

Wavelength Discrimination Deteriorates with Illumination in Blue Cone Monochromats

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Two types of incomplete congenital achromats were studied: one type (blue cone monochromats) has a conspicuous short wavelength cone mechanism, and the other type (deutan incomplete achromats) has a conspicuous long wavelength cone mechanism. The photoreceptor mechanisms were inferred from color matches and from test action spectra measured on rod-saturating backgrounds of different wavelengths. Interestingly, the illumination-dependency of color discrimination (for 5° bipartite fields that were centrally fixated) differed between the two patient types, even though rhodopsin photoreceptors were common to both. As illumination level increased, the ability to discriminate wavelength differences deteriorated for the blue cone monochromats, whereas, for the deutan achromats, wavelength discrimination remained relatively constant even near 100,000 scotopic trolands. The performance decrement in the blue cone monochromats was probably not associated with rod saturation, as the field action spectrum to cause a just-noticeable-difference (jnd) decrement in discrimination was poorly fitted by a rhodopsin action spectrum. In addition, the blue cone monochromats had rhodopsin photoreceptors that did not saturate in bright illuminations. The authors hypothesize that the deterioration of wavelength discrimination at high illuminations is not an abnormality of blue cone monochromacy. Rather, it may be a property of the normal color mechanism through which signals from the short wavelength cones pass. *Invest Ophthalmol Vis Sci* 26:1543-1549, 1985

Congenital achromatopsia usually refers to a syndrome of total colorblindness and poor visual acuity in patients with an unremarkable fundus.¹⁻³ Because these patients lack functioning cone photoreceptors and see mostly with rods, their visual resolution (eg, as measured by visual acuity, visual field isopters, or Weber fraction) greatly depends on illumination level. Like normal observers, their visual performance initially improves with increases in scotopic illumination. But the similarity ends there. With further increases in the ambient illumination, the patients' visual resolution deteriorates. At daylight illumination, patients typically report that their visual field is "washed-out" or "homogenized."^{1,4} Optimal visual resolution for these patients occurs at an intermediate retinal illumination, eg about 50 scotopic trolands.⁵

Related to congenital achromatopsia is a reportedly rarer disorder called "incomplete achromatopsia."^{6,7} Incomplete achromatopsia resembles complete achro-

matopsia with a main difference. The incomplete achromat has rudimentary color vision. There appears to be two main classes of these patients, those with residual color discrimination in the short-middle and those with discrimination in the longer wavelength portion of the spectrum. Patients with short-middle wavelength discrimination have been referred to as blue cone monochromats or pi 1 incomplete achromats.^{3,6,8-11} The disorder has an X-linked recessive transmission, so most patients are male. These patients are dichromats and their color matches are mediated by a short wavelength sensitive (sws) cone (with 445 nm peak sensitivity) and a rhodopsin photoreceptor (510 nm peak).⁸ Interestingly, the photopic rhodopsin photoreceptors of several patients were found to be cones^{8,10}; however, those of a patient studied by Daw and Enoch¹¹ were reportedly rods.

The second type of incomplete achromats are patients with residual long wavelength discrimination. Although there is large variability in the sensitivity of the residual cone mechanisms,^{7,12} classification of these patients into two subtypes appears meaningful. Some patients have a residual protan¹³ mechanism as evidenced by large field flicker photometry. According to Smith et al,¹³ these patients are dichromats and they discriminate colors on the basis of rhodopsin and middle wavelength sensitive (mws) cone photoreceptors. Other patients have a pi 5 or deutan mechanism.^{7,12,14-16} According to Pokorny et al,⁷ these pa-

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tients may be trichromats and their color vision is either mediated by rhodopsin, mws cone, and long wavelength sensitive (lws) cone photoreceptors; or is mediated by rhodopsin, sws cone, and lws cone photoreceptors.

Although incomplete (as well as complete) achromats are hemeralopic, there has been little systematic effort devoted to studying their color discrimination at photopic illumination levels. In the present paper, we report on our efforts to elucidate an aspect of the rudimentary color vision in two types of incomplete achromats. We examined the dependency of wavelength discrimination on illumination in incomplete achromats with a conspicuous sws cone or in those with a conspicuous lws cone mechanism. The results of our study suggest an interesting property about the mechanism underlying color discrimination in these patients.

