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X-Linked Incomplete Achromatopsia with more than One Class of Functional Cones

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Five affected males in the fifth generation of a large pedigree of X-chromosomal incomplete achromatopsia were tested. All had SWS cone function. A 19-year-old affected man was a classical blue cone monochromat on color matching and spectral sensitivity. A 16-year-old boy showed evidence of a long wavelength sensitive cone active in 8° color matches. With a blue-green background, his cone spectral sensitivity function peaked near 550–560 nm. Three younger boys, aged 7–10 yrs were evaluated only with color matching. All showed evidence of long wavelength cone function with an 8° field and one showed long wavelength cones in 2° matches. An independent observation concerning the family was the finding that deuteranomaly was introduced in the third generation. The fourth generation women, all obligate carriers of X-linked achromatopsia, had a 0.5 chance to carry deuteranomaly. Neither carrier state per se is usually associated with expression of deuteranomaly. Three of the five tested expressed deuteranomaly. This finding of deuteranomaly in the carrier females might be a consequence of a double carrier state indicating association between the genes for deuteranomaly and X-linked achromatopsia. Invest Ophthalmol Vis Sci 24:451–457, 1983

An X chromosome-linked form of incomplete achromatopsia was first described by Spivey, Pearlman, and Burian[1] and Spivey.[2] Many of the findings are similar to those described for autosomal recessive complete or incomplete achromatopsia. The visual acuity is reduced, in the range 0.3 to 0.1; there is pendular nystagmus, light aversion, and the minimal fundal abnormalities reported in complete achromatopsia. As in other X chromosomal disorders there may be associated myopia.[3]

X-linked incomplete achromats are reported to show residual cone function[4,5] characterized by short wavelength sensitive (SWS or "blue") cone function in the absence of other cone function. Alpern, Lee, and Spivey[4] used the term π_1 cone monochromacy, and this entity was identified with the "blue" monocone monochromacy, described earlier by Blackwell and Blackwell.[6,7] Visual function has been studied actively in X-linked incomplete achromatopsia[3-10] because the observers offer an opportunity to study the SWS cone response in isolation. The various studies all converged in their agreement that the retina of the X-linked incomplete achromat contains but two types of active photoreceptor: SWS cones and rods.

We have had the opportunity to study color vision in a pedigree with X-linked incomplete achromatopsia in which there were five affected members. We employed color matching paradigms that have previously proved sensitive indicators of receptor types in autosomal recessive incomplete achromatopsia,[11-14] increment threshold procedures,[8,15] and conventional color vision tests.[15] One patient was a classical blue cone monochromat, but the others all showed evidence of more than one class of cone function.

Materials and Methods

Case Reports

The pedigree (Fig. 1) shows incomplete achromatopsia in males, transmitted by unaffected women, which could be traced for five generations. Five generation V males were affected with incomplete achromatopsia.

Case V-4: A 19-year-old man had best-corrected visual acuity of the right eye: 0.33 (20/60); the left eye: 0.167 (20/120) with refractive error of −7 diopters (D) in both eyes. There was congenital nystagmus. On electroretinography the photopic response was strongly diminished; the scotopic response was reduced mildly.
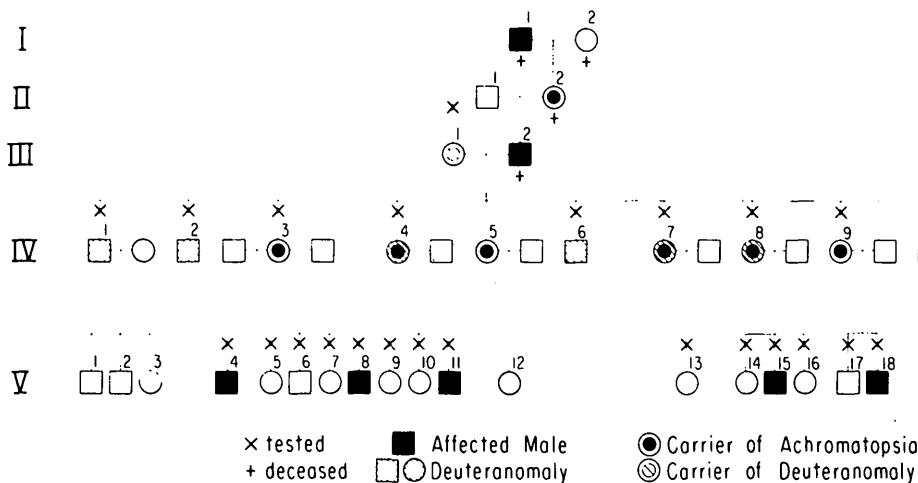
Fig. 1. Pedigree of family with X-linked achromatopsia. I-1 and III-2 were affected males according to family reports.

