Pigmentation & Melanin
Melanin Production

Diagram showing the production of melanin involving enzymes and intermediates such as Tyrosine, DOPA, DOPAchrome, DHICA, and DHI, leading to eumelanin (black form) or pheomelanin (yellow to red).
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

- Unaffected "Carrier" Father
- Unaffected "Carrier" Mother
- Unaffected 1 in 4 chance
- Unaffected "Carrier" 2 in 4 chance
- Affected 1 in 4 chance
Ocular Albinism

• ↓ eye pigment
• Due to mutation in \textit{GPR143} gene
• Melanin accumulates in melanosome (creating macromelanosomes)

• X-linked
X-linked Inheritance

XX → XY → XX

XX → XY → XX → XY
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

Normal

Albinism
Foveal hypoplasia

- Common in albinism
- Results in reduced vision
GLARE!

- Common
- Due to ↓ pigment
Glasses = Refractive Error

- Simply wearing glasses ≠ 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 ≠ 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\textsuperscript{st} 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