The Human Visual System

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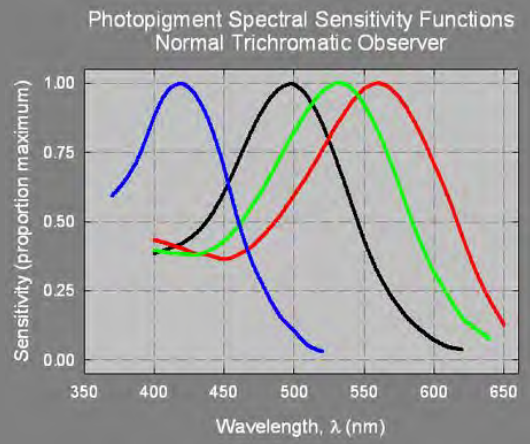
The genetics of trichromacy in humans

- In humans, in general three genes govern the synthesis of the three cone opsins:

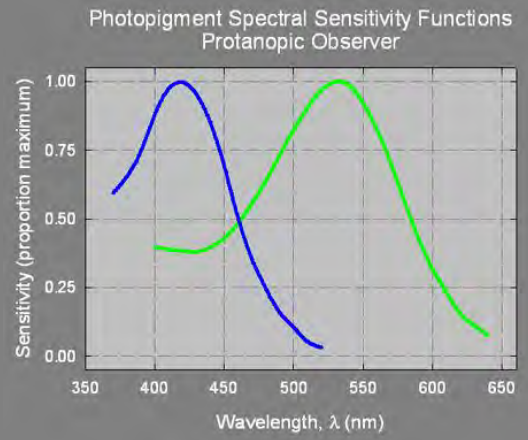
blue (cyanolabe, short-wavelength, λ_m=420)
green (chlorolabe, medium-wavelength, λ_m=530)
red (erythrolabe, long-wavelength, λ_m=560).

- The SW gene (OPN1SW) is located on chromosome 6 (autosomal)
- The MW and LW genes (OPN1MW and OPN1LW) are located on the X chromosome.
  - This is why more men than women are color-defective.

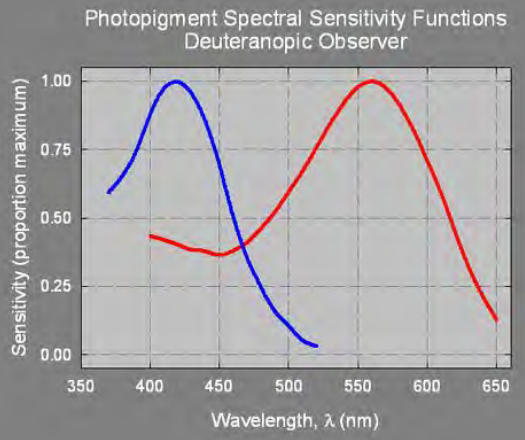
Types of “color blindness”



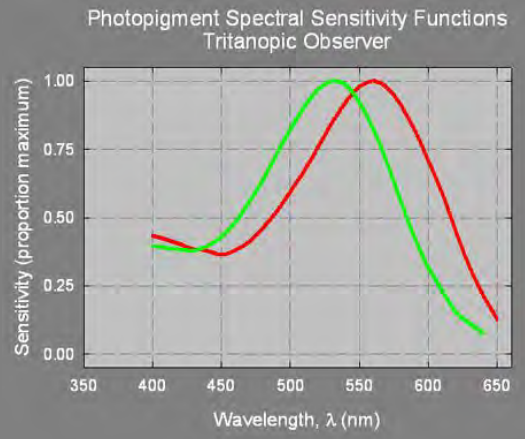
Normal trichromat



Protoanopic dichromat (“red-blind”)



Deuteranopic dichromat (“green-blind”)

