



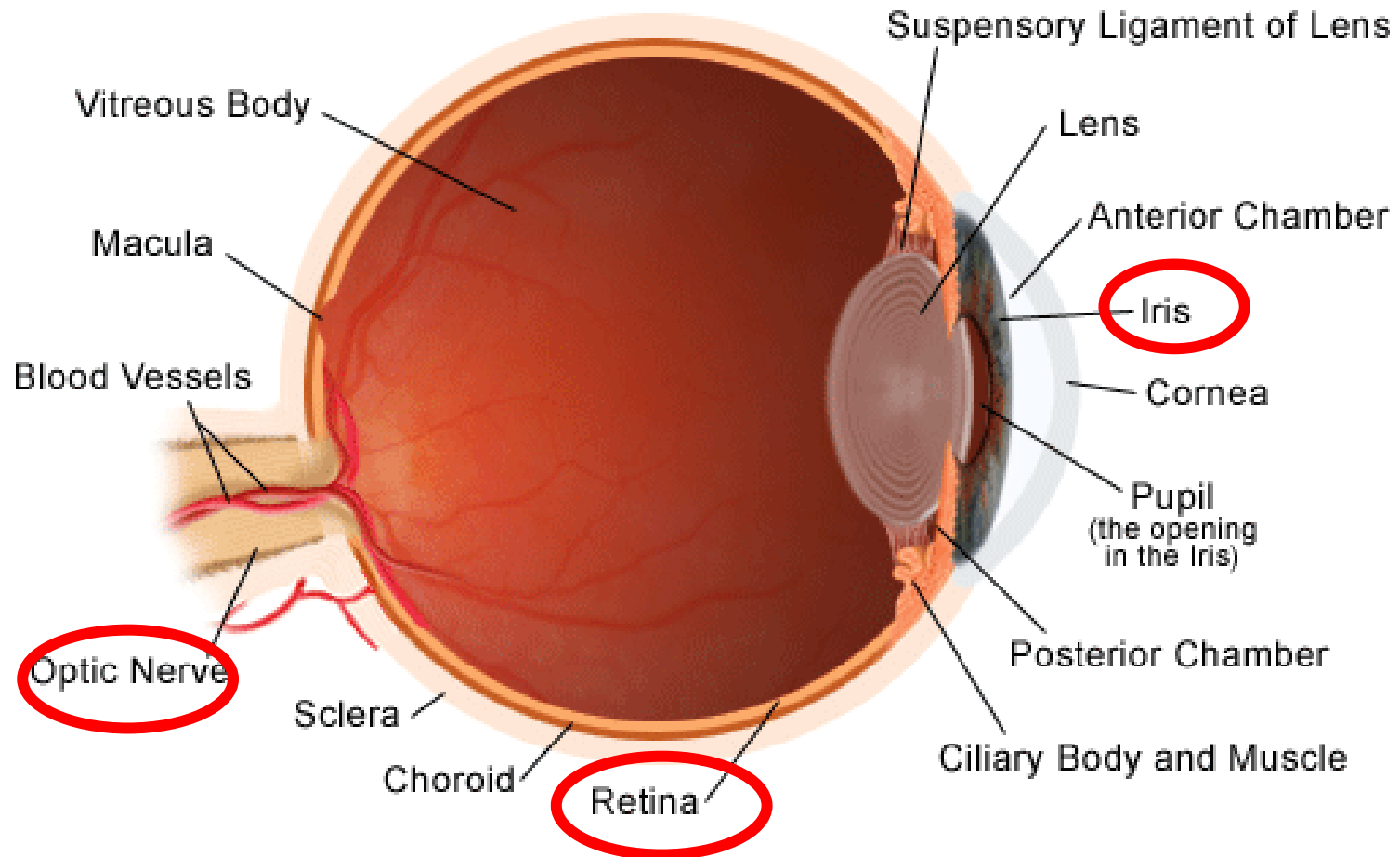
**An Introduction to Albinism
+
An Update on New Research**

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Belfast**

What I will cover:

- The eye
- What is Albinism
- The eye signs
- How is the diagnosis made
- Management
- Outcomes
- The genetics of oculocutaneous Albinism
- The genetics of ocular albinism
- The NINA study findings
- Registration in Northern Ireland

The eye



Melanin

- Is found in hair, skin, retinal cells and iris cells
- Inside these cells melanin is made and stored in melanosomes
- To have normal pigmentation, the cell must make melanosomes and enough melanin must be found in of these vesicles.
- In all of us the colour of our skin and eyes is determined by the amount of melanosomes each cell has rather than the number of pigmented cells we have.

What is albinism?

- Albinism refers to disorders of reduction of pigmentation compared with others of the same ethnic and racial backgrounds with characteristic eye involvement.
- If there is normal pigmentation of the skin and hair, the disorder is referred to as ocular albinism
- If there is a reduction of pigment of the skin and hair, the disorder is called oculocutaneous albinism.

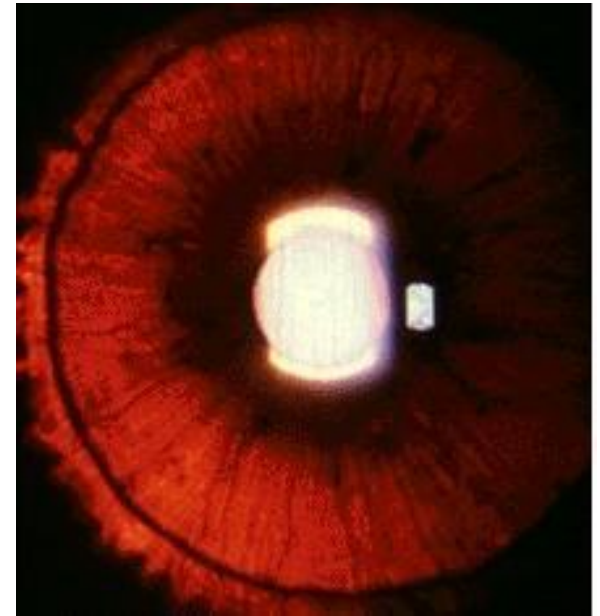
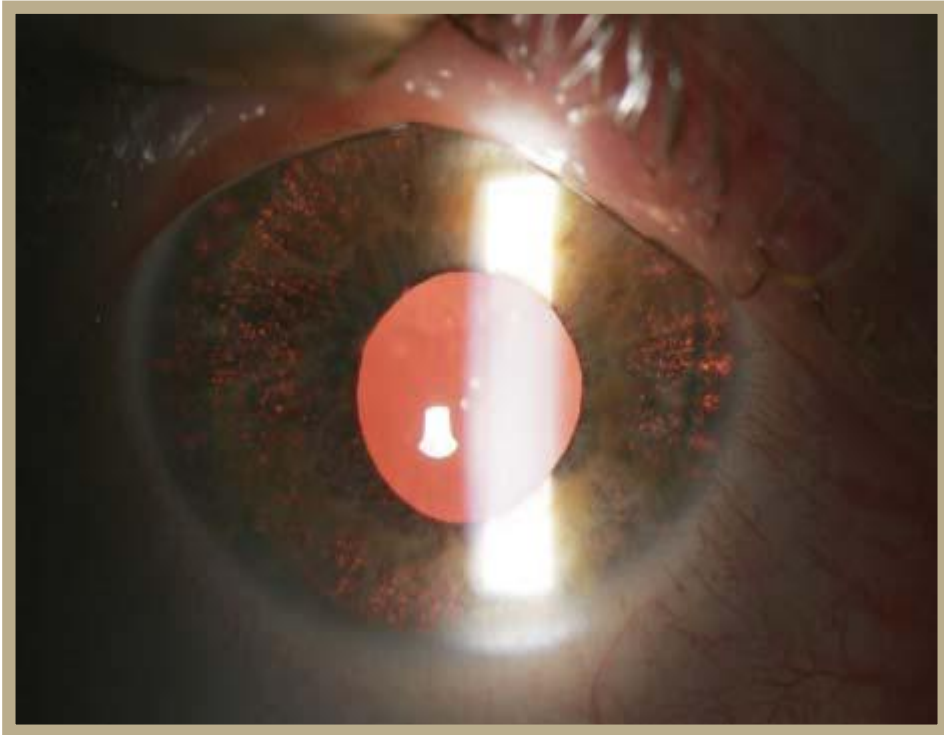
1 in 18,000 World Wide
1 in 10,000 in Northern Ireland