Materials and Methods

Patient Selection

Patients for this experiment were found over a 4-yr period during which congenital achromats were being recruited for another study.¹⁷ Our methods for recruitment included newspaper advertisements, physician referrals, or examinations of family members with similar disorders. From a group of prospective subjects, we found four patients who unequivocally had residual color discrimination (see Results). The four patients had a history of a congenital, stationary disorder that is associated with poor color vision, poor visual acuity, some nystagmus and photophobia. Ophthalmoscopic examination of their fundus revealed little, if any, pathology.

Patient 1 was a 26-yr-old male who belongs to a multiplex family. His brother 2 and sister 3 have complete achromatopsia. Patient 4 was a 16-yr-old female of a second multiplex family. Her sister, patient 5, a complete achromat, was referred to one of us by Morton F. Goldberg, M.D. Patient 6 was a 17-yr-old male with a x-linked recessive pedigree; his maternal uncle has the same disorder. Patient 7 was a 14-yr-old male with no family history for this disorder; he was referred to one of us by Gerald Fishman, M.D.

General Procedures

Informed consent was obtained after the procedures had been fully explained. Patients were always dark-adapted for a minimum of 20 min prior to the start of each experimental session. Then, they were light-adapted to the lowest illumination condition first. The patients were instructed to observe two main precautions. (1) They should not squint (ie decrease retinal illumination by partially closing their eyelids), partic-

ularly at high illumination levels. (2) They should not utilize information from after-images produced by blinking. Studies have shown that such after-images provide information that differs from that obtained from the real images.¹⁸ The experimenter monitored the patients by watching their eyelids in the darkened experiment room. When the patients blinked or partially closed their eyelids, the experimenter could often see the reflection on the patients' eyelids.

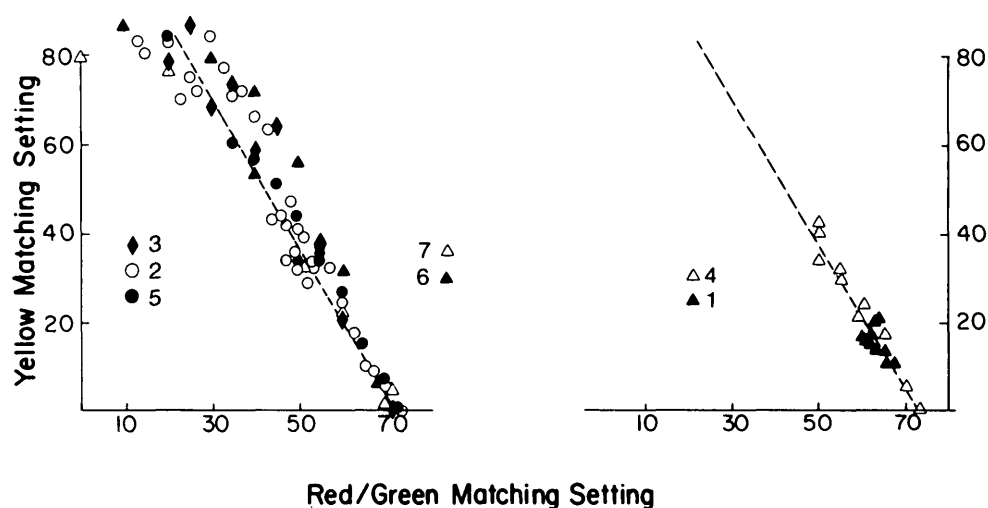
Increment Thresholds

Most of the increment thresholds were collected on a three-channel Maxwellian viewing apparatus described elsewhere.¹⁹ More recent measurements collected at Texas Tech Health Sciences Center were made on a 4-channel system modified to provide suitable test and background field diameters. A biteboard was used to maintain the patients' alignment with the instrument. Threshold measurements consisted of the patient adjusting the test flash to be "just visible," if the flash initially could not be seen or "just invisible," if the flash could be seen. Within a given session, the patient typically made six measurements in an up-and-down fashion.

