COMBINED FORMS OF CONGENITAL COLOUR DEFECTS*

A PEDIGREE WITH ATYPICAL TOTAL COLOUR BLINDNESS

BY

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There exist rare cases of monochromatism in which the characteristic symptoms accompanying typical total colour blindness are lacking. In such cases visual acuity is normal, nystagmus and photophobia are absent, the pupillary reactions are normal, the macula lutea exhibits no ophthalmoscopic anomalies, and the luminosity curve is not scotopic.

It has often been supposed that cone function is absent in typical, but present in atypical monochromatism, and atypical total colour blindness is hence called "cone monochromatism". This term is misleading, first because it has not been definitely proved that cone function is totally absent in typical total colour blindness,† and secondly because it is not yet clear whether the functional disturbance in atypical total colour blindness is located in the cones or in some other structure.

Atypical monochromatism is a very rare condition, of which only twenty or thirty cases have been reported in the literature. Although most of these case reports are incomplete, the group of persons with atypical total colour blindness does not seem to be homogeneous. Some cases are due to an acquired cerebral disturbance (König, 1891; Rohrschneider, 1928; Schober, 1948), others to a combination of congenital red-green blindness and acquired tritanopia (König, 1891, in a protanope; Köllner, 1909, in a deuteranope), and others are probably congenital. In the cases of Pitt (one, 1944), Sloan (one, 1946), and Weale (three, 1953), the maximum of the luminosity curve was shifted in the short wavelength direction, approximating to the protanopic luminosity curve.

Pitt (1944) considered his patient to be a congenital protanope and tritanope. Weale (1953), who discovered his patients with the aid of a published inquiry, believed that, in his patients, the disturbance occurred on a higher, post-receptoral level. This was because the luminosity curves did not coincide with those of protanopes, and also because the investigation of Fincham (1953) of the role of chromatic aberration in the accommodation

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† Walls and Heath (1954) have advanced the hypothesis that in typical total colour blindness the function of the "blue" receptors is still intact.
reflex suggests that, in Weale’s subjects, colour differentiation did exist in the retina.

Genetic data about “cone monochromatism” are extremely rare, but in a few cases colour vision defects appear to be familial. Raehlmann (1899) described a subject with a protanopic son; Vierling (1928) reported a patient with a completely colour-blind uncle; Weale’s subject J. G. was a compound heterozygote for protanomaly and deutanomaly.

Hylkema (1943), who examined a subject who is further analysed below, wrote that one son was protanopic and the other deutanopic. This female “cone monochromate” would thus have to be a compound heterozygote for protanopia and deutanopia, and consequently akin to Weale’s patient J. G.

Theoretical discussions of the last two of these cases by Walls (1955) are of considerable interest, although this author was unfortunately misled by errors on the part of Hylkema (1943), whose findings, which were recorded when both sons were still very young, have not been substantiated in the present investigation. More probably our case of atypical monochromatism (Case 2 (II, 10), below) was due to a combination of deuto-defects and trito-defects. Other atypical colour vision disturbances, which could be equally well interpreted as combinations of congenital colour vision defects, were discovered in the same family (Fig. 1).

Fig. 1.—Pedigree chart.
Method of Examination

In nearly all the subjects, the following tests of colour vision were done as routine:

(1) The plates of Ishihara (1943, Nos. 1–25) and the last four plates of Stillig (1936, Nos. 31–34) which are concerned with blue discrimination. Later, through the kindness of Dr. W. D. Wright, I also had at my disposal for the same purpose the pseudo-isochromatic plate of Farnsworth, with which Wright (1952) had been able to discover many cases of tritanopia in a mass examination.

(2) Further investigation of colour vision was conducted with the modified anomaloscope (Crone, 1955). Routine examination included:

(a) The Rayleigh equation.*
(b) An equation for tritanomaly, a mixture of green (0·513μ) and blue-violet (0·455μ) being matched with desaturated blue (0·480μ).†
(c) The threshold of wave-length discrimination in yellow (0·590μ) and in blue (0·480μ).
(d) Determination of relative brightness of green (0·530μ) and red (0·650μ) with the flicker photometer. This ratio is computed for a spectrum of equal energy, and is referred to below as the "luminosity quotient". The quotient is, for normal individuals, about 7·8, with a fairly broad spread. It is much higher for colour vision disturbances of the proto-group, lower for the deuterogroup. Heterozygous women with normal colour vision may often have an abnormal luminosity quotient (Schmidt, 1934; de Vries, 1948; Walls and Mathews, 1952). The figures for the luminosity quotient are given with some reserve since the clinical value of this quotient has not yet been tested on a large number of normal individuals and persons with typical colour vision disturbances.