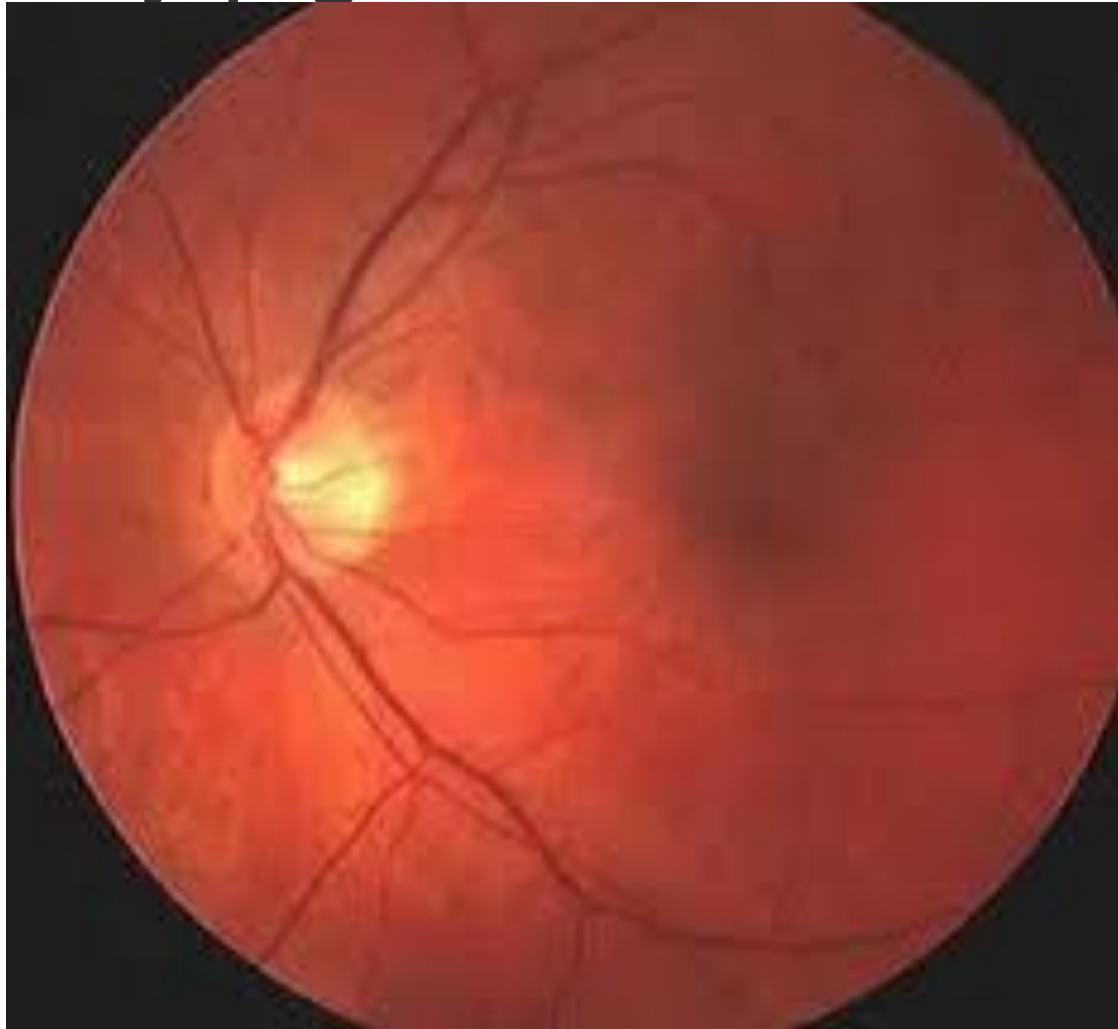
Eye signs

- Changes within the eye
- Changes in the optic nerve
- Changes in the movement and position of the eye

