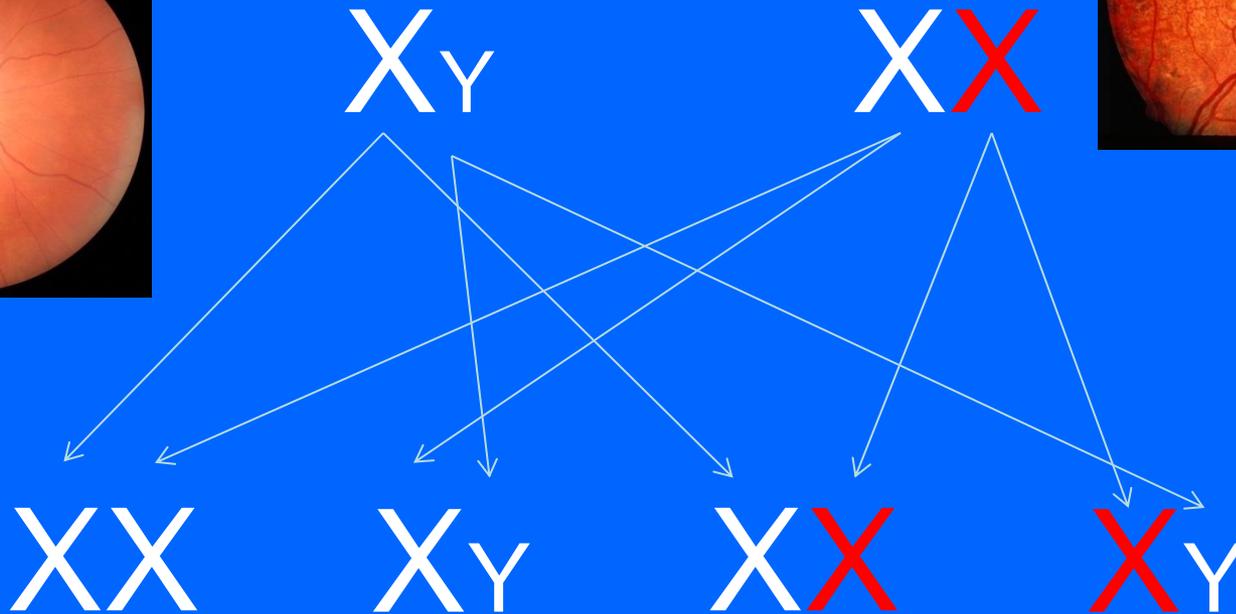
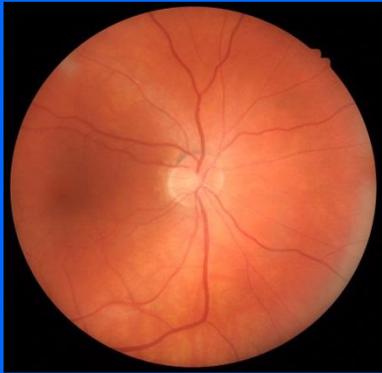
# Recessive Inheritance