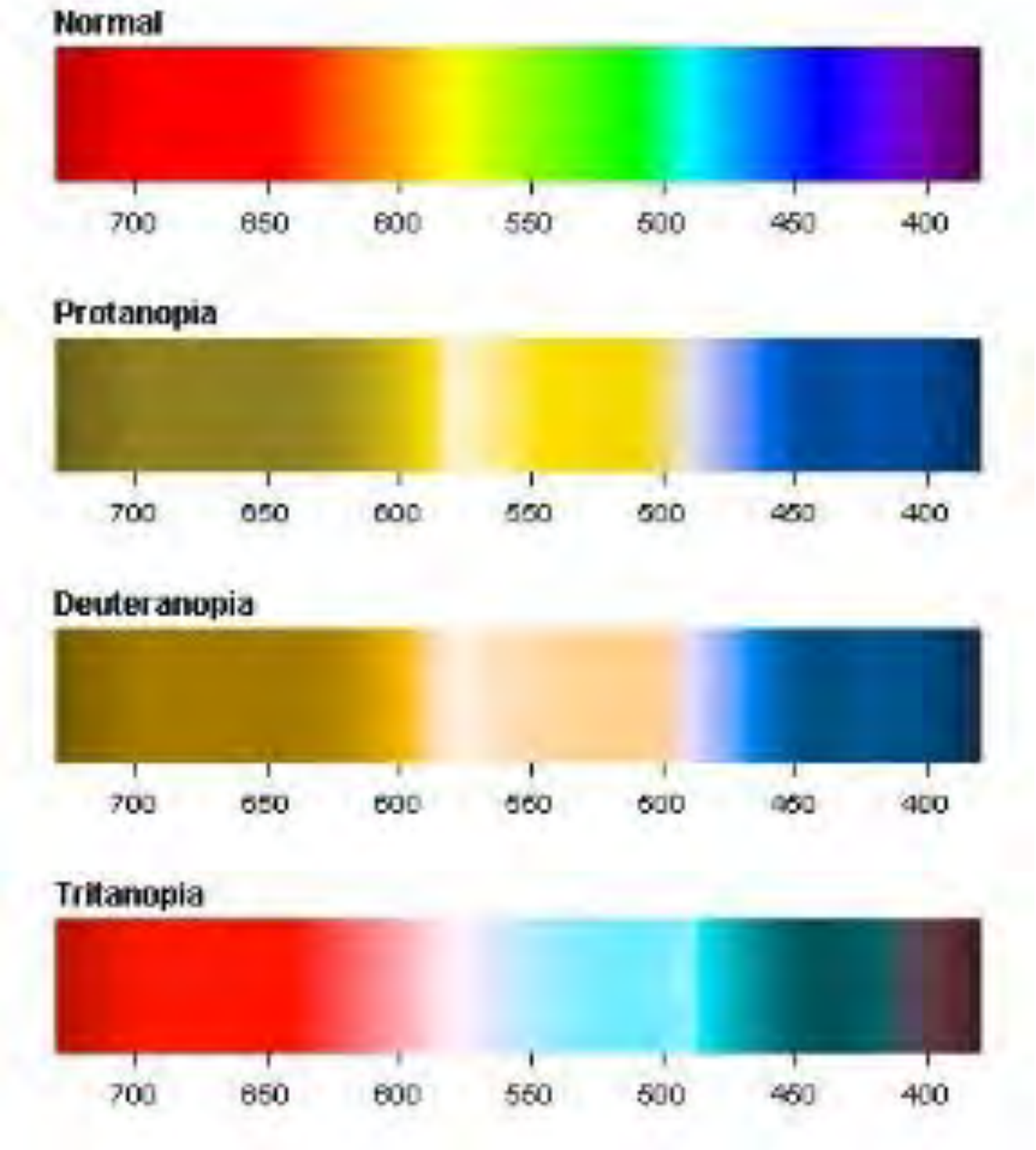


Tritanopic dichromat (“blue-blind”)

Color deficiency statistics

- 1 in 12 people have some sort of color deficiency. About 8% of men and 0.4% of women in the US.
- 0.38% of women are deuteranomalous (around 95% of all color deficient women).
- 0.005% of the population are totally color blind.
- 0.003% of the population have tritanopia. (tritanopia is not sex-linked).
- Protanomaly occurs in about 1% of males.
- Deuteranomaly occurs in about 5% of males. It's the most common color deficiency.