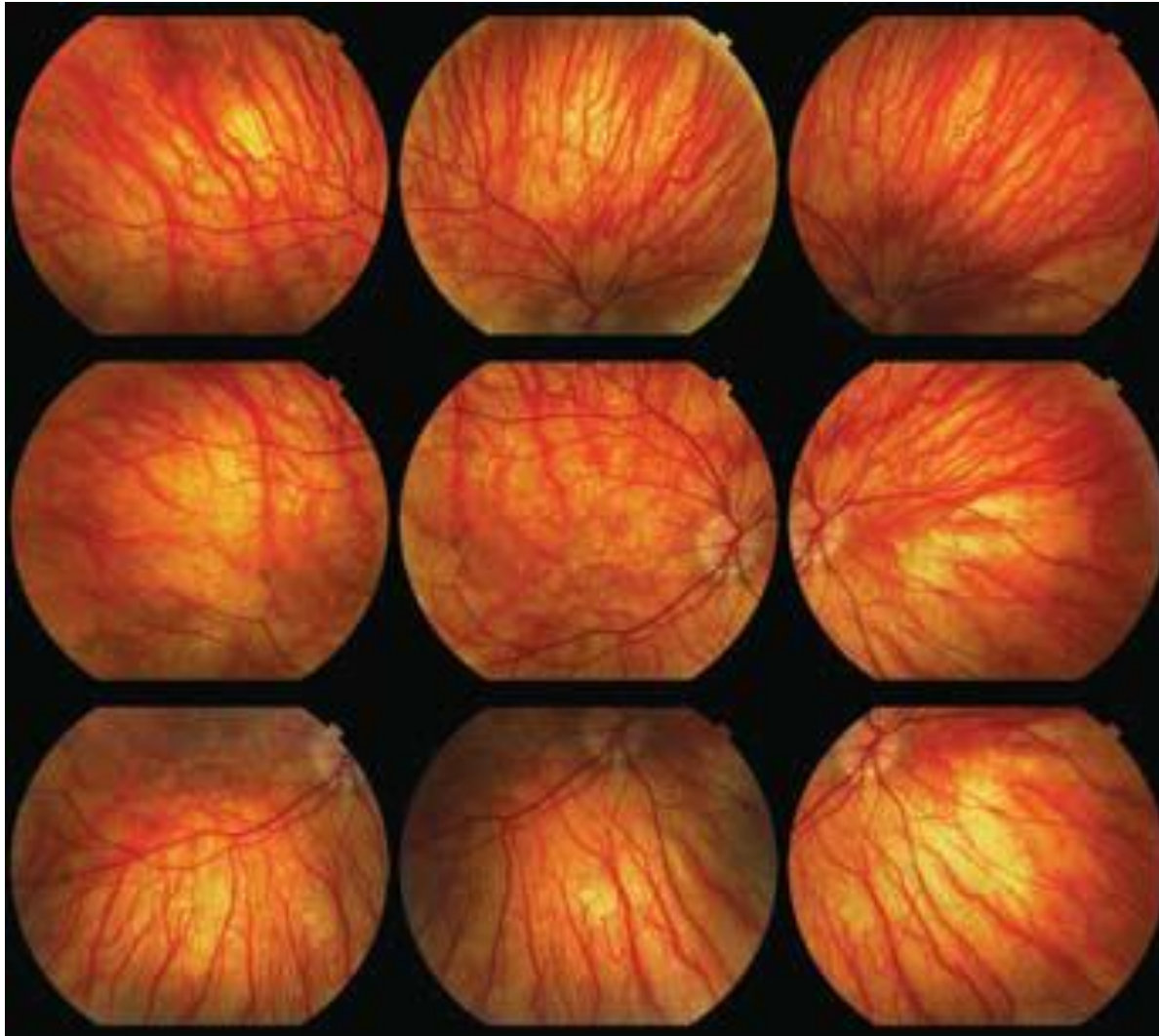
Iris transillumination



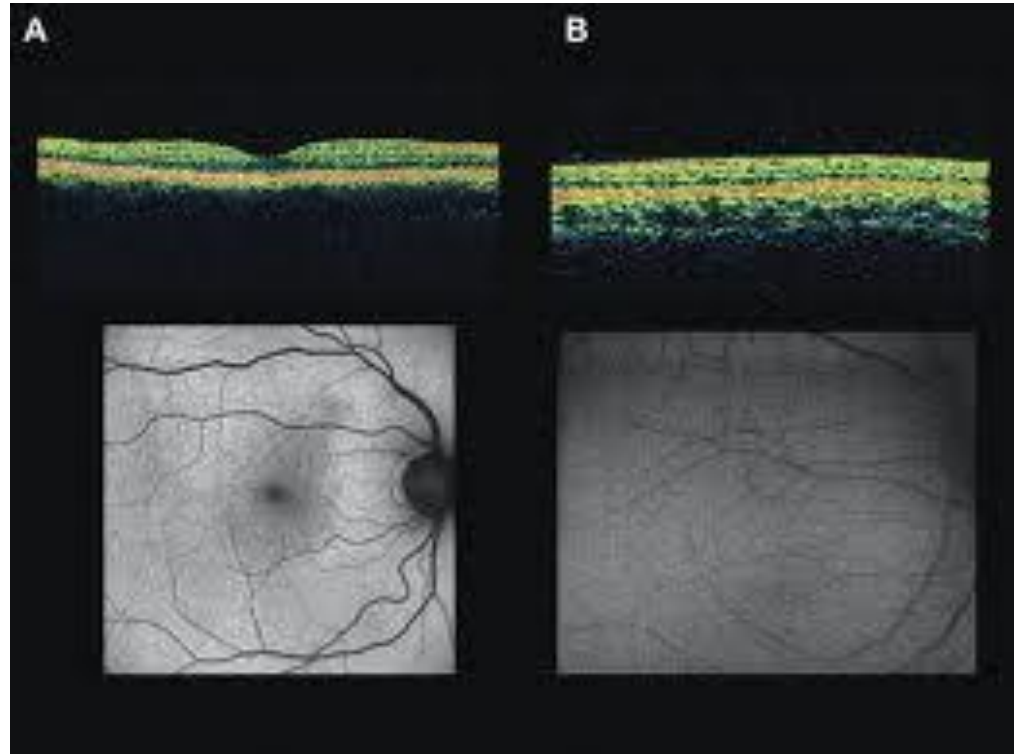
Normally pigmented Fundus



Fundal hypopigmentation

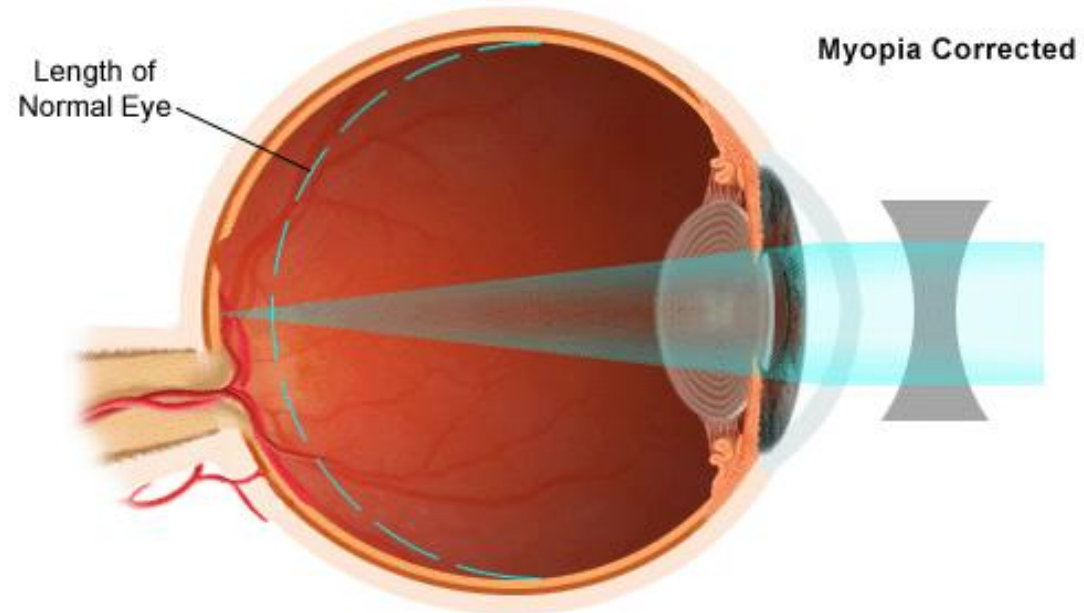
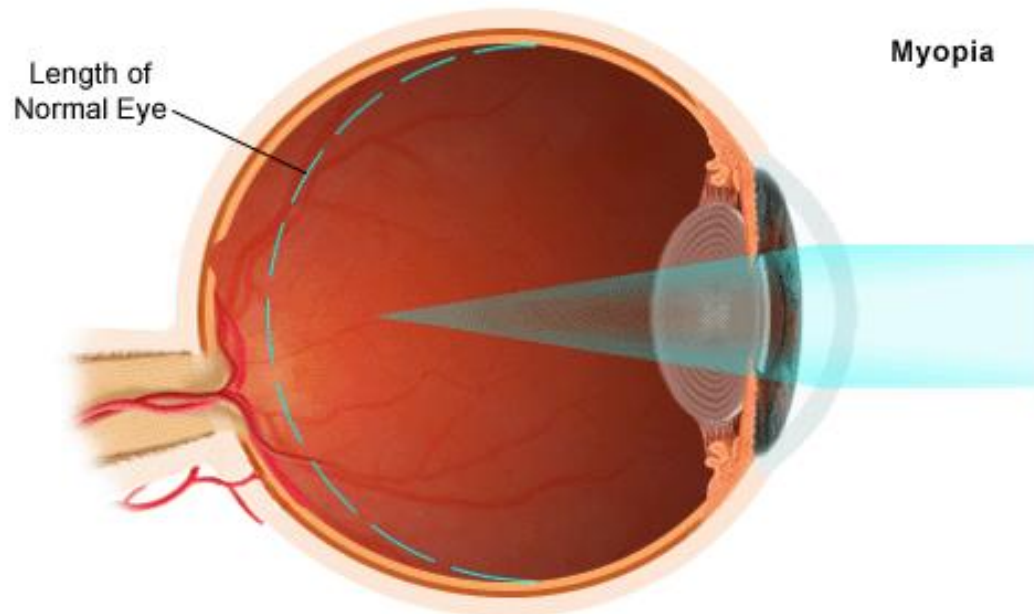


Foveal hypoplasia



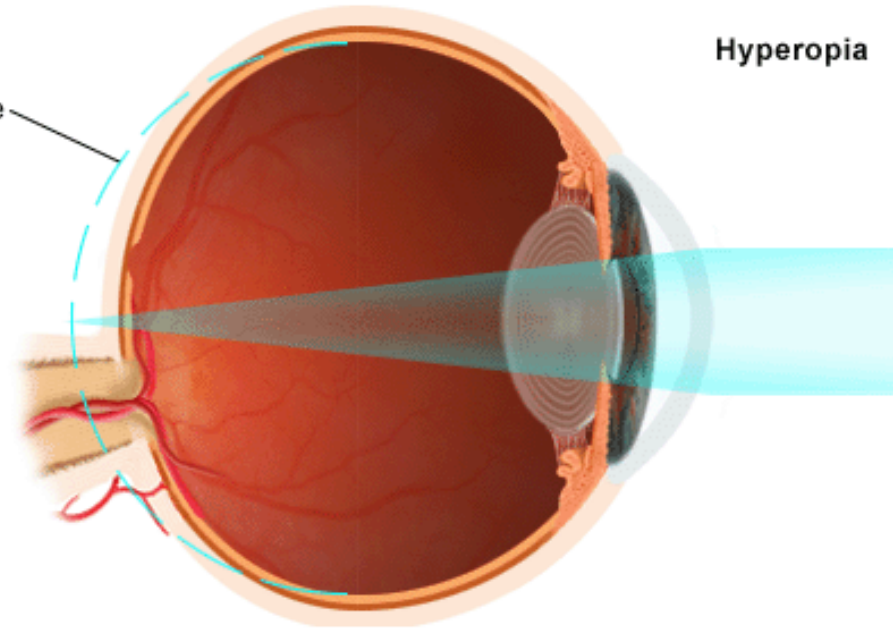
Refractive error

- Myopia = short sightedness = **eye ball is too long**
- Hypermetropia = long sightedness = **eye ball is too short**
- Most babies are born long sighted