# Ocular Albinism

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

# X-linked Inheritance



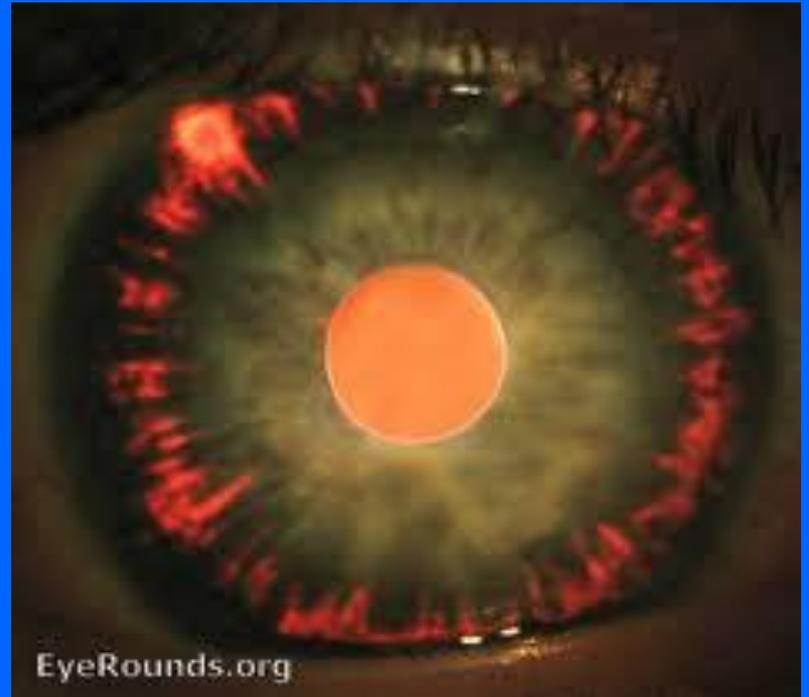
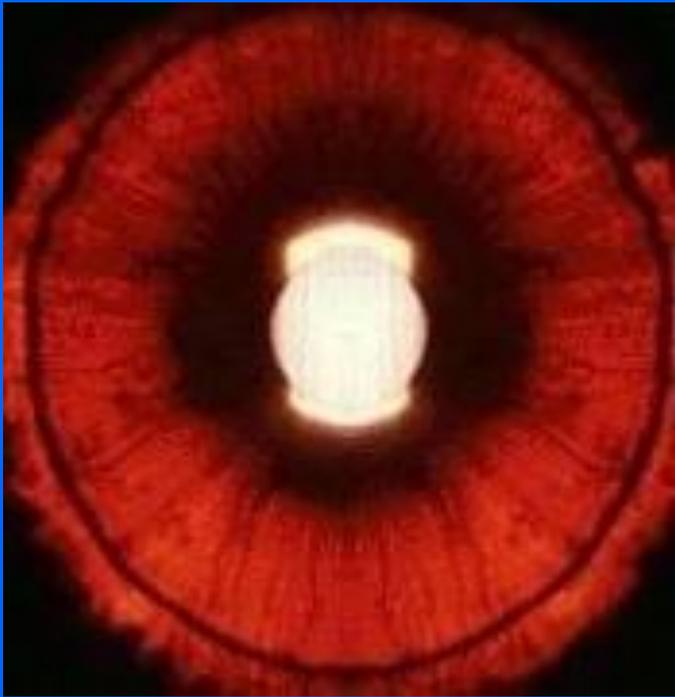
# Albinism with systemic features

