

Chapter 3, Tovée

Retinal Color Vision

Why do we need more than one cone pigment?

Humans (& Old World primates) have 3 cone classes

Red (565 nm absorption maximum)

Green (530 nm absorption maximum)

Blue (420 nm absorption maximum)

To distinguish colors two or more cones must be compared

A single cone cannot distinguish between 2 colors and one color at 2 intensities (*Univariance*)

Increases in number of cone classes increases color discrimination

Nonprimate animals have 2 cone classes

Birds have 5 cone classes

Trichromacy

Trichromatic theory for human vision-- developed by Thomas Young (1802)

All colors can be represented as a combination of three colors

Trichromatic theory can't explain why only some colors blend

Bluish-green---possible

Yellowish-red--possible

Greenish-red--not possible

Yellowish-green--not possible

Alternative theory ---Opponent interaction theory (Ewald Hering)

Comparison is made in an opponent interaction

Color Opponency--

-Red-Green or Green-Red center surround organization

-Blue-(Red + Green=Yellow) or Yellow-Blue center surround

Luminance detection does not depend on color (achromatic)

Color opponency arise without prewired pattern of cones in retina

Red and green cones are randomly arranged in the retina

Blue cones (small density) form a ring around fovea

Non primate mammals have only yellow cones in central fovea

Achromatic central vision

Primates have an extra red cone giving color vision

Red and green cone absorbance maxima are necessarily similar

Central red and green cones provide information about luminance

Luminance differences provide information about scene detail

Similar absorption peaks allow each cone to detect similar luminances to a single color → improves luminance detection
Small differences in absorption maxima ensure little chromatic aberration

Light of different frequencies are focused at different distances (eg. eye focal length varies with wavelength)
Chromatic aberration causes image degradation

The genetics of visual pigments

Color blindness arises from genetic variations

Types of color blindness--

Missing Pigments (or cones containing pigment)

◆ Tritanope--missing blue pigment

◆ Deuteranope--missing green pigment

John Dalton & his brother were deuteranopes

Determined by DNA analysis of his preserved eyes

◆ Protanope--missing red pigment

Altered pigments (Anomaly)

◆ Tritanomaly--altered blue pigment

◆ Deuteranomaly-- altered green

◆ Protanomaly-- altered red

Molecular genetic studies provide insight in genes for cone pigments

Anomalous pigments are caused by many absorption differences

A continuum of absorption maximums are observed

Normal absorption maxima are the most probable ones

Some women may have 4 cones

Congenital color blindness is common in men; rare in women

Occurance--8% men, 0.5% women

Red-green abnormalities--common

Blue abnormality-----rare

Acquired color blindness

Blue cone loss is most common

Blue cones sensitive to high light intensities

Blue cones are sensitive to oxygen deprivation

Inheritance of red & green pigments is sex-linked

Inheritance of blue cones is not sex linked (autosomal dominant)

Organization of genes for pigments

Pigment = opsin + retinal (opsin determines absorption spectra)

Chromosomal location of genes

Blue pigment--chromosome 7

Red pigment--chromosome 3

Green + red-- chromosome X (tandem array of green-red)

Green gene shows 97% similarity to red gene

Upstream promotor regions are similar

Green gene shows 40% similarity to red gene

Unequal crossing over during meiosis causes abnormalities

One X chromosome may carry 2 green genes and 1 red gene

Males who inherit these genes will be normal

Other X chromosome carries 1 red gene and no green genes

Males who inherit this gene will be deuteranopes

Green genes on X-chromosome varies between individuals

Number of green genes varies between 1 to 6; 2 is most common

Green (or red) genes possess 6 exons and 5 introns

Differences between red and green genes arise in exons 2-5

Green exons 1-5 + red exon 6 = hybrid gene (green pigment)

Substitutions in exons 2,3 or 4 cause 2-5 nm absorption shifts

Substitution of exon 5 cause 15-21 nm absorption shift

Anomalous trichromats possess a hybrid gene

Hybrid gene is extra (besides red and green gene)

Hybrid gene substitutes for red or green pigment genes

Hybrid gene formed by unequal crossing over (recombination)