The spectrum according to various observers



The world according to…

…a protanope

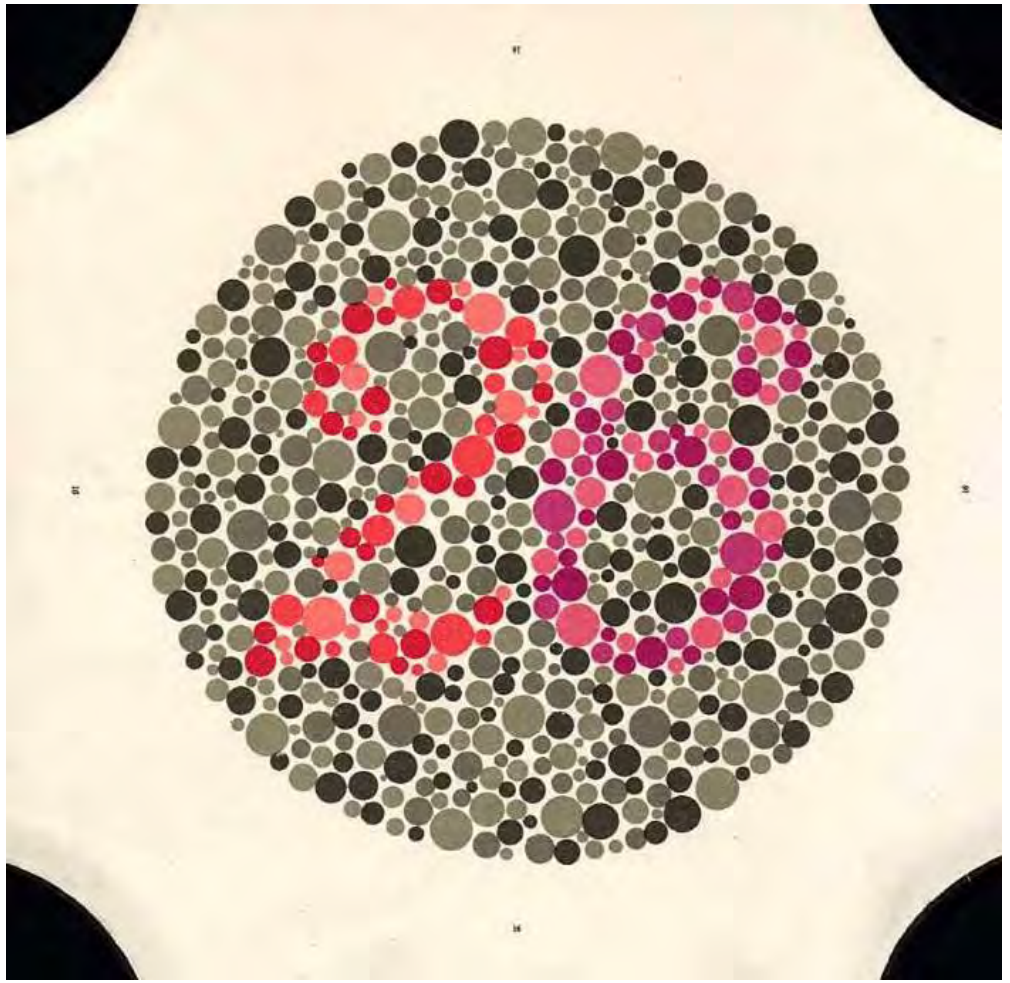
… a deuteranope…

… and a tritanope.