(3) In special cases, routine examination was supplemented with the following determinations:

(a) Enumeration of various spectrum colours.
(b) Determination of the neutral point in the spectrum.‡
(c) Determination of the entire luminosity curve.
(d) Determination of the entire wavelength discrimination curve.
(e) Determination of the purity discrimination curve (for first step from white).

(4) The visual field for colour was also determined in two cases.

Pedigree

The pedigree of this family of Dutch nationality is shown in Fig. 1. The data on colour vision are set out in the Table (opposite). The following six cases require special consideration:

Case 1 (II, 5) Colour Amblyopia.—No peculiarities seen in routine ophthalmological examination. Both eyes had a visual acuity of 5/5 and were emmetropic. The visual fields for red and blue were limited concentrically to less than 30° of the centre (objects of 1 cm.² at 30 cm. distance). Dark adaptation was normal.

* In order to be independent from the source of light used, the ratio is given, for each subject, between his red-green mixture and the mean normal red-green mixture which could be matched with yellow. When a subject adjusts the left control of the anomaloscope at r units, while n is the normal adjustment of the Rayleigh equation, the quotient obtained is:

\[
\frac{n}{73-r} \times \frac{73-p}{p}
\]

According to Trendelenburg (1943), the quotient for deuteranomaly varies between 2 and 5, and for protanomaly between 0·13 and 0·6.

† The result is expressed as a quotient. A high value indicates tritanomaly or yellow coloration of the eye media.

‡ Colour temperature of the white matching field: 5,000° K.
<table>
<thead>
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<th>Generation</th>
<th>Sib No.</th>
<th>Age (yrs)</th>
<th>in Ishihara</th>
<th>in Stillinger</th>
<th>Farnsworth</th>
<th>Quotient of Rayleigh Equation</th>
<th>Quotient of Equation for Tritanomaly</th>
<th>Threshold of Wavelength Discrimination at 0.760 µ</th>
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The patient had not been aware of any disturbance in colour vision. The colours of a collection of coloured pieces of paper were identified correctly as long as the saturation level was not too low. Only a few of the Ishihara plates were read correctly. The errors typical of subjects with red-green disturbances were not usually made. In the plates for the discrimination of proto- and deutero-disturbances, sometimes the red figure was not seen, and at other times the purple figure was missed. The Rayleigh equation and the equation for tritanomaly were normal, though with an enlarged adjustment width. There was no pathological contrast; yellow, when placed next to red or green, was called whitish-yellow in both cases. There was no neutral zone in the spectrum. All the spectrum colours were well identified; the white comparison field was always seen as white. The wavelength discrimination was greatly reduced over the entire spectrum, with a minimum in green (Fig. 2a).

The luminosity curve was shifted somewhat to the right; the luminosity quotient was 4.4 (Fig. 2b).

The discrimination of differences in saturation, measured for the first step from white toward spectrum light, was lowered over the entire spectrum. The shape of the curve was similar to the normal (Fig. 2c).

Case 2 (II, 10) Atypical Monochromatism.—No peculiarities seen in routine ophthalmological examination. Both eyes had a visual acuity of 5/5 (+0.5D Cyl., axis vertical). No nystagmus or photophobia. Rapid pupillary reactions to
light. Normal fundi, normal visual fields for white. The dark-adaptation curve was normal and exhibited the characteristic kink after 6 min. (pre-adaptation with 2,000 lux.).

The patient had never been able to visualize colours. She believed that she could differentiate a few colours, but spoke only vaguely about "dark" and "light" colours.

Not one of the plates of Ishihara, Stilling, and Farnsworth was identified, rightly or wrongly. In the colour apparatus, each spectrum colour could be matched with white. The luminosity curve, determined with the flicker photometer, showed a shift of the maximum to the right (Fig. 3); luminosity quotient 4.

Case 3 (II, 11) Colour Amblyopia.—
No signs of abnormal ocular pigmentation in macula or lens. No peculiarities seen in routine ophthalmological examination. Visual acuity normal. Many mistakes were made in the Ishihara plates, but the characteristic confusions were not apparent. Normal Rayleigh equation; no pathological contrast-sensations; tendency to call green "yellow". Very broad adjustment width in the tritanomaly equation; blue occasionally called "green".

No neutral zone in the spectrum; white comparison field always seen as white.