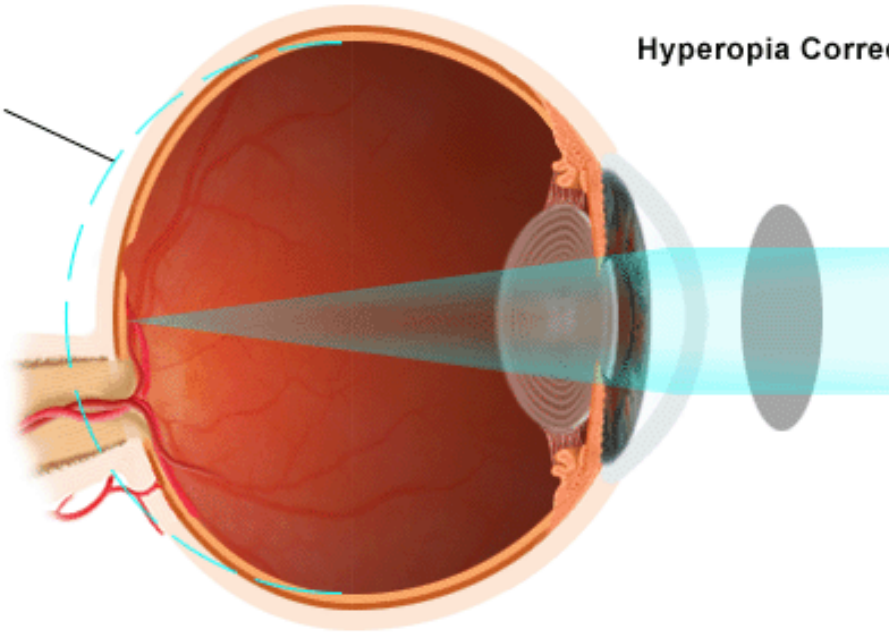
Length of Normal Eye

Hyperopia

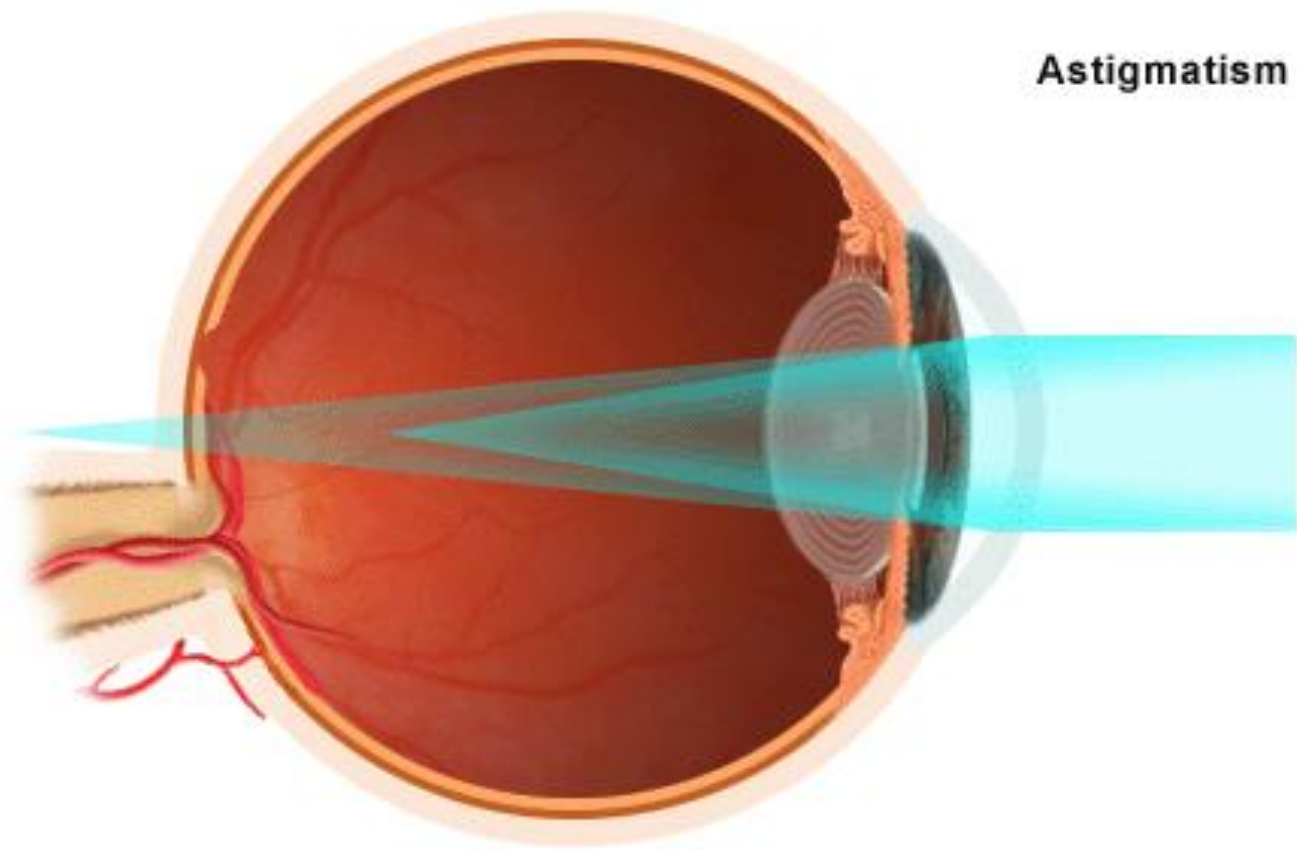


Length of Normal Eye

Hyperopia Corrected

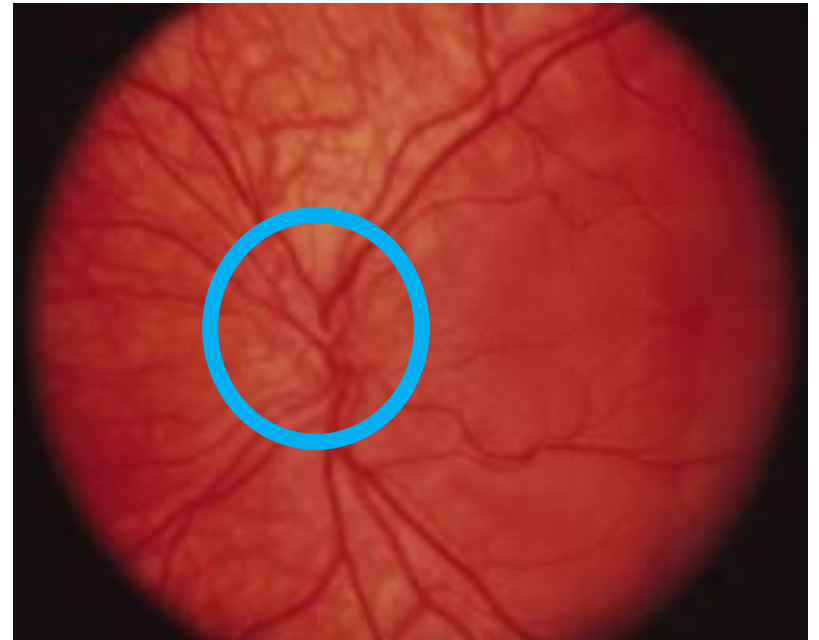
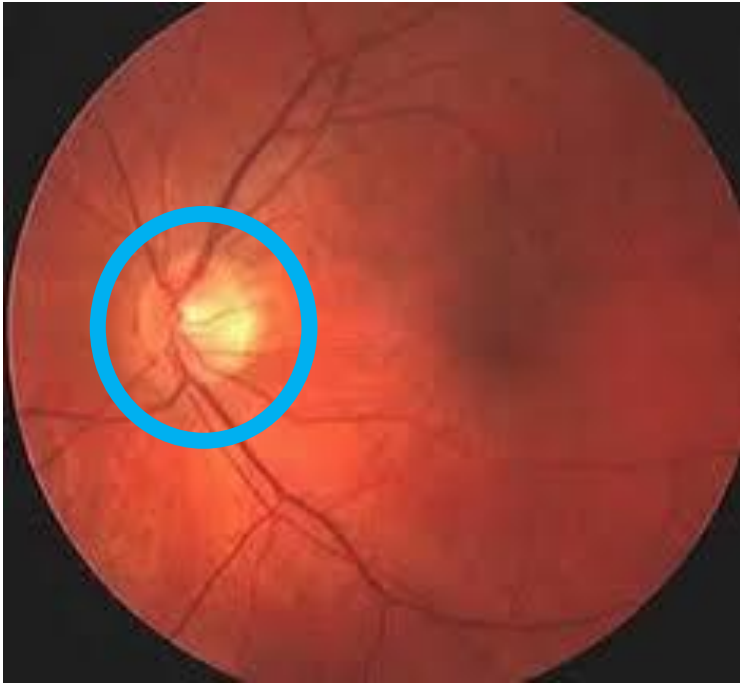