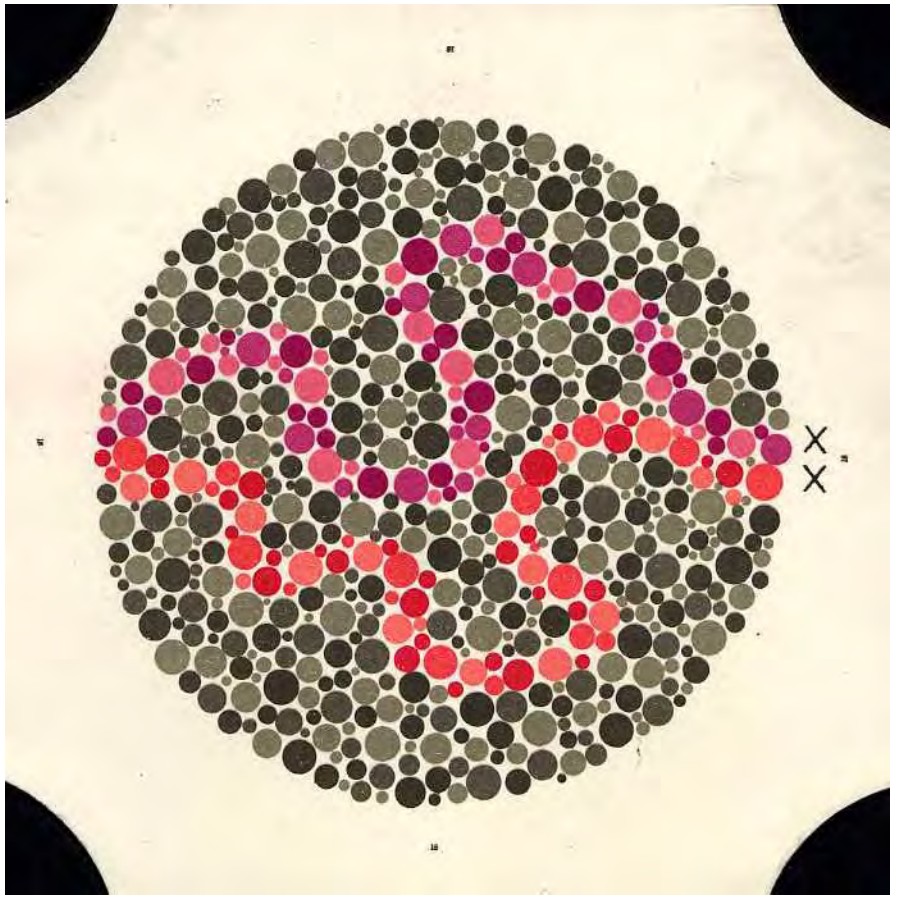
Dichromatic and trichromatic views of the world

