iBiology.org Teaching Tools
Jeremy Nathans' Lecture Part 3:
The Evolution of Color Vision

Teaching Tools were prepared by Jeremy Nathans.

Contents

---

1. Keywords and Terms

evolution, color vision, New World primate, Old World primate, color opponent processing, spectral tuning, X-chromosome inactivation, blue cone monochromacy, locus control region, genetically engineered mice

2. Lecture Notes

Primate evolution: the plate tectonic separation of South America and Africa led to a division into New World and Old World primate lineages. Humans and our great ape relatives are Old World primates.

What is the selective value of trichromatic color vision relative to dichromatic color vision? Possible answers include an improved ability to identify ripe fruit, especially among dappled foliage, and an ability to assess individual differences among members of one's own species.

Non-primate mammals (dogs, cats, mice, cows, etc.) have an S-pigment and a single longer wavelength pigment (encoded on the X-chromosome). The X-chromosome-encoded pigment is the ancestor of the M and L pigments of Old World primates.

The neural circuitry that supported an ancient system of dichromatic color system among non-primate mammals still exists essentially unchanged in the primate retina. Current evidence suggests that the comparison between M and L cones uses a neural circuit that evolved to measure spatial variation in light intensity by comparing the activities among a homogeneous set of photoreceptors at different locations in the retina. These photoreceptors contained a visual pigment that was the ancestor of the present-day primate L and M pigments.

The difference in absorbance spectra between the primate L and M pigments is principally due to amino acid differences at only three locations (out of 364 amino acids total).

The requirement for expression of L and M pigments in distinctive cone types has been solved in two different ways by New and Old World primates.

In most New World primates, the X-chromosome carries only a single cone pigment gene, but sequence variation within this gene in the population creates a series of cone pigments (typically three) that differ in their absorbance spectra. These species use X-chromosome inactivation – a phenomenon common to all female mammals that randomly silences most of the genes on one of the two X-chromosomes – to create a mosaic of spectrally distinctive cone photoreceptors. Of course, this mechanism is only useful for those females who inherited two different alleles of the X-linked visual pigment gene. A striking aspect of this mechanism is that it generates a random mosaic of spectrally distinct cones, since the decision to inactivate one or the other X-chromosome is stochastic.

In Old World primates, the choice between expressing L or M visual pigment genes also appears to be governed by a random process. In this case, current evidence suggests that there is a random choice to activate either the L or the M promoter, but not both, in each cone that is destined to express one of the X-linked visual pigment genes. This mechanism succeeds because the X-chromosomal location of these genes means that this random choice is made on only one chromosome in each cone cell. (Recall that males have only one X-chromosome, and female cells express the L and M pigment genes only from the one active X-chromosome.)

These primate mechanisms of gene choice have been examined by genetic engineering experiments in mice. First, gene expression by random gene choice of either the L or M pigment genes from an human gene array has been reproduced in mice, showing that this phenomenon does not require any primate-specific proteins. Second, by replacing the mouse M pigment gene (located on the X-chromosome) with sequences coding for a human L pigment, a mouse line has been engineered to carry X-chromosome visual pigment gene variation of the type that characterizes New World primates. By color vision testing, heterozygous female mice were found to have acquired the ability to distinguish lights of different wavelength that normal mice could not distinguish. These experiments show that the mammalian brain has an inherent plasticity that permits it to take advantage of new sensory inputs so that evolutionary variation in sensory receptor cells can be immediately useful for the species – without a need to evolve more complex neural circuits to extract or compare this new information. This type of plasticity is likely to be used in the evolution of other sensory systems.

3. Recommended Reading

1. Jacobs, G.H. and Nathans, J. (2009) The Evolution of Trichromatic Color Vision. Scientific American (April), pp. 32-39.
2. Nathans, J. (1999) The Evolution and Physiology of Human Color Vision: Insights from Molecular Genetic Studies of Visual Pigments. Neuron 24: 299-312.

4. Review Questions

1. What is the principal evolutionary and geographic division among primates?
2. In which of the two primate divisions do humans belong?
3. Name two potential ways in which trichromatic color vision might confer an advantage over dichromatic color vision.
4. Of the 364 amino acids in the M and L pigments, how many positions play an important role in distinguishing their absorbance spectra?
5. What is X-chromosome inactivation?
6. How was the M and L pigment gene locus control (LCR) region first identified?
7. In a three-way forced choice test of color vision, how one can one be sure that the subject (either human or nonhuman) is not using differences in light intensity to discriminate between the reference lights and the test light?
8. In a three-way forced choice test, how well would the subject (human or nonhuman) perform if he or she was simply guessing randomly rather than making a real discrimination?

5. Answers to Review Questions

1. Old World primates encompass the African and Asian species. New World primates encompass the Central and South American species.
2. Humans and our nearest primate relatives (the great apes) are Old World primates.
3. First, finding fruit among dappled foliage and recognizing its degree ripeness, an ability that would be expected to increase food consumption. Second, distinguishing differences in coloration among members of one's own species, an ability that might improve an assessment of potential mates or competitors.
4. Only three: positions 180, 277, and 285.
5. X-chromosome inactivation is an epigenetic process that occurs during embryonic development in female mammals and it silences most of the genes on one of the two

X-chromosomes. In each embryonic cell in which X-chromosome inactivation occurs, the initial choice of which of the two X-chromosomes to silence is random and independent, but once the decision occurs it is stably inherited in all of the progeny of that cell. As a result, female mammals are mosaics.