Wavelength Discrimination

Wavelength discrimination was performed on a 4-channel Maxwellian-view optical system described elsewhere.²⁰ Refractive errors of the eye were corrected with trial lenses; chromatic aberrations were minimized with an achromatizing lens. A biteboard was used to maintain the patients' alignment with the instrument. The stimulus was a 5° bipartite field, one half serving as a standard and the second half, as a test field. The test wavelengths were varied on a grating monochromator which had about 8-nm dispersion (in order to obtain high intensities); the standard field was filtered with an interference filter of about 8-nm half-band pass width. The observer's task was to obtain a metameric match between the test and standard fields only by adjusting the intensity of the test field. If a match was possible, the experimenter increased the difference between the test and standard wavelength. Conversely, if a match was not possible, the difference was decreased. The experimenter varied the test wavelength in a staircase fashion until the range of just-noticeable-wavelengths could be bracketed. Estimates of the just-noticeable-wavelengths were usually obtained once on the long wavelength side and once on the short wavelength side of the standard. Within a given session, the wavelength of the standard was fixed. At the start of each session, the patient dark adapted fully and then light adapted to the stimulus display for about 2 min. During light adaptation, the observers practiced setting

Fig. 1. Color matches obtained on a Nagel Model 1 anomaloscope. *Right:* Color matches for patients 4 and 1 who have residual color discrimination in the red/green portion of the spectrum. *Left:* Color matches for patients 7 and 6 who have no discrimination in the red/green. For comparison, the color matches of three complete rod achromats (2, 3, 5) are also plotted. The line through the data points represents the locus of color matches for an observer whose color discrimination in the red/green portion of the spectrum is mediated by rhodopsin photoreceptors.



isomeric matches in order to establish a stable criterion for equality.

The field flux for optimal wavelength discrimination was estimated from the results of the effect of field illumination on wavelength discrimination. First, a best-fitted regression equation was found using a time-series analysis program; the data were well fitted by one of three general equations: $Y = A + BX + CX^2$, $Y = A + BX + CX^2 + DX^3$, or $Y = A + B/X + C/X^2$. Then, the field flux for optimal discrimination was derived by finding the solution at which the first derivative of the regression equation was equal to zero.

Results

Photoreceptor Mechanisms of Two Types of Incomplete Achromats

The color vision of patients 4 and 1 differs from that of 7 and 6. Patients 4 and 1 have residual long wavelength discrimination, as documented by their color matches (Fig. 1, right). Patients 4 and 1 could only accept matches over a limited range of red-green mixtures; outside this range, the patients discriminated the red-green mixture from the spectral yellow. Interestingly, their color matches lie on the same locus as that for patients with complete rod achromatopsia. Thus we infer that one of the photoreceptors underlying the color vision of 4 and 1 contains rhodopsin. The action spectra of the underlying mechanisms were further elucidated by increment threshold measurements in which we measured the patients' test sensitivities on rod-saturating backgrounds of various wavelengths (Fig. 2, top). At least two identifiable mechanisms were found to underly the fixation region of their eyes. One action spectrum resembles Stiles' pi 5 mechanism; the

second spectrum resembles that of Stiles' pi 0, ie a rhodopsin-like spectrum. Over all, patients 1 and 4 appear similar to incomplete achromats with long wavelength discrimination reported previously^{12,15,16}, and also with those classified as incomplete achromats with a deutan luminosity,¹⁴ although we do not know whether they fall within Pokorny's group III (with mws cones) or group IV (with sws cones) classifications.⁷

In contrast, patients 7 and 6 had little or no color discrimination over the long wavelength portion of the spectrum. Their color matches were similar to those of complete rod monochromats over the entire red-green mixture range (Fig. 1, left). Patients 7 and 6, however, denied total colorblindness and measurements of their test sensitivities on rod saturating background fields supported their claim (Fig. 2, bottom). In the region of the patients' fixation, the patients have two conspicuous mechanisms. One mechanism resembles that of Stiles' pi 3 spectrum. The second mechanism as revealed by increment thresholds (or color matches) is consistent with a rhodopsin action spectrum. For example, patient 7's test sensitivity (on a 4.46 log sc td background) has a pi 0-like spectrum for wavelengths greater than 476 nm. Over all, patients 7 and 6 appear similar to the blue cone monochromats previously reported.^{6,8-11}

Wavelength Discrimination as a Function of Illumination for the Blue Cone and for the Deutan Incomplete Achromats