Ishihara Plate 1

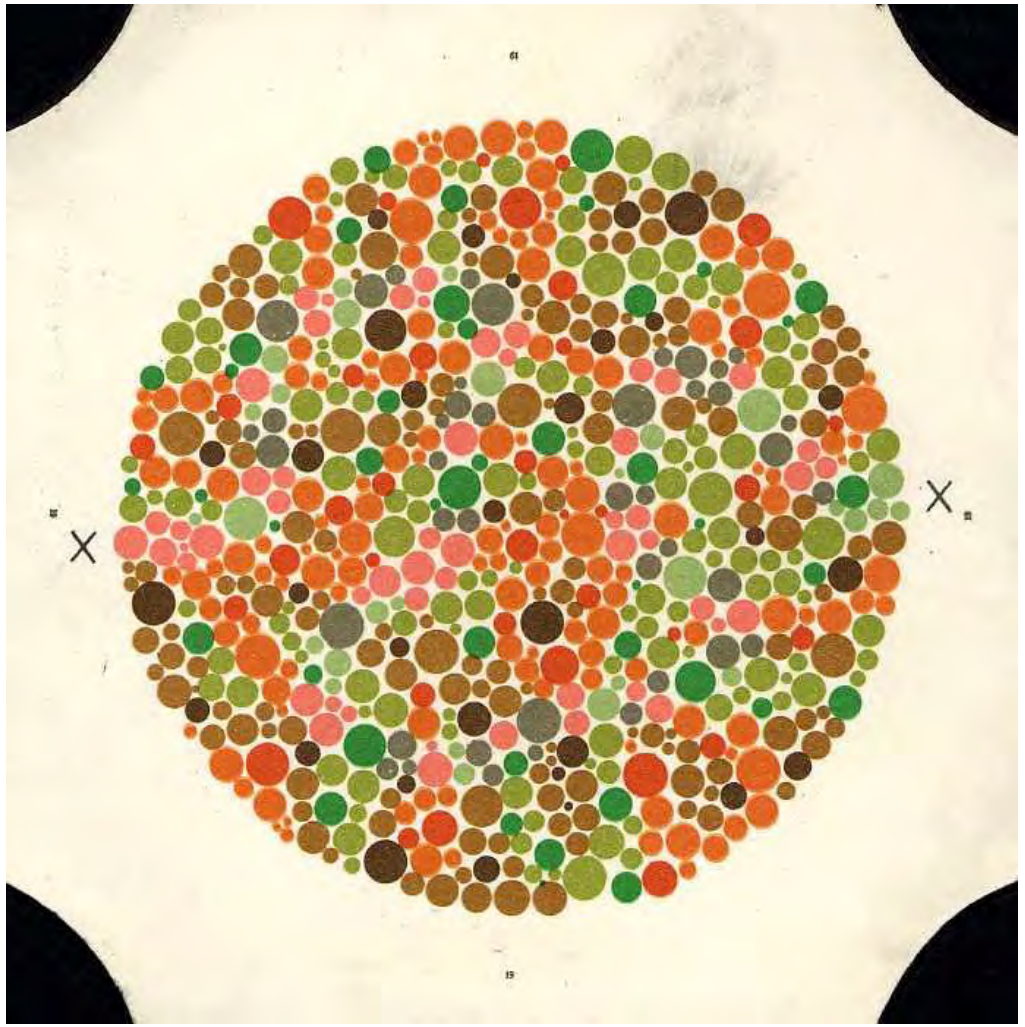
Ishihara Plate 16



Ishihara Plate 18



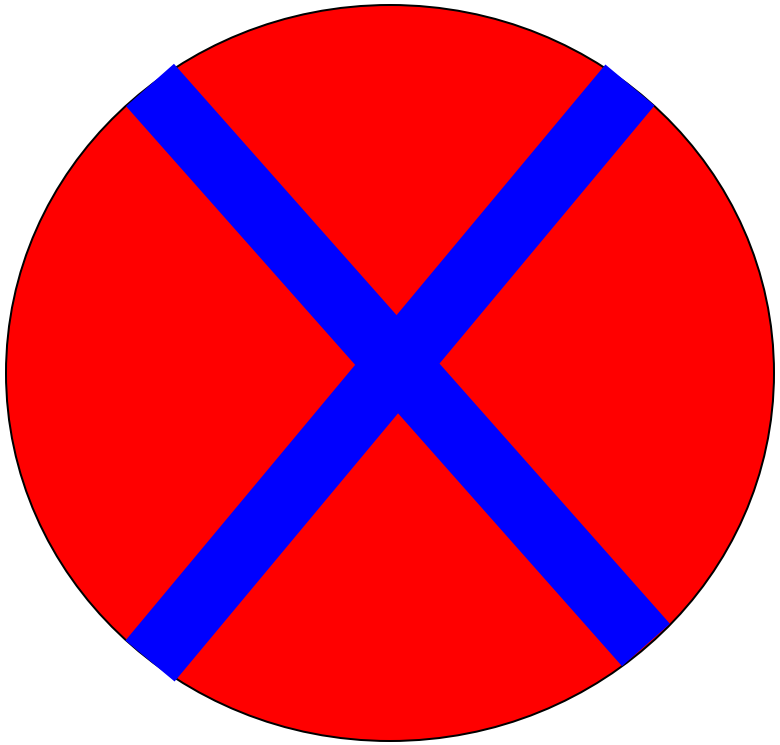
Ishihara Plate 19



Blue cone anomalies and evolution

- They constitute only about 2% of the total number and are found outside the fovea centralis where the green and red cones are concentrated.
- Although they are much more light sensitive than the green and red cones, it is not enough to overcome their disadvantage in numbers. However, the blue sensitivity of our final visual perception is comparable to that of red and green, suggesting that there is a somewhat selective "blue amplifier" somewhere in the visual processing in the brain.
- The visual perception of intensely blue objects is less distinct than the perception of objects of red and green. This reduced acuity is attributed to two effects.
  - The blue cones are outside the fovea, where the close-packed cones give the greatest resolution.
  - The refractive index for blue light is enough different from red and green that when they are in focus, the blue is slightly out of focus (chromatic aberration).