Anomalous trichromat --

One (or more) green genes +

Hybrid gene

Substitutions in exons 2,3,4, or 5 (or any combination of exons)

Not all pigment genes on X-chromosome are expressed

Particular genes in tandem sequence are preferentially expressed

Achromatic vision is rare (missing red and green pigments) [1/100,000]

Possible explanations--

1. Green gene deleted by unequal recombination event

A mutation in red gene

2. Deletion of gene(s) necessary for the function of the red & green cones

3. Deletion of gene(s) needed for red & green gene transcription

Blue cone pigment

Blue cone pigments are inherited autosomally on Chr 7 (not sex linked)

Blue pigment gene is dominant

Loss of blue pigment can be caused by mutations at 2 sites of gene
Mutations in blue gene cause changes in blue opsin

Rhodopsin, night blindness and retinitis pigmentosa

Changes in rod function cause--

- ◆ Night blindness (sensitivity to light is reduced)
- ◆ Retinitis pigmentosa (degeneration of rods & retina=complete blindness)

Progression of disease--

- Death of rods
- Death of cones (constriction of visual fields)
- Loss of retinal blood flow & retinal thinning
- Melanin pigment deposition in the retina

Inheritance of night blindness and retinitis pigmentosa

Autosomal dominant trait

Abnormal opsin in rhodopsin

Abnormal opsin folding (Sung et al., 1991; Rao et al., 1994)

Abnormal breakdown of bleached rhodopsin

Activated opsins permanently stimulate the cell til death occurs

Night blindness mutation is less severe & doesn't lead to cell death

Better color vision in women?

X-chromosome mutations in red & green genes could lead to 4-5 pigments

- Females possess 2 X-chromosomes
- Mutations in red & green genes would produce different pigments

Conditions for possessing a usable 4 or 5 color visual system

- Anomalous & normal pigments must be produced in separate cells
- Comparison circuitry must exist (center-surround opponency cells)
- Comparisons are used by visual cortex network to perceive color

Female tetrachromats may exist (Jordan & Mollon, 1993)

Three pigments in normal human color vision?

Non polar opsin amino acids substitution for polar ones=absorption shifts
(amino acids containing -OH are polar)

Work of Neitz, Neitz & Jacobs (1991)--

- Exon 3> Nonpolar substitution at residue 180 \Rightarrow 5.3 nm increase
- Exon 5> Nonpolar substitution at residue 277 \Rightarrow 9.5 nm increase
- Exon 5> Nonpolar substitution at residue 285 \Rightarrow 15.5 nm increase

Work of Merbs & Nathans, (1992a,b)--

- Exon 4> Nonpolar substitution at residue 233 \Rightarrow 20 nm increase
- Residue 233 substitutions modulate changes at 180, 277, & 285

Work of Asjano, Rim & Oprian (1994)--

Seven residues (out of 364) play key role in red/green absorption

Key amino acid residues--

116

180

277

285

230

233

309

Color blindness test is bimodal Rayleigh distribution⇒ two red pigments

Rayleigh match--ratio of red to green to match orange

Distribution of ratios arises from two distributions (bimodal)±

two red pigments

Microspectrophotometric absorption measurements of 1 red cone

Two red pigments are observed (Darnall et al., 1983)

Serine/alanine (180) cause changes consistent with two red pigments

(Merbs & Nathans, 1992; Winderickx et al., 1992b)

Two green pigments were found with serine / alanine (180)

(Neitz, Neitz and Jacobs 1993)

Many other red or green anomalous pigments are likely in population

The evolution of primate color vision

Non-primate animals are dichromats

Blue pigment

Red-green pigment (555 max absorption)

Second gene site on X-chromosome likely arose by gene duplication

Corellates with separation of lineages of Old & New World monkeys

Possible mechanisms for generation of trichromacy

① Point mutation produces two different red-green pigments

② Unequal recombination ⇒ 2 alleles on X-chromosome

③ Mutation causes 2 genes to be expressed in different cones

④ Mutation causes absorption shifts to produce red & green cones

Trichromacy of Old World primates may have selective advantage over dichromacy