Wavelength discrimination curves for the two types of incomplete achromats are illustrated in Figure 3. There is some intersubject difference, particularly between patients with the residual pi 5 mechanism. Pa-

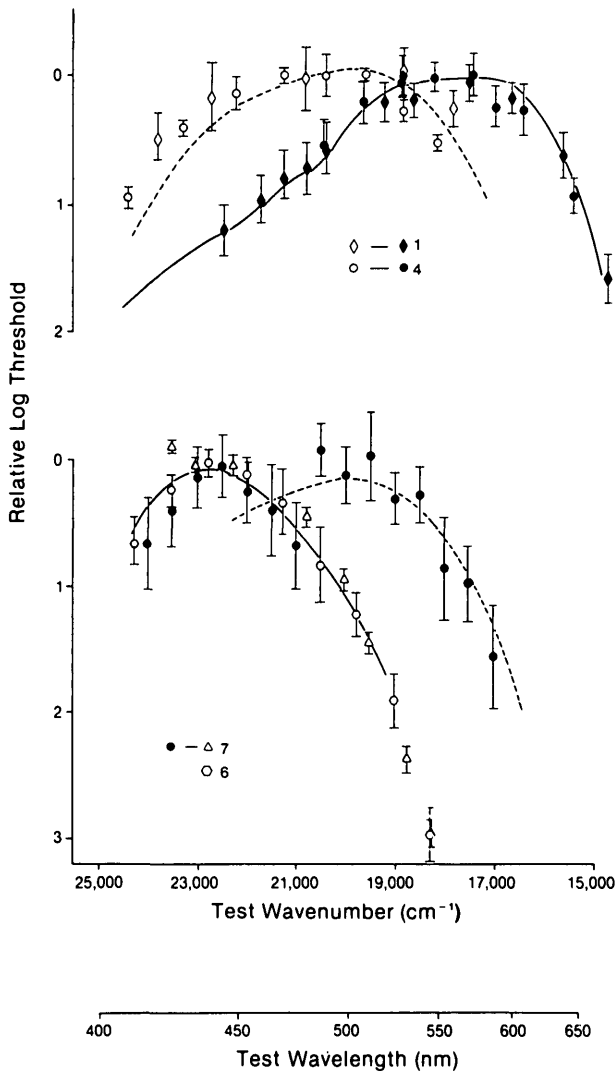


Fig. 2. Test field action spectrum measured on chromatic backgrounds for the two classes of incomplete achromats. *Top:* For patients 1 and 4, the spectral sensitivity measured with a 5°, 200 msec flash on a 8° steady background field (500 nm, 4.46 log scotopic trolands) resembled Stiles' pi 5 (solid curve). The spectral sensitivity measured on long wavelength backgrounds (for 1, 580 nm at 3.93 log scotopic trolands; for 4, 560 nm at 3.02 log scotopic trolands) resembled that of the CIE scotopic luminous efficiency curve when plotted on a quantum basis (dash curve). *Bottom:* For patients 7 and 6, the spectral sensitivity curve (open symbols) measured with a 1° test flash on a 8° background of 550 nm at 4.46 log scotopic trolands, resembled that of Stiles' pi 3 mechanism (solid curve). In addition, on a 430-nm background set at 4.15 log scotopic trolands, 7 spectral sensitivity for a 5° test flash (solid symbols) resembled that of Stiles' pi 3 mechanism for wavelengths less than 480 nm and that of the CIE scotopic luminous efficiency curve for wavelengths greater than 480 nm (dash curve).

tient 4 only begins to discriminate wavelength differences at illuminations that have nearly reached optimal levels for patient 1. From preliminary increment threshold experiments, we relate this difference to our finding that pi 5 sensitivity of patient 4 is poorer than

that of 1. Despite the individual difference, however, several generalities can be drawn from our results. At very low illumination levels, only rods function and none of the patients are capable of color discrimination. As illumination level is increased, color discrimination of the incomplete achromats generally improved, as indicated by reductions in the discriminable wavelength differences. As illuminations increased further, however, wavelength discrimination deteriorated for the blue cone monochromats (Fig. 3, top); whereas for the deutan incomplete achromats, relatively little or no change could be appreciated (Fig. 3, bottom).