Effects of chromatic aberration



It’s hard to see blue letters on a red background

Or red letters on a blue background

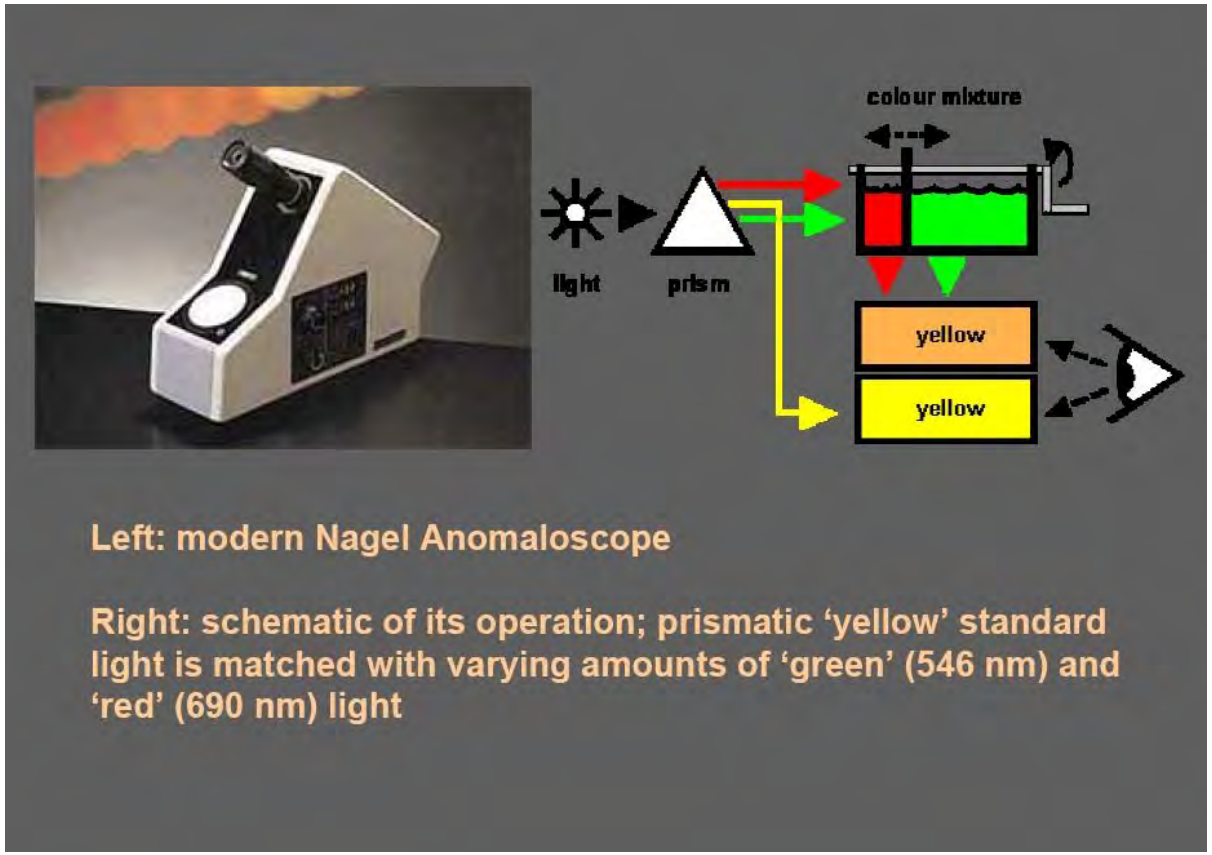
Genetics and color-vision anomalies

- Achromatopsia (rod monochromatism). Three genetic causes (autosomal)
  - Mutant CNGA3 gene, which codes for the α subunit of the sodium channel in the cones.
  - Mutant CNGB3 gene, which codes for the β subunit. (Note Pingelipese islanders; 1775 typhoon bottlenecked the population to 20; now 5-10% have mutant CNGB3, a >1000-fold increase over Europeans)
  - Mutant CNAT2 gene, which codes for the α subunit of cone-specific transducin (the G-protein associated with rhodopsin).

Genetics and color-vision anomalies

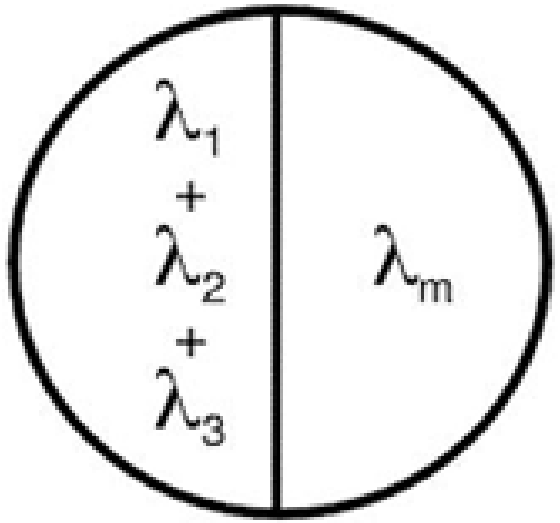
- Blue-cone monochromatism: two causes
  - Mutations in both the MW and LW genes
  - Mutation in the LCR (locus control region) upstream of the MW and LW genes, preventing transcription.