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

# Eye features of Albinism

- Variable
- Some very obvious
- Others less obvious

# Reduced pigment



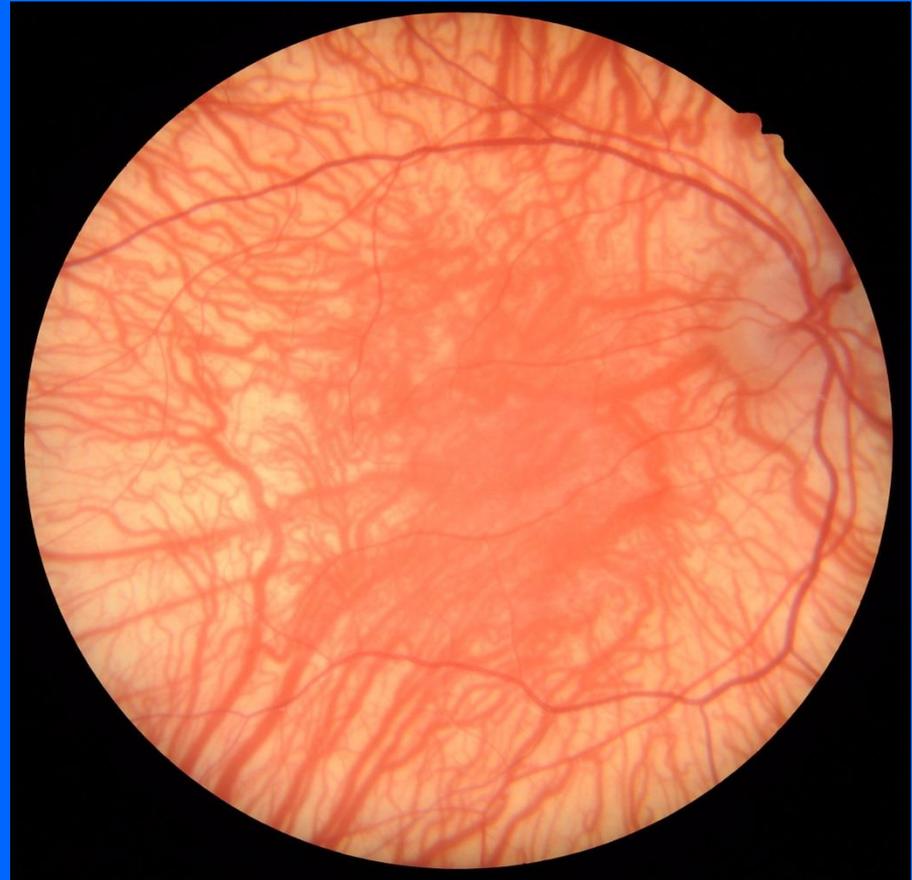
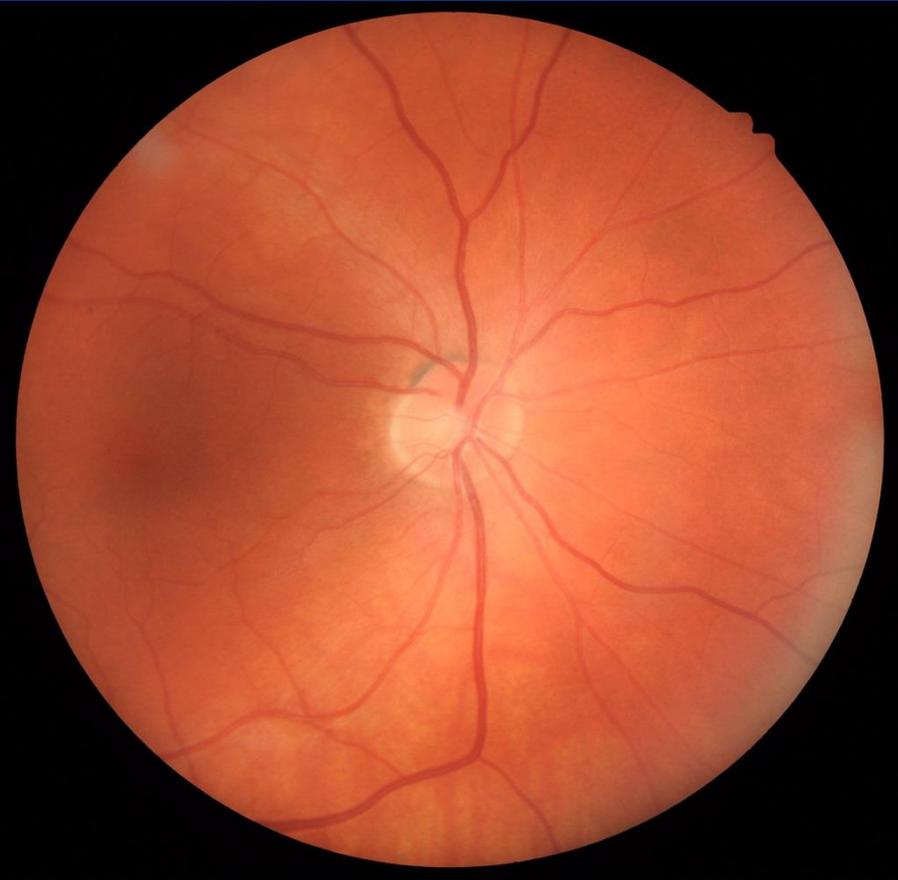
# Eye features of Albinism