Astigmatism





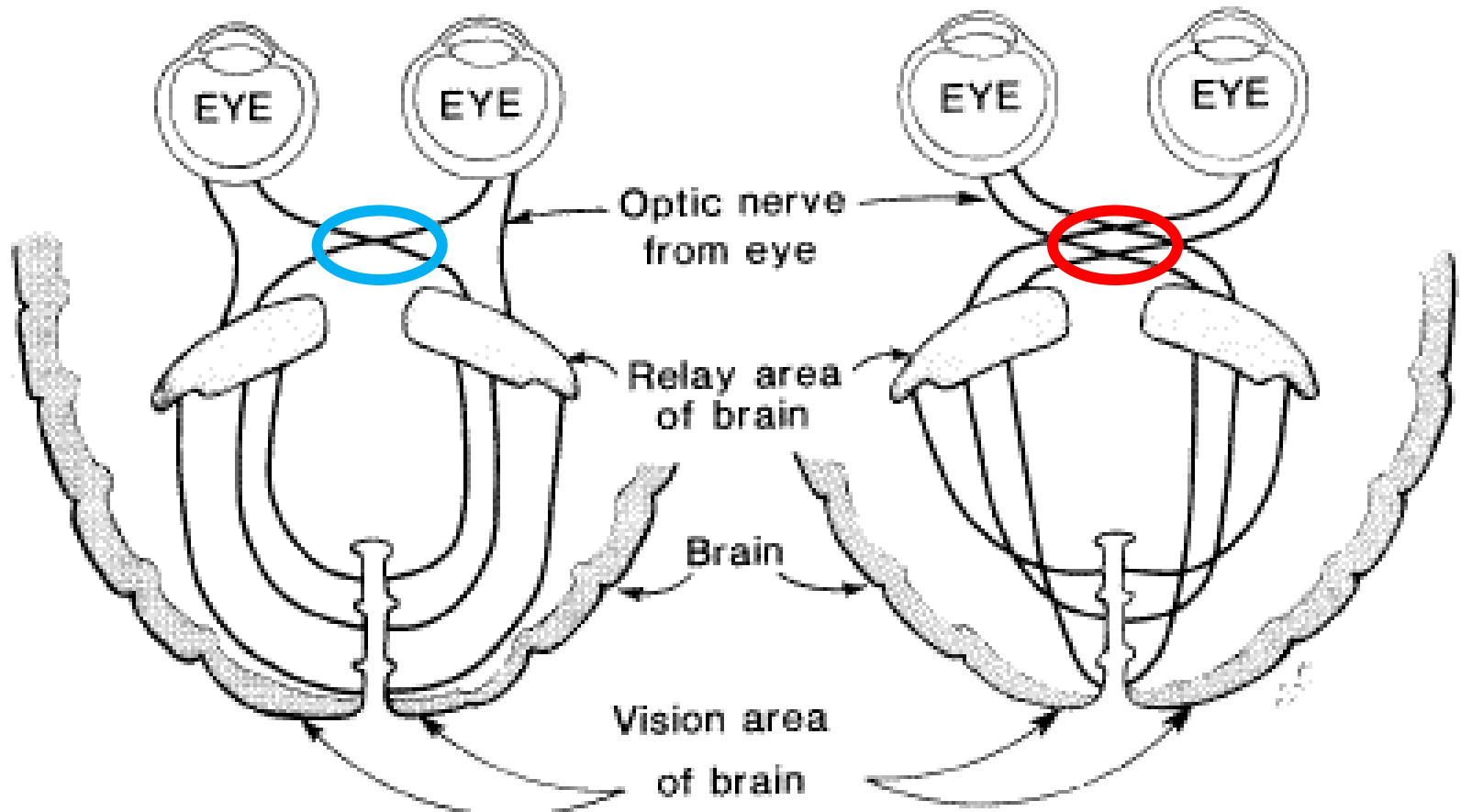
Optic nerve hypoplasia



Abnormal decussation of the visual pathways

**EYES AND BRAIN IN
NORMAL PIGMENTED PERSON**

**EYES AND BRAIN IN
PERSON WITH ALBINISM**



Strabismus



Exotropia (eye turns out)



Esotropia (eye turns in)

Nystagmus and head turn



How is a diagnosis made?

- History
- Examination
- Tests- cycloplegic refraction, OCT, VEP
- Time and re-examination
- Looking at other family members colouring and eyes.

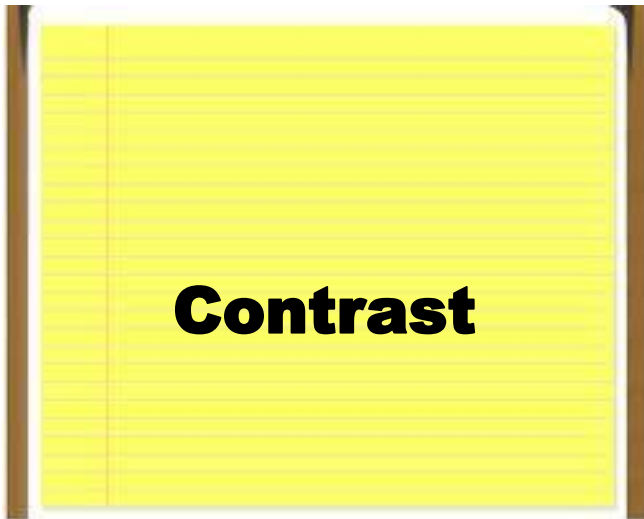
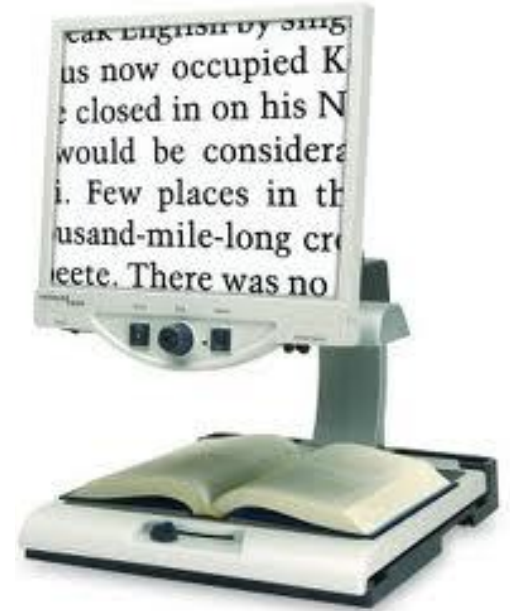
Managing the condition

- Glasses and contact lenses
- Low vision aids
- Protect the eyes and skin from ultraviolet light

Glasses and Contact Lenses



Low visual aids





SUN SAFETY

GLUG
GLUG GLUG



Slip Slop Slap

REHYDRATE

What can a person with albinism see?

- It is a spectrum.
- Evidence shows that those with more severe foveal hypoplasia have higher refractive errors and poorer vision.