Rayleigh matches and polymorphism

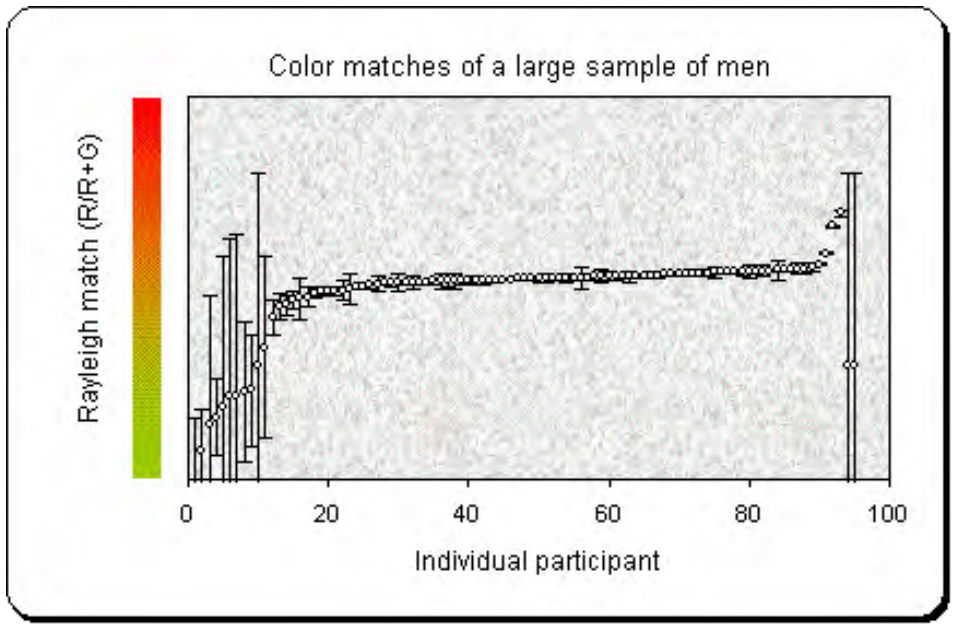


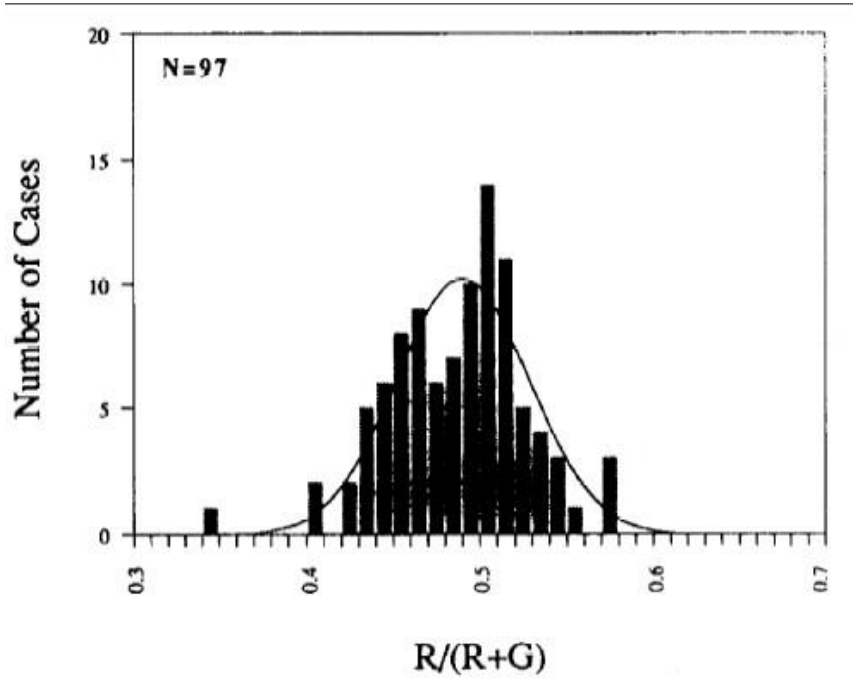
Left: modern Nagel Anomaloscope

Right: schematic of its operation; prismatic 'yellow' standard light is matched with varying amounts of 'green' (546 nm) and 'red' (690 nm) light



