Colorblindness

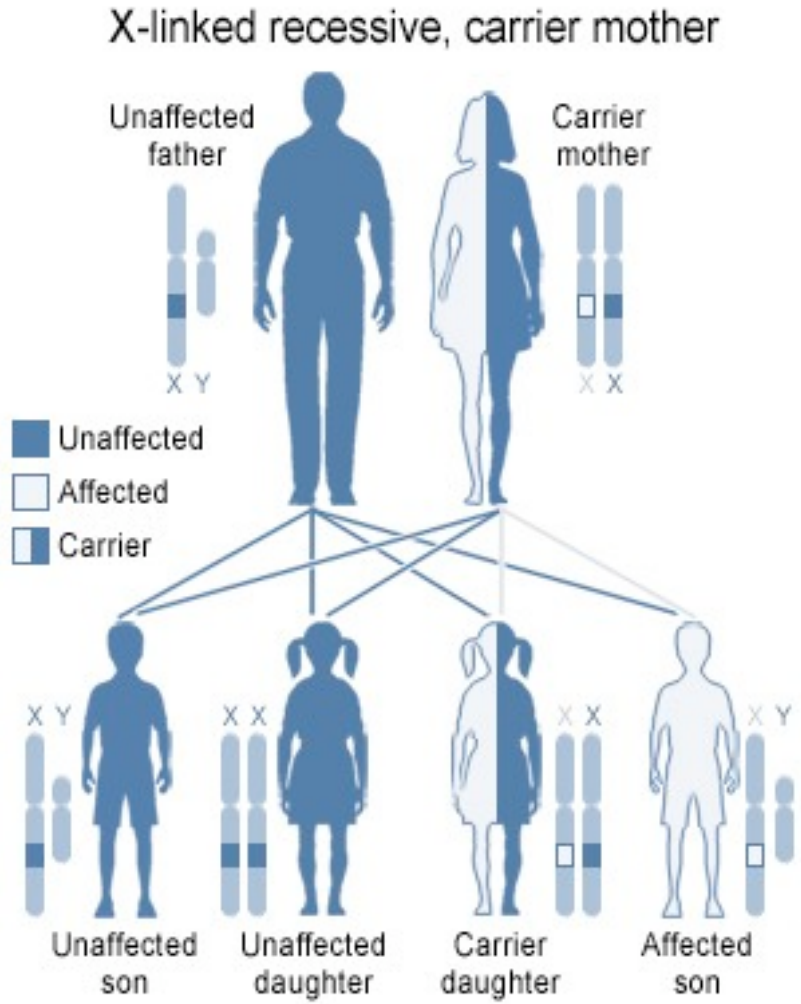
Presentation by Steven Sandoval

Types

- Mutations in >19 different chromosomes may cause color blindness.
  - Red-green.
  - Blue-yellow.
- Mutations affect presence/effectiveness of:
  - Rods - contain color-sensitive pigments

Inheritance

- Deuteranopia: "Green Colorblindness"
  - Sex-linked: Common in males
  - Located on X chromosome
- One of the first genes to be mapped to a specific chromosome.

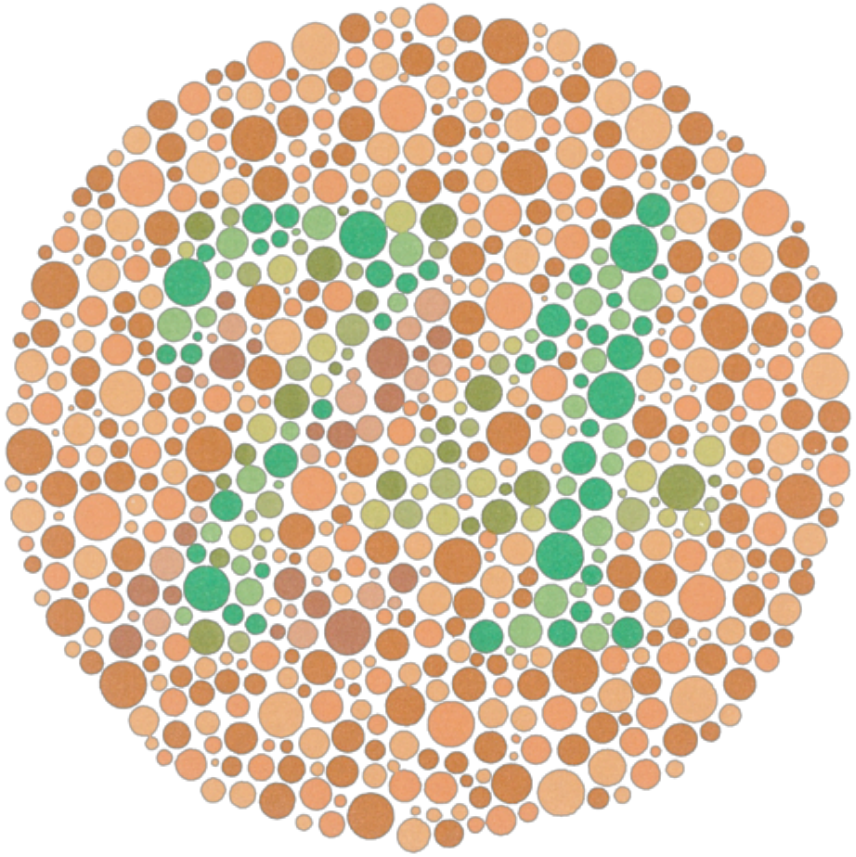

X-linked recessive, carrier mother
Unaffected father
Carrier mother
X Y
X X
Unaffected
Affected
Carrier
X Y
X X
X X
X Y
Unaffected son
Unaffected daughter
Carrier daughter
Affected son
U.S. National Library of Medicine

Diagnosis

- Ishihara Color Test
- Genetic Testing (Research Only)

Gene Symbol	Chromosomal Locus	Protein Name
OPN1LW	Xq28	Red-sensitive opsin
OPN1MW	Xq28	Green-sensitive opsin

Ishihara Color Test



Pseudoisochromatic Plates

Example: Pingelap

- Catastrophic typhoon in 18th century
- 10% population total color blind
- 30% population are carriers
- Appearance of phenotype discernible 4 generations from single ancestor (typhoon survivor: Nahnmwarki Mwanenised)

Treatment

- No genetic treatment available
- Early detection in high school
- Tinted contact lenses