× tested   ■ Affected Male   ◉ Carrier of Achromatopsia
+ deceased   ☐◯ Deuteranomaly   ◎ Carrier of Deuteranomaly

Color vision screening showed a red-green color vision defect. There were no fundal abnormalities.

Case V-8: A 10-year-old boy had visual acuity of the right eye: 0.067 (20/300); the left eye: 0.10 (20/200), without refractive error. There was congenital nystagmus. On electroretinography the photopic response was extinguished; the scotopic response was normal. Color vision screening revealed a red-green defect. Ophthalmoscopic examination revealed granularity of the fundus and pale optic discs.

Case V-11: A 16-year-old boy had best-corrected visual acuity of the right eye: (20/200); the left eye: 0.1 (20/200), with refractive error of −0.5 D in both eyes. There was no nystagmus. On electroretinography the photopic response was diminished; the scotopic response was normal. Color vision screening showed a red-green color vision defect. The fundus appearance showed unusual macular pigmentation with two concentric streaks near the upper temporal side of the disc.

Case V-15: A 10-year-old boy had reduced visual acuity and congenital nystagmus from birth. On electroretinography, the photopic response was strongly diminished; the scotopic response was normal.

Case V-18: A 7-year-old boy had best-corrected visual acuity of the right eye: 0.133 (20/150); the left eye: 0.2 (20/100). The refractive error was right eye: −2S, −2C × 20°S; the left eye −2.55S, −1.5 C × 160°. There was right convergent strabismus with variable nystagmus. Electroretinography revealed reduced photopic response; the scotopic response was within normal limits. Color vision screening revealed a red-green color vision defect. The fundus appeared blond with congenital conus but was otherwise normal.

Procedure: Color Matching

We studied color matching for 2° and 8° fields using a modified Moreland anomaloscope.[16] To determine Rayleigh equations we used primaries of 545 nm (green) and 670 nm (red) matched to a test field of 589 nm (yellow). Additional test wavelengths of 570 nm and 610 nm were also available for some subjects. The field luminance at the normal match was 5 cd/m². The Rayleigh equation was assessed in the affected males and in as many family members as we could reach. One family member was tested with a Nagel anomaloscope. To determine the Moreland equation we used primaries of 500 nm (green) and 430 nm (blue) matched to a test field of 480 nm using a fixed amount of desaturant of 580 nm. This equation is a sensitive indicator of SWS cone function.[17] The field luminance at the normal match was about 5 cd/m². The Moreland equation was assessed in the five affected males. We used a procedure similar to that described by Linksz.[18] For each primary mixture ratio the patient adjusted the flux in the test field and then reported if a color match could be obtained. Matches were reported as the proportion of 670 nm (Rayleigh equation) or 430 nm (Moreland equation) in the match.

Increment Threshold

We studied increment threshold spectral sensitivity using a portable instrument described previously.[19,20] A 1° test field of variable wavelength was pulsed at 0.5 Hz on a 10° chromatic background field. Thresholds were obtained following 2 min of preadaptation by a tracking procedure in which the radiance of the test field was increased in small steps by a motor driven neutral wedge. By pressing a button the observer reversed the wedge direction and decreased the radiance of the test. The patient maintained the test field at visibility by controlling the direction of the wedge. The wedge position was plotted on chart paper. The test wavelength was changed after three to five reversals. By use of a calibration table, log relative spectral sensitivity was plotted for a series of test wavelengths. For our studies we used a tungsten white adapting field of 1000 tds to reveal SWS cone function, and a blue-green adapting field of 6300 tds to suppress SWS cones and rods and thus to reveal other possible cone function.[20] We obtained spectral sensitivities on affected patients V-4 and V-11. The three

younger patients did not give reliable data on this test.

Chromatic Discrimination

We obtained an index of chromatic discrimination in nonaffected family members using clinical color vision tests, including the Ishihara plates and the Farnsworth-Munsell 100 hue test. These tests were given binocularly under light from a Philips "color 57" fluorescent source. One patient (IV-3) was tested in natural daylight using screening plate tests.