Wavelength discrimination lowered over the entire spectrum (Fig. 4a). The luminosity curve of this subject (Fig. 4b) was the only one in this pedigree to
exhibit a shift to the left. Although the subject, aged 44, was a poor observer, the data recorded here were later substantiated in a re-examination.

**Case 4 (II, 18) Tritanomaly.**—No peculiarities seen in routine ophthalmological examination. Visual acuity normal. Farnsworth plate and Stilling’s plates 33 and 34 not read; no errors in the Ishihara plates. Rayleigh equation normal; very large adjustment width in tritanomaly equation. No neutral zone, but wavelength 0.575 was “nearly white”.

Enumeration of spectrum:

\[
\begin{align*}
0.440-0.540 & : \text{"blue"} \\
0.540-0.570 & : \text{"green"} \\
0.575 & : \text{"yellow, nearly white"} \\
0.610 & : \text{"light red"} \\
0.630 & : \text{"red"}
\end{align*}
\]

Wavelength discrimination greatly lowered in blue and green (Fig. 5); luminosity quotient normal.

**Case 5 (III, 4) Tritanomaly.**—No peculiarities seen in routine ophthalmological examination. Visual acuity normal. Only No. 32* among the Stilling plates was recognized; one mistake was made in the Ishihara plates. Tritanomaly equation 1.5; normal equation not accepted. Rayleigh equation normal. Wavelength discrimination curve exhibited moderate disturbance in blue and green (Fig. 6).

**Case 6 (III, 11) Incomplete Monochromatism.**—Alternating convergent strabismus; visual acuity for both eyes 5/5 (Sph. +4 D); normal fundi. Only the first Ishihara plate was read correctly, and the confusion numbers were mostly not read. Of the Stilling plates only No. 32* was recognized. In the colour apparatus the entire spectrum could be matched with white, but on larger sheets of

* This plate is of minor significance. I recently saw two patients with definite tritanopia who could read it without effort.
coloured paper the colour was usually given correctly. There was, therefore, total colour blindness in a small, central visual field. The data necessary to establish the nature of the colour vision for a larger visual field were lacking. The low luminosity quotient showed that the maximum of the luminosity curve was shifted slightly to the right.

Discussion

I shall endeavour to disentangle the genetic connexions among the various forms of colour blindness which appear in this family.

Case 1 (II, 5) is the best starting point; this woman had three children (III, 4, 5, 6):

1. A daughter with tritanomaly (Case 5: III, 4) and a low luminosity quotient, thus being perhaps heterozygous for a deutero-defect.
2. A son with deuteranopia (III, 5). There can be no uncertainty about this diagnosis in view of the presence of a neutral zone, a (not pictured) wavelength discrimination curve with one minimum, and a low luminosity quotient. This man has a broad adjustment width in the tritanomaly quotient, which is not usual in classical deuteranopes, and, for simple deuteranopia, a somewhat low wavelength discrimination in blue. We shall have to ascribe to him, besides a factor for deuteranopia, a factor for a trito-defect.
3. A daughter who is normal (III, 6) and, judging from the normal luminosity quotient, not heterozygous for a deutero-defect.

From this it follows that Case 1 (II, 5) is probably heterozygous for deuteranopia, and possesses at the same time a factor for trito-defect, which must be assumed for other members of her family. This tallies with her clinical picture in so far as the trito-factor is concerned; it is true that the tritanomaly quotient does not exhibit any important deviation from the normal, but there is severe reduction of blue-green colour discrimination. The deutero-factor appears to be less certain: in a heterozygote for deutero-defect, one would expect a normal red-green discrimination, but this subject has a major disturbance in red-green discrimination, as testified by the many errors made in the Ishihara plates. She cannot be a deuteranope, since there is no neutral point in the spectrum, and the hue discrimination curve has two minima instead of one. Deuteranomaly is just as unlikely, since the Rayleigh equation is normal. With reference to the red-green discrimination, this subject can be considered to be an incomplete deuteranope, a transitional form between normal trichromatism and deuteranopia. This is a direct transition, not by way of the stage of anomalous trichromatism. In accordance with the theory of Young-Helmholz: “There is no change in the spectrum sensitivity of the system of ‘green’ cones, but only a lowering of the sensitivity”.

The presence of a trito-factor seems to be responsible for the intermediate character of the otherwise recessive deuteranopia gene in this respect. This is only a hypothesis, but it receives support from Jaeger (1951), who described two sisters who were both non-allelic compound heterozygotes for
protanopia and deuteranomaly. One was phenotypically tritanomalous and slightly deuteranomalous, the other phenotypically protanopic. Here also genes for red-green disturbance had probably lost their recessive character through the presence of a trito-factor.