Nystagmus

- Concentration, being tired or stressed makes nystagmus worse

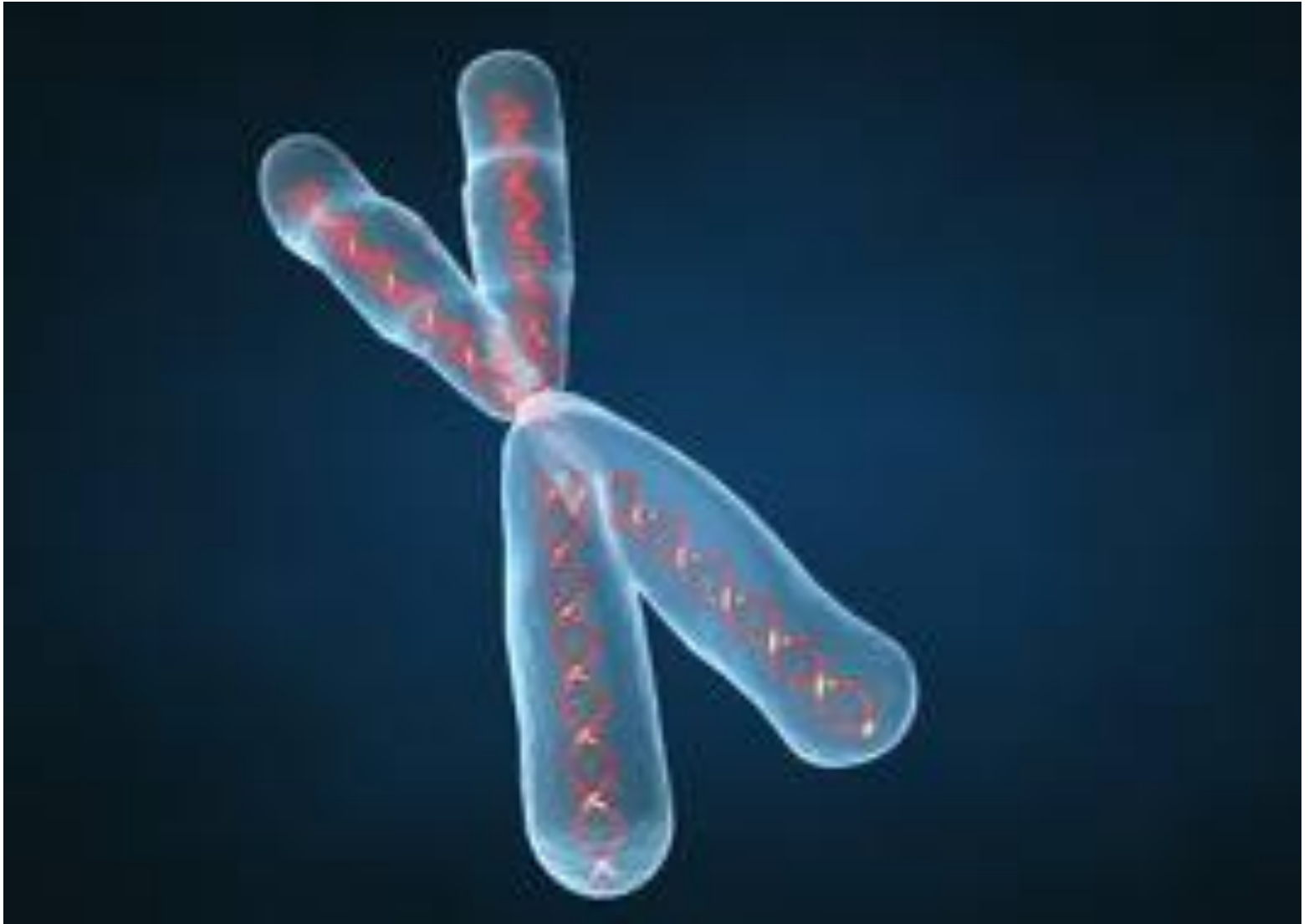
What can a person with albinism do?

- May not be able to drive a car.
- USA study of 78 children, normal neurodevelopment, behaviour, academic achievement regardless of visual acuity.