Results

Color Matching in Affected Males

With a 2° field, four affected males had a full range Rayleigh match. The brightness matches were typical of matches made by complete achromats. The color matches indicated that with preferred fixation and a 2° field of 5 cd/m² rods were the only functional photoreceptors for wavelengths above 540 nm. The fifth patient (V-18) did not have a full range match. His matches extended from the normal match at 0.5 to the red primary; flux settings were those of rods. His data indicated the presence of a cone photoreceptor active at long wavelengths with a 2° field. With an 8° field (Fig. 2) only one of the affected males (V-4) made a full range scotopic match. The matching range for V-8 extended from 0.7 to the red primary. Narrow matches at 0.86, 0.83, and 0.88 were obtained from V-11, V-15, and V-18. The brightness matches for these narrow color matches were similar to those for the 8° field, indicating that rods were participating. In addition, a cone photoreceptor active at long wavelengths participated in the matching. The matching positions of three narrow matches made by

Fig. 3. Moreland equation for affected males. The flux in the 480 nm and 580 nm test is plotted as a function of the proportion of 430 nm primary in the mixture field. The dashed line shows matches of complete achromats; the dot-dashed line shows matches of tritanopes. Hatched rectangle shows 8° match positions of normal trichromats. Color matches are for an 8° field, except for V-18 who was tested with a 2° field. Symbols: $\triangle$ = V-4; ∇ = V-8; $\square$ = V-11; $\diamond$ = V-15; $\bigcirc$ = V-18.

V-11, V-15, and V-18 were consistent with a cone visual photopigment with λ max near 550–560 nm.[21]

Using a 2° field, none of the affected males made full range Moreland equations. The matching ranges were, however, wide except for patient V-18 who made a narrow normal match. The wide matching ranges of the other patients included the normal matching position. With an 8° field, the matching ranges of patients V-4, V-8, V-11, and V-15 narrowed, and these 8° color matches are shown in Figure 3 together with the narrow 2° match of patient V-18. The Moreland equation matches indicated SWS cone activity together with rod or other cone photoreceptor activity.

Increment Threshold Spectral Sensitivity in Affected Males

Increment threshold spectral sensitivity was measured on a white background (Fig. 4). Patient V-4 (right eye) showed maximal sensitivity at short wavelengths and a steep decline in sensitivity at longer wavelengths (Fig. 4). No thresholds could be measured above 530 nm. His data were characteristic of SWS cones; a SWS cone sensitivity function[22] (solid line) was fit by eye to the data. Patient V-11 also showed peak sensitivity at short wavelengths. His data showed a more gradual decline in sensitivity, and thresholds could be measured even at 611 nm (Fig. 4). The data of this patient were consistent with SWS cone activity below 500 nm and rod function above
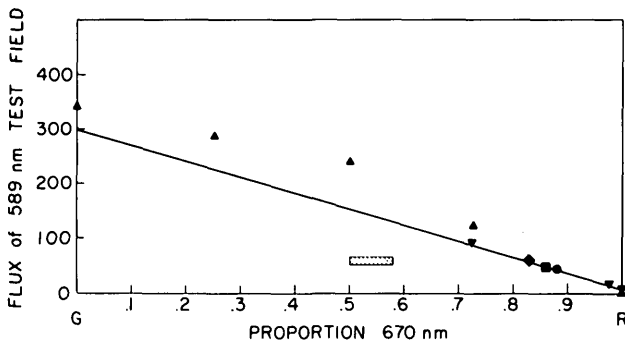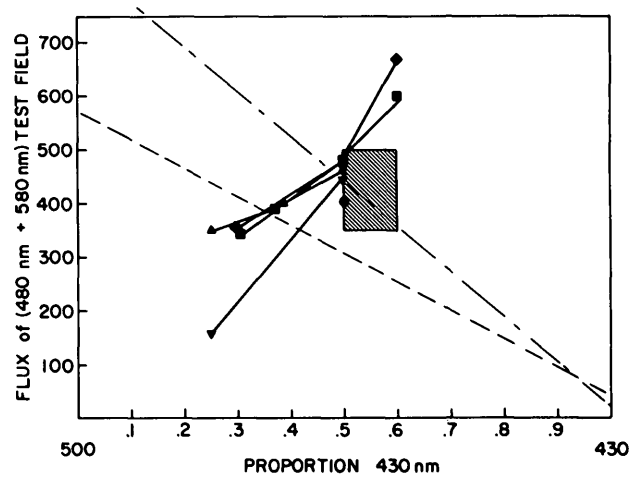