Additional data describing the relationship between discrimination and illumination for one of the blue cone monochromats, 7, are illustrated in Figure 4. The results illustrate two general points. First, regardless of the wavelength of the standard field, discrimination always reaches an optimum at some intermediate illumination and then deteriorates at higher illuminations. Second, discrimination also varies as a function of the wavelength of the standard field. The smallest discriminable difference reaches a minimum for stan-

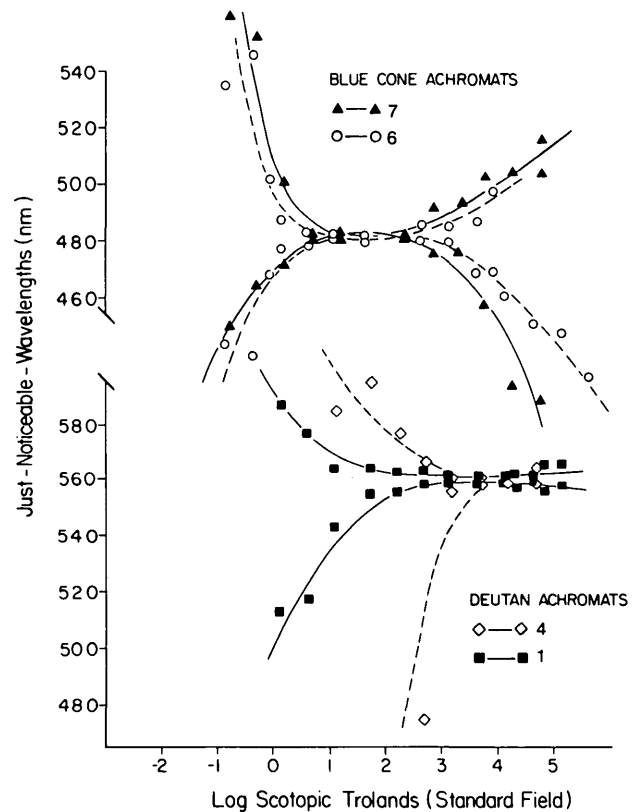
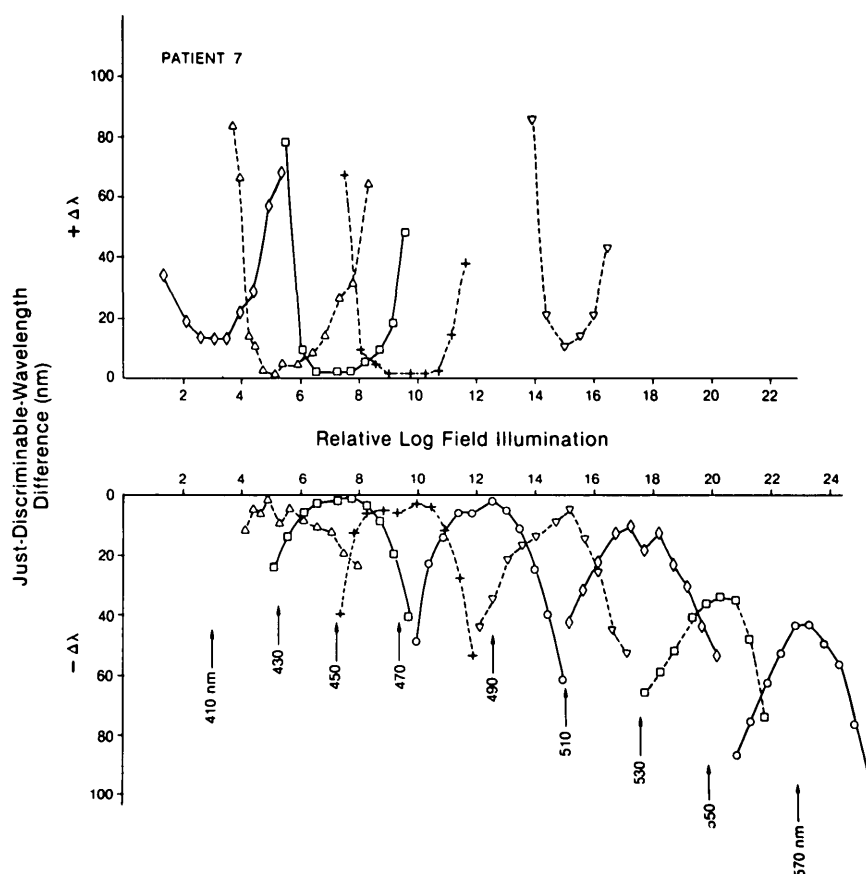