Subject II, 7, a sister of Case 1 (II, 5), is phenotypically deuteranopic. There can be no doubt about the diagnosis, since there is a neutral point in the spectrum and the hue discrimination curve has one minimum. She has two typically deuteranomalous sons (III, 9 and 10) and two normal daughters (III, 7 and 8).

The mother is therefore homozygous for deuto-defects; since her sister has a factor for deuteranopia and her sons are deuteranomalous, her genetic composition is, in all probability: deuteranopia-deuteranomaly. Clinically, she ought to have been deuteranomalous, following the rule that the gene for anomaly dominates over the gene for dichromatism. One can, as in Case 1, accept the influence of a trito-factor, which increased the expressivity of the deuteranopia gene, although for the rest the trito factor remains latent.

Case 4 (II, 18) has an isolated trito-disturbance. Since he is not a true tritanope (there is, among other factors, no neutral point) I have called the disturbance tritanomaly, although the normal tritanomaly equation was accepted, and the subject might as well be called an incomplete tritanope.

There are differences from, but also important similarities to, the classical cases of Engelking (1925). The fact that Case 4 has a deuteranomalous son (III, 19) is of no significance for this pedigree, since this deuteranomaly must be derived from his mother (II, 19).

Case 2 (II, 10), the most prominent subject of this pedigree, with atypical or cone monochromatism, derives her remarkable colour vision defect from a combination of deutero- and trito-defects.

Since she has one deuteranomalous son (III, 12), phenotypically she herself should be tritanomalous-deuteranomalous, which, following a simple addition sum, can never result in total colour blindness. For this, one would a priori expect a combination of tritanopia and deuteranopia. We have seen in Case 1 (II, 5), and perhaps also in Subject II, 7, that a trito-factor can increase the penetrance of a gene for deutero-disturbance; besides, the presence of tritanopia is not at all improbable in view of the varied manifestations of trito-defects in this family.

Case 6 (III, 11) has colour discrimination for larger colour surfaces, but his vision is monochromatic when examined in the colour apparatus.

We may regard this colour defect as incomplete atypical monochromatism. Genotypically we may assume a deutero-defect (deuteranopia or deuteranomaly) and a less marked trito-defect than that of his mother (Case 1: II, 10).
Case 3 (II, 11) resembles Case 1 (II, 5) in many respects. There is a severe disturbance in the red-green discrimination, although the defect cannot be classified as dichromatism or anomaly, and the blue-green discrimination is also severely disturbed, indicating the presence of a trito-factor.

This patient differs from all the other members of the family by a high luminosity quotient, such as exists in proto-disturbances. If a factor for proto-disturbance is present, it is not discernible from whence it is derived; if the factor had come from the father (I, 4), one would expect a high luminosity quotient in the subject’s sister (II, 12) also; if it had come from the mother (I, 5), one would expect a higher luminosity quotient in the mother. It seems, however, that a shift in the luminosity curve (“Schmidt’s sign”) is not invariably present in proto-heterozygotes*. In the present state of our knowledge it is perhaps advisable to regard Case 6 (II, 11) as a heterozygote for protanopia with an associated trito-defect.

Now that a schematic picture has been devised for the genetic connexions of the various colour vision defects which appear in this pedigree, more can be said about the transmission of the trito-disturbances.

The familial incidence of tritanomaly originally suggested a recessive sex-linked transmission (Hartung, 1926), but Kalmus (1955) proved that tritanopia was due to one or more autosomal dominant genes. He studied the tritanopic subjects of Wright (1952) and their tritanopic relatives; there were 29 tritanopic males and eighteen tritanopic females, a sex incidence which is compatible with neither recessive nor dominant sex-linkage. There were no consanguineous marriages. Our own data are in accordance with the findings of Kalmus. The trito-defects of this pedigree must be due to a dominant gene with incomplete manifestation. In many cases, perhaps, this gene remains completely latent, in others it can cause light tritanomaly (Case 5: III, 4) or severe tritanomaly (Case 4: II, 18); in special cases it may even give rise to tritanopia (? Case 2: II, 10).

Summary

A pedigree is described which includes:

Two cases of tritanomaly;
Two cases of deuteranopia, and three of deuteranomaly;
Two cases of atypical monochromatism, and two of colour amblyopia (with reduced colour discrimination over the entire spectrum).

The cases of atypical monochromatism and colour amblyopia are considered to be combination forms of trito-defects and red-green defects.

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* Walls (Personal communication).
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