Fig. 2. 8° Rayleigh matches for affected males. The flux in the 589 nm test field is plotted as a function of the proportion of 670 nm primary in the mixture field. The solid line shows average matching position of complete achromats. Hatched rectangle shows 8° match positions of normal trichromats. Symbols: $\triangle$ = V-4; ∇ = V-8; $\square$ = V-11; $\diamond$ = V-15; $\bigcirc$ = V-18.
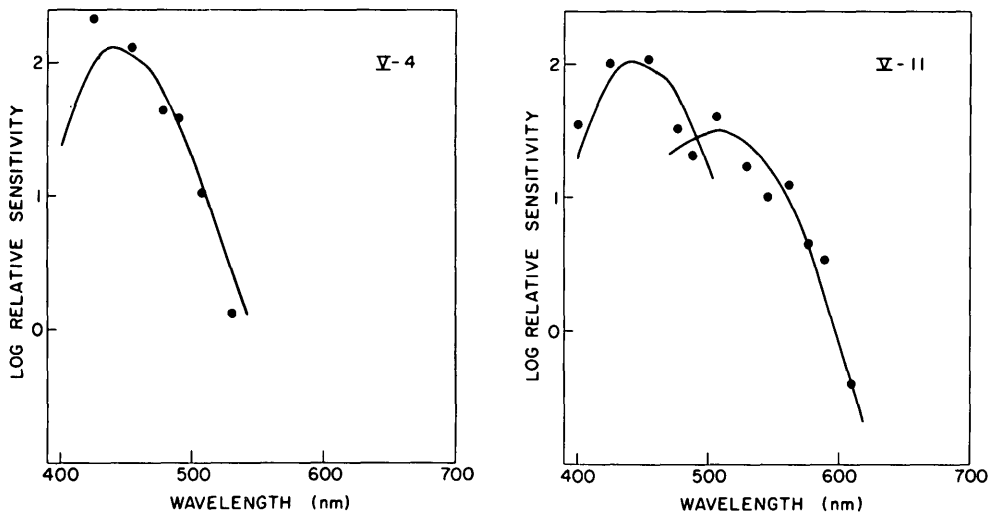
Fig. 4. Spectral sensitivity functions measured using increment detection on a white background for patients V-4 and V-11. The solid lines are curves for SWS cone (from reference 22) and rod (from reference 23) sensitivity. These functions were fit to the data by eye using an arbitrary vertical adjustment.

500 nm. The solid lines represent SWS cone[22] and rod[23] sensitivity functions fit by eye to the data.

On a blue-green background patient V-4 was unable to detect the test field even at full radiance. Patient V-11 could detect the test field for the majority of wavelengths (Fig. 5). His data suggested LWS cone function, and an LWS cone sensitivity function[22] was fit by eye to the data points. We inferred function of a cone photoreceptor with a λ max in the 550–560 nm spectral region.
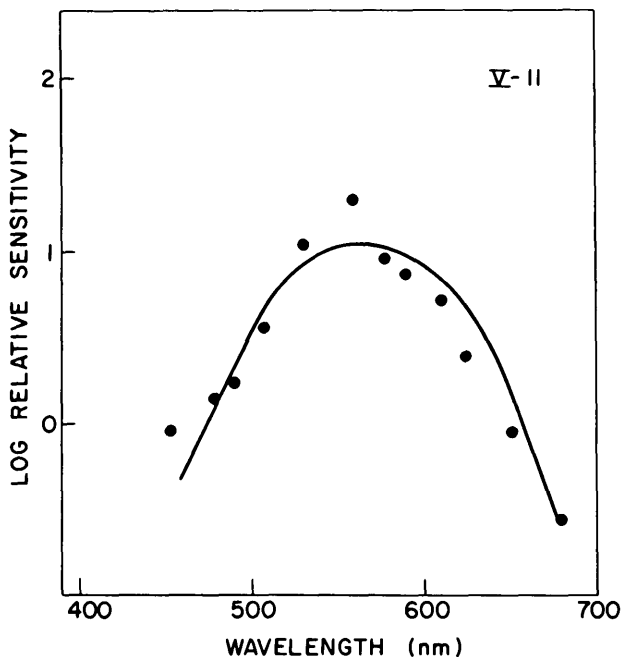

Fig. 5. Spectral sensitivity function measured using increment detection on a blue-green background for patient V-11 (average of two runs). The solid line is a curve for LWS cone sensitivity (from ref. 22). The curve was fit to the data points by eye using an arbitrary vertical adjustment.