Fig. 3. Wavelength discrimination as a function of illumination for the two types of incomplete achromats. *Top:* Discrimination from a standard wavelength of 480 nm for blue cone monochromats. *Bottom:* Discrimination from a standard wavelength of 560 nm for deutan incomplete achromats. The curves were drawn to illustrate the general trend of the data points; they have no theoretical significance.

Fig. 4. Wavelength discrimination as a function of illumination for representative wavelengths (blue cone monochromat, patient 7). Discriminable differences from the long wavelength side of the standard are plotted as positive wavelength increments; discriminable differences from the short wavelength side are plotted as negative increments. Zero represents no difference from the standard wavelength. The numbers 410, 430, 450, etc. (below) are the standard wavelength for each curve. The curves were displaced along the relative log illumination abscissa for the sake of clarity. The actual illumination values, however, can be derived from the data in Figure 5.



standard wavelengths between 430 and 490 nm and then appears to increase with shorter or with longer standard wavelengths.

The field illumination at which discrimination was optimum also varies with the wavelength of the standard field (Fig. 5). The plot of the field flux for the different wavelengths illustrates an action spectrum with peak sensitivity between 430 and 480 nm. The results, however, are poorly described by a rhodopsin (or for that matter, a sws cone) action spectrum. This finding suggests that the illumination for optimal discrimination is not determined by a single photoreceptor mechanism.

Discussion

Although the residual color vision of either type of incomplete achromats is mediated in part by rhodopsin photoreceptors, our results (Fig. 3) show that color discrimination of the blue cone monochromats behaves differently from that of the deutan achromats. The blue cone monochromats exhibit a marked decrease whereas the deutan achromats exhibit less, if any change, in wavelength discrimination at high illuminations. The current literature contains little about the illumination-

dependency of color discrimination in incomplete achromats. However, our results on the blue cone monochromats appear consistent with the subjective descriptions of similar patients reported by Blackwell and Blackwell.⁶ At mesopic illuminations, Blackwell's patients described short wavelength fields as appearing "blue," long wavelength fields as "yellow," and fields of wavelengths between about 450 and 510 nm as "neutral." But at photopic illuminations, the same patients lost the color-naming ability and were appraised to function as "monochromats." Likewise, our results on the deutan achromats appear consistent with the only study we know which deals with the illumination-dependency of residual long wavelength discrimination. Using Farnsworth 100 Hue Test, a previous study¹⁶ showed that color discrimination in the red-green spectrum of an incomplete achromat improved at intermediate and remained nearly the same at high illuminations. This finding parallels our wavelength discrimination results of deutan achromats. (Interestingly, the same patient also had residual blue-yellow discrimination. Her blue-yellow discrimination, like that of our blue cone monochromats, improved at the intermediate and then worsened at the high illumination level.)