Why does this happen

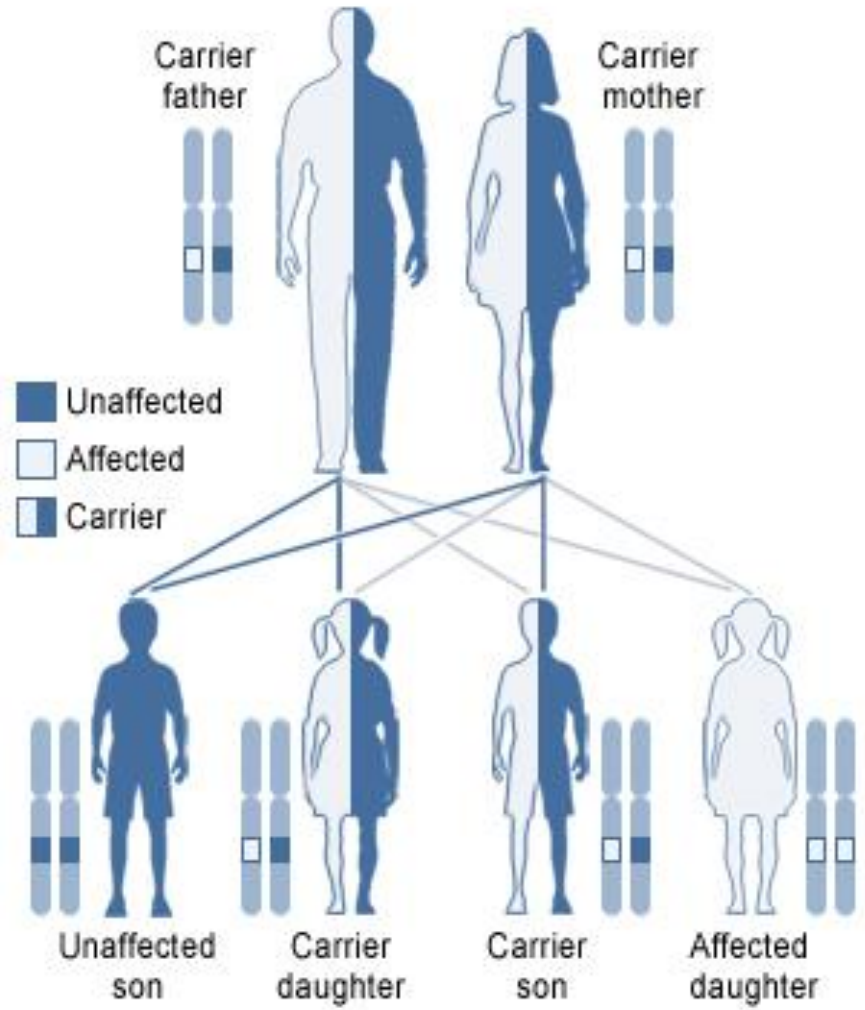
- Genetics





Autosomal Recessive

Autosomal recessive



Albinism

Oculocutaneous

Ocular

OCA1

OCA2

OCA3

OCA4

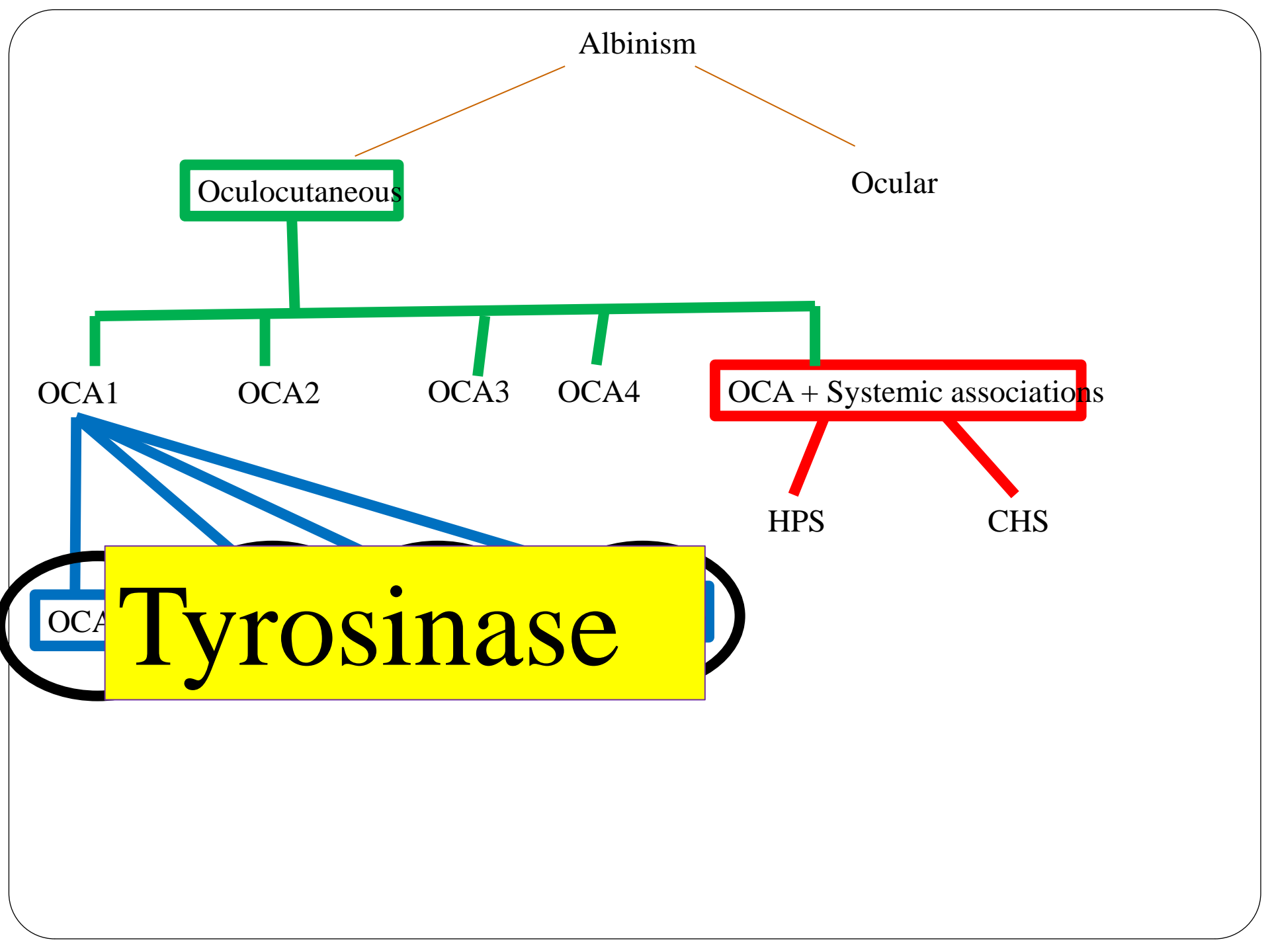
OCA + Systemic associations

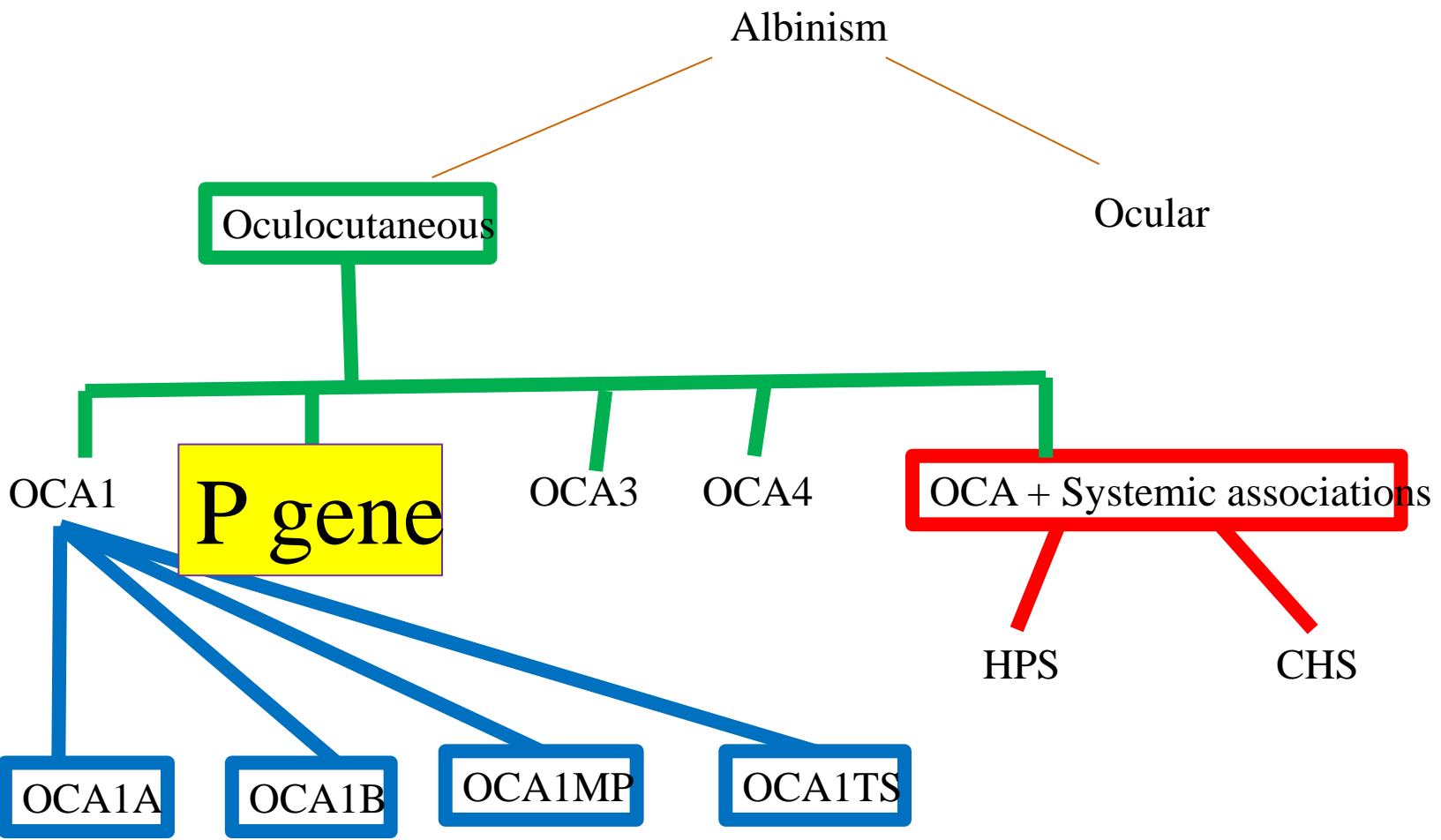
HPS

CHS

OCA

Tyrosinase





Albinism

Oculocutaneous

Ocular

OCA1

Tyrosine related protein

Genetic associations

CHS

OCA1A

OCA1B

OCA1MP

OCA1TS

Albinism

Oculocutaneous

Ocular

Membrane associated transport protein

OCA1

OCA1A