Color Screening in Family Members

We performed color screening on 20 unaffected family members (Table 1). We found X-linked deuteranomaly in the five generation IV males, indicating that III-1 was a carrier for deuteranomaly. We were able to test five of the six generation IV females; three expressed deuteranomaly and two had normal color vision. The three deuteranomalous women showed similar behavior on the Ishihara and Farnsworth Munsell 100-hue tests as their deuteranomalous brothers (Table 1). Table 2 shows 2° Rayleigh matches at three test wavelengths for the seven siblings for whom data were obtained using the Moreland anomaloscope. The mid-matching positions are similar. The matching ranges are narrow with the exception of IV-2 (a male) who showed wide matching ranges. We tested nine unaffected members in the fifth generation; eight, including two male children with affected brothers, had normal color vision. One woman (V-14) made excessive errors on the Ishihara plate test, indicating minor color abnormality, perhaps associated with the carrier state.

Discussion

We have documented long wavelength cone function in X-linked incomplete achromatopsia. There was variability in the strength of the long-wave cone response. One patient showed only SWS cones, he appeared as a "classical" blue cone monochromat; but the remaining patients did not. A large field was necessary to reveal long-wave cone function in three patients, but long wavelength cones were active in a 2° field for one patient. Previous studies have not used field sizes greater than 4°. We suspect that other X-linked incomplete achromats may show long wavelength cones with sufficiently large fields.

Table 1. Color vision in unaffected family members of pedigree with X-linked achromatopsia

Patient	Age	Sex	FM-100	Ishihara	Rayleigh match	Conclusion
III-1	67	F	136 Blue-yellow	3 errors	PseudoPA†	Type III acquired defect
IV-1	42	M	150 No axis	22 errors	DA‡	Deuteranomaly
IV-2	40	M	53 Deutan	22 errors	DA	Deuteranomaly
IV-3		F*	—	2 errors	—	Normal color vision
IV-4	37	F*	150 No Axis	—	DA	Deuteranomaly
IV-6	34	M	74 No Axis	24 errors	DA	Deuteranomaly
IV-7	32	F*	328 No Axis	23 errors	DA	Deuteranomaly
IV-8	31	F*	—	21 errors	DA	Deuteranomaly
IV-9	27	F*	36 No Axis	3 errors	N	Normal color vision
IV-10	25	M	122 No Axis	14 errors	DA	Deuteranomaly
IV-11	23	M	—	17 errors	DA	Deuteranomaly
V-5	15	F	—	No errors	N	Normal color vision
V-6	14	M	—	No errors	N	Normal color vision
V-7	12	F	—	No errors	N	Normal color vision
V-9	19	F	77 No Axis	—	N	Normal color vision
V-10	18	F	40 No Axis	—	N	Normal color vision
V-13	8	F	—	No errors	N	Normal color vision
V-14	11	F	—	8 errors	N	Minor color abnormality
V-16	6	F	—	No errors	N	Normal color vision
V-17	10	M	—	No errors	N	Normal color vision

* Carriers of X-linked achromatopsia.
N = Normal.
DA = Deuteranomaly.

† Pseudoprotanomaly for 2° field, normal match for 8° field.
‡ Tested with Nagel anomaloscope.

This pedigree proved unusual since a gene for deuteranomaly was introduced by III-1. In the fourth generation, of 11 offspring, all five males and three of the five women we tested expressed deuteranomaly. One woman (IV-5) was unavailable for testing. Expression of deuteranomaly is extremely rare in women proven heterozygotic for the defect.[24] We and others[25,26] have noted red-green (deutan) discrimination defects in some carriers of X-linked achromatopsia; these defects occasionally take the form of deuteranomaly. In this pedigree, the generation V women except V-3, had a 0.5 chance to inherit a gene for X-linked achromatopsia. Of the seven whom we tested, none expressed deuteranomaly, one (V-14) made excessive errors on the Ishihara plate test.