- Effects quality of vision
- Effects eye movements

# Quality of vision

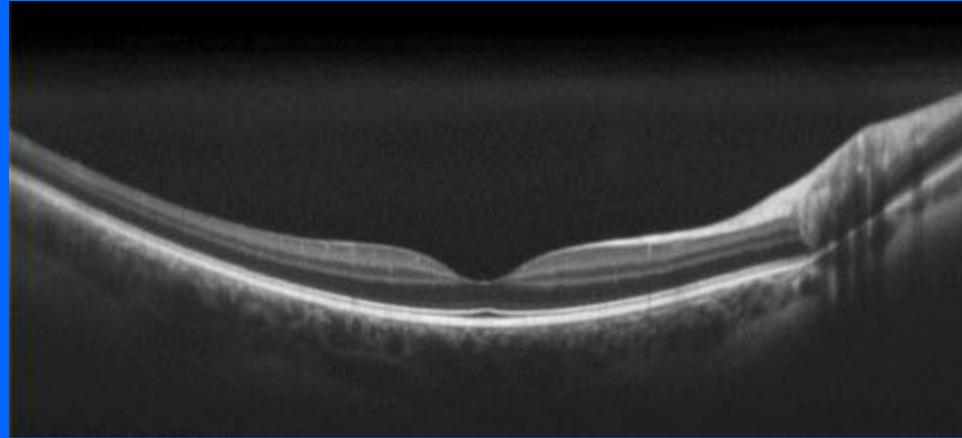
- Abnormality in eye
- Abnormal co-ordination of eyes
- Glasses problems