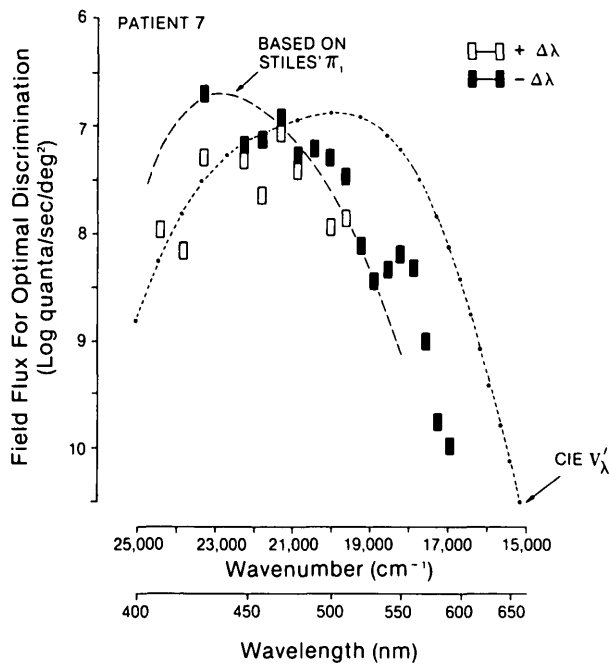


Fig. 5. Quantum field flux for optimal discrimination as a function of standard wavelength (blue cone monochromat, patient 7). Derivation of the data is described in the Methods section. Discrimination from the long and short wavelength sides of the standard are represented respectively by the open and solid symbols. The dash curves represent estimates of the action spectrum for the sws cones and for rhodopsin photoreceptors.

The mechanism underlying the loss of wavelength discrimination in blue cone monochromats is poorly understood. Blackwell et al⁶ initially hypothesized that it could be attributed to a color mechanism that was mediated by sws cones and by rods. Accordingly, color discrimination at mesopic illumination was possible because both sws cones and rods functioned. But at photopic illumination, the rods would no longer contribute to visual discrimination; so the patient was left with only sws cone function. The deterioration of color discrimination, therefore, was associated solely with a loss of rod function at high illuminations. However, the assumption that all rhodopsin photoreceptors were rods (and therefore, all rhodopsin signals would saturate at photopic illuminations) was not supported by subsequent investigations. First, the rhodopsin photoreceptor mechanism in (at least) some patients did not saturate at illumination levels that the normal rod mechanism would.^{8,9} In our study (Fig. 2), the rhodopsin mechanism measured on a 4.46 log sc td background for blue cone monochromat 7 has a Weber fraction of about 0.30, which is about 100 times smaller than the value expected for the Stiles' pi 0 thresholds on a saturating background.²¹ (The rhodopsin mechanism for the deutan incomplete achromats also did not appear to saturate. The Weber fractions for the rhodopsin mechanism were 0.20 and 0.17 for patients

1 and 4, respectively). Second, other investigators showed that the rhodopsin photoreceptor mechanism had the directional sensitivity of cones.⁸ Third, the rhodopsin mechanism dark adapted with a time constant that was more consistent with the recovery of a cone mechanism.^{8,10}

Additionally, our results provide little evidence that the rhodopsin photoreceptor (either rod or cone) is responsible for the loss of discrimination at high illumination levels. If the rhodopsin photoreceptor were solely responsible (as Blackwell's hypothesis suggests), the action spectrum to cause a just-noticeable-difference deterioration in discrimination should correspond with a rhodopsin action spectrum. However, because the smallest discriminable wavelength difference increases with standard wavelengths greater than 500 nm (Fig. 4), the action spectrum to cause a 1 jnd deterioration in discrimination must be either similar to or more narrow than the optimal field flux spectrum shown in Figure 5. Therefore, we conclude that the rhodopsin mechanism is not solely responsible for the loss of discrimination with illumination.

In view of the more recent findings, we propose an alternative to Blackwell's hypothesis.⁶ We hypothesize that the loss of discrimination with illumination in the blue cone monochromat may relate to a property of the normal color mechanisms in man. Under certain conditions, color discrimination for normal observers also deteriorates with illumination. For example, the Weber fraction of Stiles' pi 1 mechanism was found to increase at high background illuminations, as if the sws cone signals saturated.²²⁻²⁵ Similarly, the foveal discrimination of wavelengths near 460 nm reaches an optimum at intermediate illuminations and then worsens with higher illuminations (McCree,²⁶ Fig. 3). The discrimination, however, does not deteriorate with illumination over portions of the spectrum that is mediated by the midspectral cone mechanisms. The deterioration of discrimination in both normal observers and in blue cone achromats may involve mechanisms associated with the sws cones.

In conclusion, the results of the present study show that wavelength discrimination in the blue cone monochromats deteriorates at high illuminations. The loss of discrimination, however, does not appear to be an abnormality that is general to all types of incomplete achromats. It also does not seem to be associated with the saturation of rod signals. We hypothesize that the loss of discrimination with illumination in the blue cone monochromats may be a property of the normal color mechanism through which signals from the sws cones pass.

Key words: wavelength discrimination, rod mechanism, short wavelength cone mechanism, incomplete achromats, blue cone monochromats

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