Table 2. 2° Rayleigh Color Matches on the Moreland anomaloscope for seven siblings with deuteranomalous trichromacy. The proportion red at the extreme of the matching range is shown for three test wavelengths.

Patient	Sex	570	589	610
IV-2	M	0-.232	.181-.335	.380-.588
IV-4*	F	.140-.168	.270	.415-.439
IV-6	M	.126-.154	.207-.258	.427-.451
IV-7*	F	.126-.181	.258-.282	.403-.427
IV-8*	F	.154	.282	.427
IV-10	M	.126	.220-.245	.392-.439
IV-11	M	.154-.181	.270	.451
Normal		.270	.470	.720

* Carrier of X-linked achromatopsia.

The generation IV women are obligate carriers for X-linked achromatopsia and have a 0.50 chance to inherit the gene for deuteranomaly from III-1. Perhaps the expression of deuteranomaly in three generation IV women represents an interaction between genes causing deuteranomaly and X-linked achromatopsia. It is of note that IV-3 and IV-9 who did not express deuteranomaly each had a non-affected son with normal color vision.

One interpretation of X-linked achromatopsia[27-29] is that the defect results from a combination of protanopia (P) and deuteranopia (D) (compound hemizygote). Such a model or loss system[30,31] requires that for each class of dichromats, no viable photopigment is governed by the defective gene.¶ Females IV-4, IV-7, and IV-8 would be compound heterozygotes, carrying a gene for protanopia and (two) alleles for deuteranomaly and deuteranopia, thus predicting the expression of deuteranomaly. Females IV-3 and IV-9 carry the P and D genes on one X chromosome. According to this hypothesis, the daughters of IV-4, IV-7, and IV-8 must carry a gene for defective color vision.

A second hypothesis is that there is a separate gene for X-linked achromatopsia; according to this hy-

¶ The alternate model of dichromacy (replacement system) is that a normal or near normal photopigment (LWS for deuteranopes and MWS for protanopes) is substituted for the absent photopigment.[32-34] By this hypothesis, a male inheriting genes for dichromacy at both loci would have a photopigment complement allowing for normal or near normal color vision. A literature survey of presumed compound hemizygotes is presented elsewhere.[35]

pothesis, females IV-4, IV-7, and IV-8 would be heterozygous for both X-linked achromatopsia and deuteranomaly, IV-3 and IV-9 for the achromatopsia only. The finding of deuteranomaly in the three generation IV women could be ascribed to a steric effect on the normal gene at the deutan locus on the same chromosome, allowing expression of the normally recessive gene for deuteranomaly on the other X chromosome. Alternatively III-2 might have had genes for X-linked achromatopsia and for a deutan defect.

An analysis of the findings in generation IV leads to the following results (ignoring IV-5 and her only daughter V-12 who were not investigated): three women in generation IV, fully expressing deuteranomaly, have two sons with achromatopsia and five daughters without apparent abnormalities, while two women in generation IV who have normal color vision have three sons with achromatopsia, two with normal color vision and two daughters without abnormalities. The observed findings are consistent with either hypothesis for achromatopsia, a combination of P + D genes or a separate achromatopsia gene. The five daughters of the deuteranomalous generation IV women will produce sons with either deuteranomaly or with achromatopsia, the two daughters of generation IV women with normal color vision will produce sons with normal color vision or with achromatopsia (unless there is crossing over between genes at separate loci). We should emphasize, however, that the combination model offers the most economical explanation of our data. The separate gene model requires additional ad hoc assumptions to explain the finding of deuteranomaly in the three deuteranomalous generation IV women.

One finding in our pedigree might add further support to the hypothesis of a combination defect. It is now recognized that protanopes and deuteranopes have residual anomalous cone function, elicited by the use of large, high luminance photopic fields with rods suppressed,[36-39] or with a foveal flicker paradigm.[40] The anomalous cone spectra of deuteranopes[41] (M'WS) peaks near 555 nm. We represented the spectral sensitivity function obtained from V-11 with blue-green adaptation by the spectral sensitivity of LWS cones. However, the data may be fit with approximately the same precision with the deuteranomalous (M'WS) cone spectrum. It is possible that the residual cone function we measured reflects the same anomalous cone function as seen in large fields with deuteranopes under high photopic illumination.

Although the data are more economically explained by the combination defect hypothesis, our results offer no proof on the issue. Perhaps the descendants of this family will yield an answer in succeeding generations.

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