6. The LCR was identified as a DNA sequence adjacent to the array of M and L pigment genes that was deleted in individuals with blue cone monochromacy, a severe inherited color vision deficiency in which both M and L cones are missing.
7. The intensities of one or both lights must be randomly varied to insure that wavelength rather than intensity is being used to make the discrimination.
8. On average, one third of the responses would be correct.

6. Discussion Questions

1. What led to the geographic and biological isolation of New and Old World species, and when did this happen? What are some other examples of geographic isolation that has led to biological divergence?
2. Name some non-primate animals that have good color vision.
3. Compare the geometric representation of human color perception based on cone photoreceptor sensitivities vs. retinal ganglion cell activity.
4. Current evidence suggests that the locus control region (LCR) adjacent to the M and L pigment gene array pairs with a single M or L pigment gene promoter to direct gene transcription in M or L cones, respectively. If this pairing and the associated transcriptional choice constitutes the only difference in gene expression between M and L cones, and if it occurs by a random choice of LCR binding to either the M or L pigment gene promoter, then what would be the predicted effect on cone identity and number of losing all but one of the pigment gene?

7. Answers to Discussion Questions

1. The New World (America) and Old World (Africa) were part of one contiguous continent until about 100 million years ago when plate tectonic movements cause

the continents to start to drift apart. Complete biological separation was probably attained by about 50 million years ago. Other examples of divergent evolution following biogeographic isolation include the evolution of different species of fruit flies on the different Hawaiian Islands, the evolution of marsupials on the Australian continent, and the evolution of distinctive species of finches on the Galapagos Islands.

2. Insects that pollinate colorful flowers, such as honeybees, have excellent color vision that includes receptors with absorbance maxima in the ultraviolet. Most likely the flower pigments that produce the petal colors co-evolved with the insect visual pigments that detect their distinctive absorbance spectra. Recall that recognition of the flower by the insect is mutually advantageous: the insects obtain nectar and the flowers exchange pollen thereby fertilizing the next generation of seeds. [Interestingly, a major class of plant pigments consist of unbranched conjugated carbon chains with the same general architecture as 11-cis retinal, the visual pigment chromophore. Compared to 11-cis retinal, which has a conjugated region consisting of six double bonds separated by five single bonds, the plant pigments have substantially longer chains of alternating single and double bonds.] Other examples of animals with highly evolved color vision include goldfish and pigeons. In general, those species with complex body coloration, such as tropical fish and many birds, have multiple visual pigments that can decipher those colors. Also, body coloration is often sexually dimorphic.

3. As first described by Maxwell in the mid-nineteenth century, we can conceptualize human color perception using a three dimensional Cartesian coordinate system with the X, Y, and Z axes representing the degree of excitation of the L, M, and S cones. Since every light stimulus is composed of some combination of wavelengths with a particular set of intensities at each wavelength, every light can be represented by a vector in the three-dimensional space. The orientation of the vector indicates the relative extents of excitation of the three cone types, and the amplitude of the vector is proportional to its intensity.

   At the level of the retinal ganglion cells, the cone signals have been processed to yield an M vs. L signal, an S vs (M+L) signal, and a lightness vs. darkness signal. Therefore, again we have three-dimensional perceptual space, but this time the axes represent (roughly speaking) redness vs. greenness, yellowness vs. blueness, and a grey scale going from black to white. This second perceptual space, which is equivalent in information content to the first one, corresponds to the representation of color and lightness-darkness information that is conveyed to the brain.

4. X-chromosomes in which the visual pigment gene array consists of only a single L pigment gene or a single L/M hybrid pigment gene as a result of homologous unequal recombination are quite common – about 1% of human X-chromosomes have this arrangement (see lecture 2). We would predict that the LCR would then simply pair with the single remaining visual pigment gene promoter, thereby producing a single class of cones from all of the precursor cells that would, in a normal trichromat, have been divided between M and L cone types. Thus, we would predict that in these individuals the spectral sensitivity of the retina to those wavelengths preferentially absorbed by the single X-chromosome encoded pigment would be higher than the corresponding sensitivity for a normal trichromat. We would also predict that the total number of cones subserving longer wavelength vision (i.e. M and L cones) would be unchanged from the number found in normal trichromats. Both of these predictions are borne out by the data.

   By contrast, if M vs. L cone types and M vs. L pigment gene expression were determined by cell-type specific transcription factors, then a visual pigment gene array with only one gene would support visual pigment gene expression in just one of the two types of cones, leaving the other cone type empty of visual pigment. This has not been observed.

8. Explain or Teach These Concepts to a Friend

1. Explain the way in which plate tectonics have shaped the continents, and the effect of these movements on the evolutionary history of primates.
2. Explain how the different chromatic opponent circuits in the retina compare cone inputs.
3. Explain how a three-way forced choice test works.

9. Research the Literature on Your Own

1. How good is the color vision of a bee? What are the spectral sensitivities of the visual pigments found in bees and how do these properties facilitate the tasks that the bees accomplish?

2. Draw a time-line and evolutionary tree showing the speciation of the major types of vertebrates, including mammals and primates.
3. Discuss the implications of X-chromosome inactivation for human genetic disease.
4. Random choices between different states of gene expression are a recurrent theme in biology. A classic example is the lysis vs. lysogeny decision in bacteriophage lambda, a bacterial virus. Contrast that process with the process of M vs. L visual pigment gene selection.