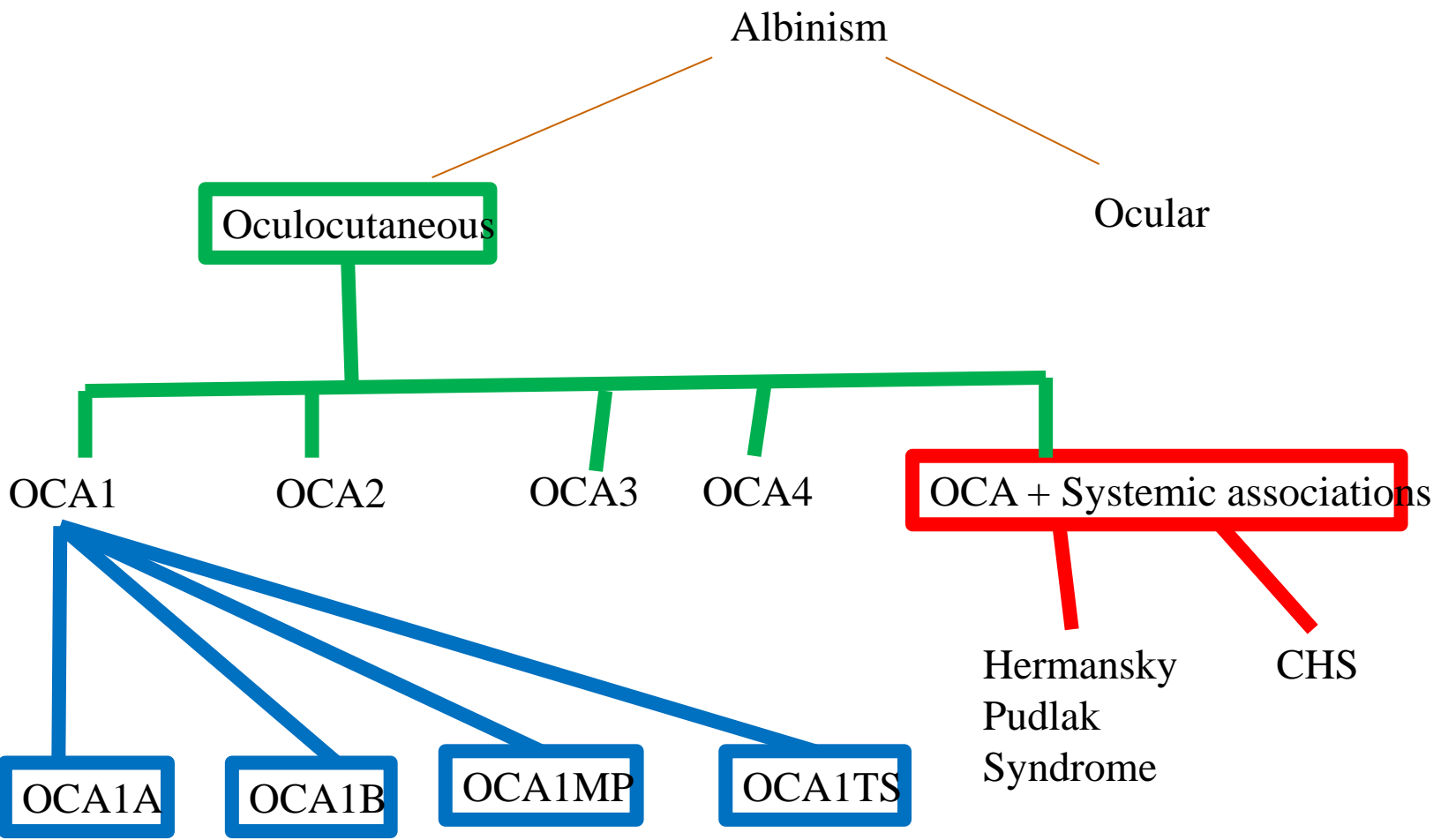
OCA1B

OCA1MP

OCA1TS

HPS

CHS



Albinism

Oculocutaneous

Ocular

OCA1

OCA2

OCA3

OCA4

OCA + Systemic associations

OCA1A

OCA1B

OCA1MP

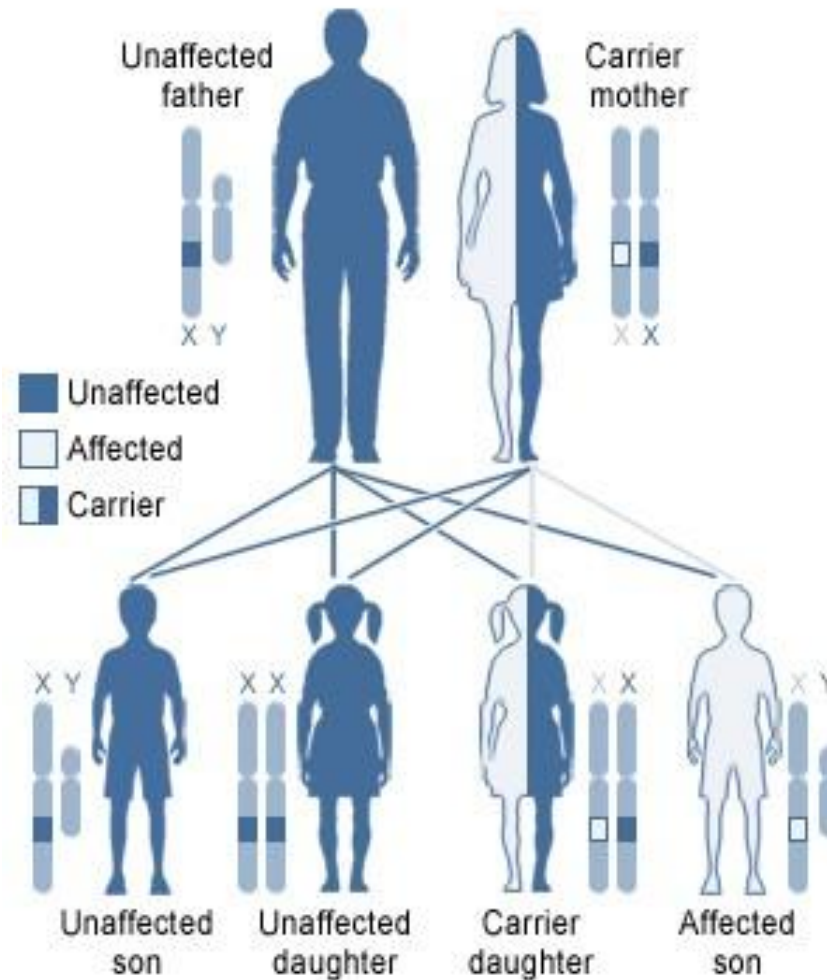
OCA1TS

HPS

Chediak Higashi Syndrome

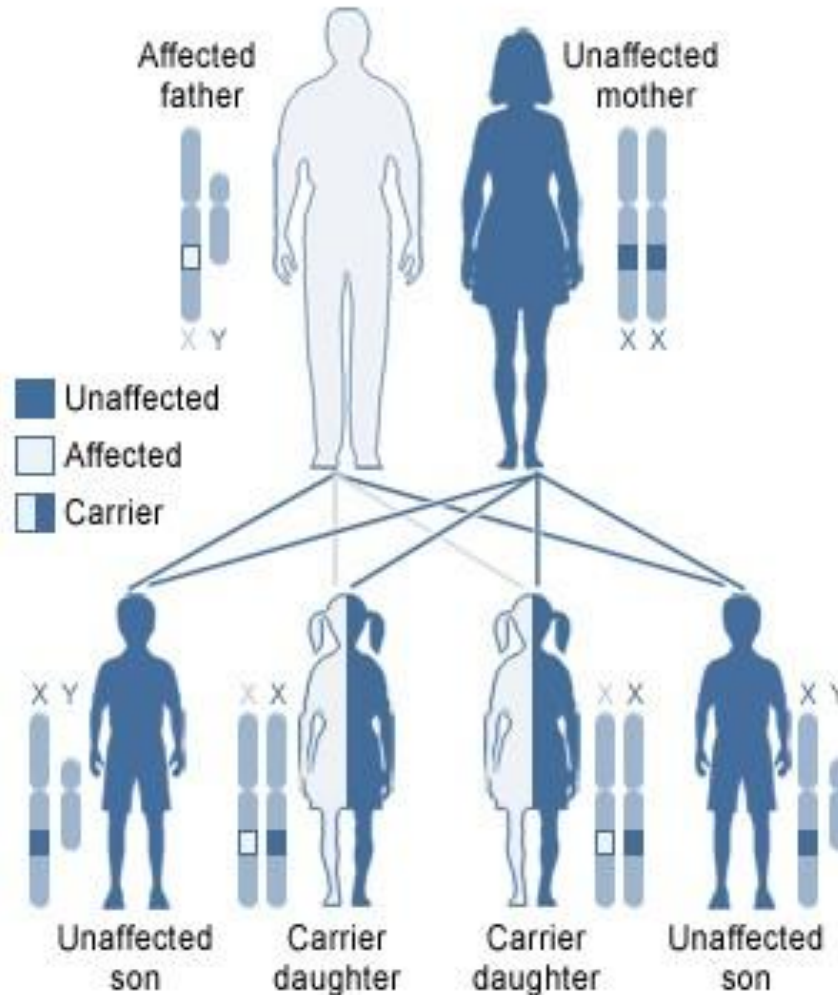
X linked Recessive

X-linked recessive, carrier mother



X linked Recessive

X-linked recessive, affected father



Albinism

Oculocutaneous

**G protein
receptor**

OCA1

OCA2

OCA3

OCA4

OCA + Systemic associations

HPS

CHS

OCA1A

OCA1B

OCA1MP

OCA1TS