# Retina in albinism

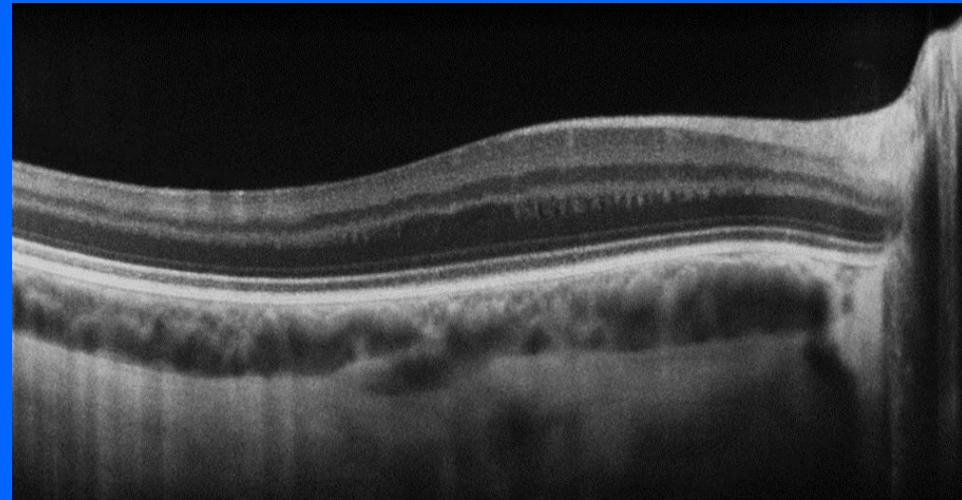


# Retina in albinism

Normal



Albinism

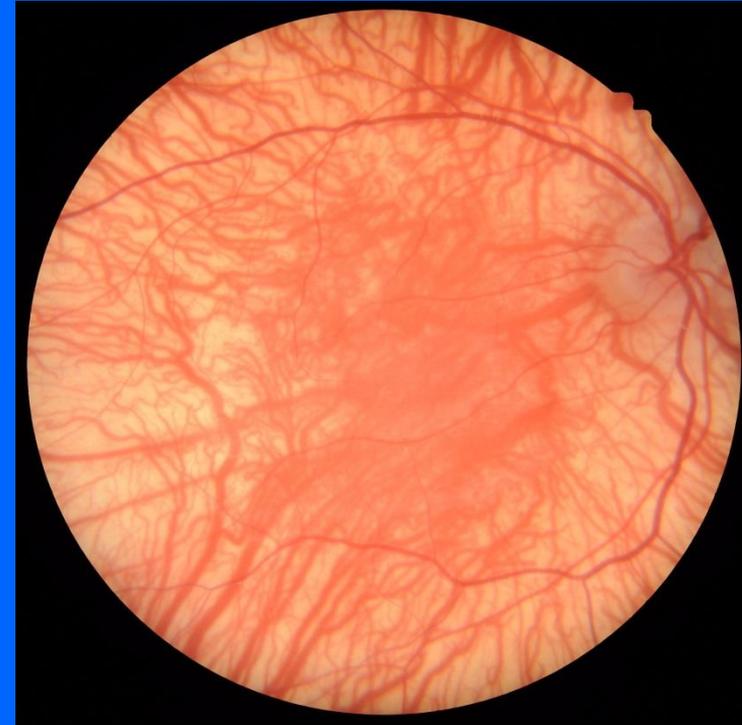


# Foveal hypoplasia

- Common in albinism
- Results in reduced vision

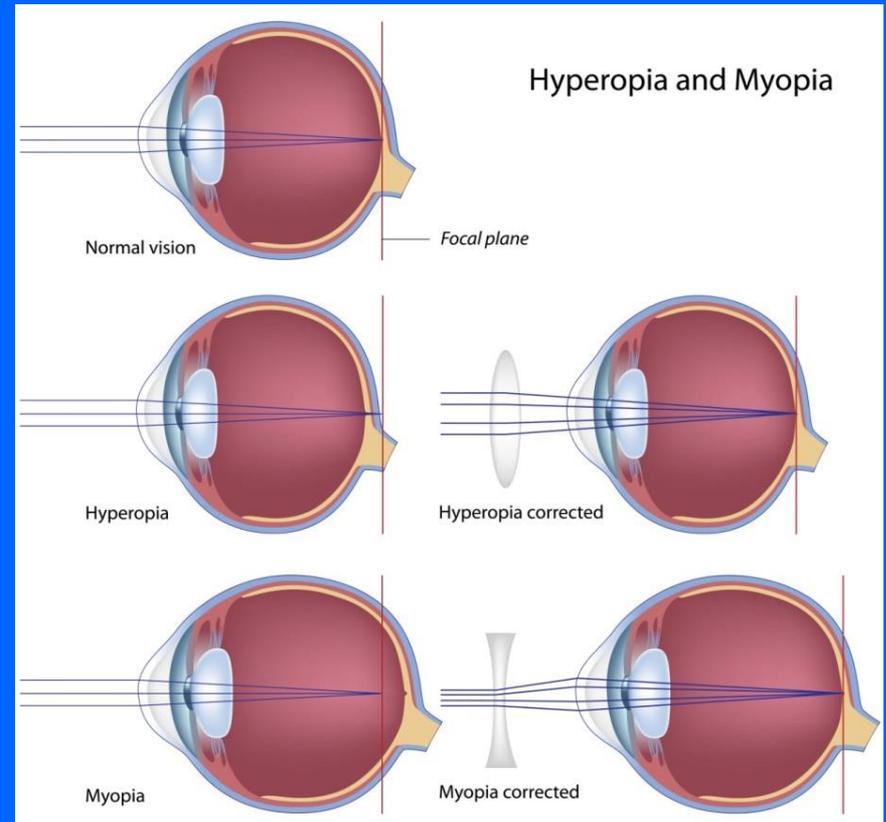
# GLARE!

- Common
- Due to ↓ pigment



# Glasses = Refractive Error

- Simply wearing glasses  $\neq$  better vision



# Eye movements

- Turned eyes = strabismus
- Wobbly eyes = nystagmus



# 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 1<sup>st</sup> 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