Rayleigh matches and polymorphism (cont'd)

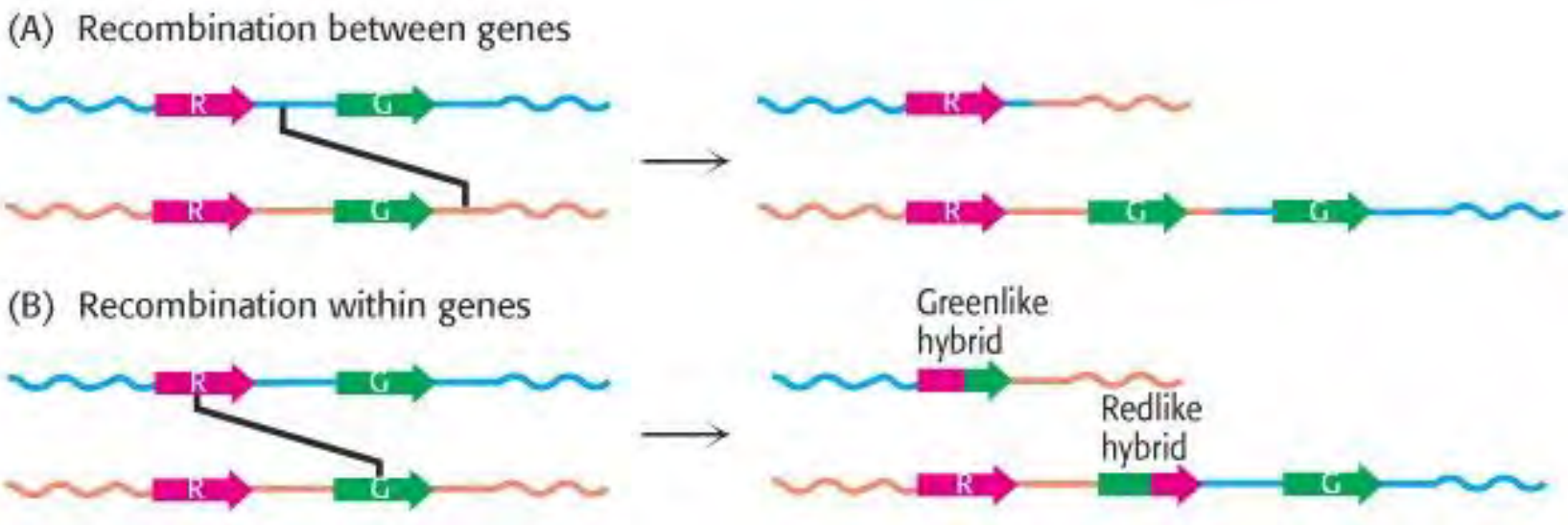




What does the distribution mean?

- Not everyone has the same pigments.
- The red and green pigment genes are polymorphic.
- Many of the LW genes contain MW sequences, from unequal crossover at meiosis I, between X chromosomes.

Crossover anomalies



Some numbers

- More than 50% of humans have two or more LW genes on their X chromosome.
- As many as 2/3 have fusion (LW+MW) genes, where the LW gene contains substantial MW sequences.
  - Virtually all LW genes on the X chromosome except for the first one are fusion genes.
  - Yet even where the only “LW” gene is a hybrid, color vision is normal.

Color vision and evolution

- Fish, birds and reptiles have up to four photopigments, but mammals, except for old-world monkeys and humans, have only two (autosomal SW and X-linked LW)
  - (did we lose one when primates were primarily nocturnal??)
- New-world howler monkeys have a kind of trichromacy, based on a polymorphic LW gene.
  - heterozygote females are trichromatic.