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Colour discrimination ellipses in patients with dominant optic atrophy

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Abstract

Many colour tests require a visual acuity of at least 0.1, making them unsuitable for low vision patients. To assess colour vision in patients with sub-normal acuity, we re-designed a previously described test so that its spatial details would be coarse enough to be resolvable by subjects with severe visual impairment. The test measures chromatic discrimination along 20 axes evenly spaced in CIE 1976 L*u*v* colour space. We detail the results for this test in a group of patients with dominant optic atrophy. Despite the lack of evidence for genetic heterogeneity in dominant optic atrophy, we observed phenotypic variation both between and within families.

Keywords: Low vision; Colour discrimination; Dominant optic atrophy; Colour vision deficiency

1. Introduction

Many colour vision tests require a minimum visual acuity of around 0.1 (6/60) [1], although some tests, such as the enlarged D-15, and the computer controlled test described by Arden et al. [2] can be used with the visually impaired. Although this dependence on visual acuity is not a drawback in assessing congenital colour deficiencies, it is a limitation when assessing those patients with colour deficiency secondary to ocular or systemic pathology, because in many cases a colour defect will only develop once visual acuity falls below 0.6 (6/10) [3].

Dominant optic atrophy (DOA) is a hereditary optic atrophy characterised by moderate to severe visual impairment with an insidious onset, a centro-caecal scotoma and dyschromatopsia. The disease follows Mendelian rules for autosomal dominant traits, displaying a high penetrance (0.98), but variable expressivity [4,5]. The onset of the disease is early in life, perhaps even within the first year, although some patients remain undiagnosed until late childhood/adolescence. Visual acuity ranges from being as good as 1 (6/6) to as poor as 0.05 (6/120) or less. The primary ophthalmoscopically visible abnormality is pallor of the optic nerve head, which may be confined to the temporal side of the nerve head. The condition has been linked to chromosome 3q28-qter in French [6], Cuban [7], Danish [8], American [9] and English [10,11] pedigrees, making it increasingly unlikely that a second locus for the condition will be found. The pathogenesis of the condition is as yet unknown, but histopathological studies do suggest that there is a loss of ganglion cells, particularly of those cells constituting the papillomacular bundle [12,13]. These findings are consistent with the electrophysiological picture: patients with DOA show pattern ERG deficits suggestive of ganglion cell pathology [14–16].

It is quite commonly stated in ophthalmologic texts that DOA invariably causes a tritan colour vision deficiency [17,18]. The seminal monograph by Kjer [4] reported that the colour defect is predominantly tritan. His findings were confirmed by Eliott et al., Hansen, Kline and Glaser, and Ohba et al. [19–22]. Krill et al. [23] reported that DOA was capable of mimicking congenital tritanopia, although the magnitude and specificity of the of colour defect could vary dramatically within families. Some families may display predominantly red-green deficiencies [24–27]. Roggeveen et al. 1985 [1] proposed that such phenotypic variations reflect genetic heterogeneity.

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Table 1
Results for normal subjects

Subject	Age	Ellipse (A, B) θ	Subject	Age	Ellipse (A, B) θ
AP	36	R (4.7, 9) 55.5 L (4.3, 8.5) 48.9	MRS	57	R (6.2, 8.7) 132.3 L (7.2, 8.8) 42.4
RO'K	28	R (4.1, 7.5) 47.0 L (4.7, 9.1) 51.4	MP	58	R (5.0, 7.4) 69.8 L (4.7, 9.1) 86.3
JS	28	R (4.7, 9) 53.5 L (6.3, 11) 54.9	TS	32	R (4.7, 10.6) 54.0 L (5.5, 11.6) 53.5
CA	35	R (5.2, 11.1) 73.2 L (5.8, 8.9) 54.4	MS	40	R (6.1, 14.4) 73.2 L (7.3, 11.3) 36.5
PD	34	R (6.7, 9.3) 73.0 L (6.5, 9.9) 79.2	LP	42	R (8.0, 10.3) 73.2 L (8.2, 17.6) 70.29
CG	58	R (4.9, 8.3) 77.0 L (4.4, 7) 79.5	SF	34	R (4.5, 11.1) 74.3 L (5.3, 8.6) 61.4
TPS	46	R (6.4, 10.5) 89.7 L (7.8, 9.0) 177.8	AS	44	R (4.9, 10.6) 52.4 L (5.7, 8) 80.5
LW	36	R (4.9, 9.3) 86.3 L (5.4, 7.8) 74.2	SR	29	R (4.5, 7.5) 72.8 L (4.3, 7.6) 64.9
TW	61	R (5.9, 9.6) 99.0 L (5.3, 7.3) 51.4			

The columns headed 'ellipse' give, for right and left eyes, the lengths of the minor and major axes (A, B) and angle of the major axis (θ) of the ellipse fitted to the chromatic thresholds. Units are 1000 times their value in the CIE 1976 u' v' diagram.

In order to assess colour vision in DOA, we aimed to design a colour test that requires only a minimum level of acuity. In addition the test should be capable of assessing patients with normal acuity, so that colour vision could be assessed in patient groups with a wide array of visual acuities. The same features would also mean that changes in colour vision could be monitored over time in conditions where there is a gradual loss in visual acuity. We have used this test to examine residual colour vision in a group of DOA patients, and to identify the nature of inter- and intra-familial variations in this group.

2. Methods

2.1. Subjects

A group of 17 subjects from nine families was assembled from a molecular genetic study of DOA being conducted at the Institute of Ophthalmology, London. The mean age of subjects was 43.1 year, and the range was 27–67 year. A total of 17 normal subjects from the Cambridge area served as a control group. The mean age of the normal subjects was 41.1 year, with a range of 28–61 year. All the normal subjects passed the FM D-15 and had normal Rayleigh matches. These subjects had a minimum visual acuity of 1 (6/6), had normal fundi, and had no history of ocular or systemic conditions known to affect colour discrimination, and were not taking any medications known to affect colour vision.

2.2. Method of examination

Each DOA patient performed a battery of colour vision tests monocularly, including the enlarged FM D-15 (the 'PV-16', Precision Vision, Villa Park, Illinois), an enlarged version of the 'minimalist test' described by Mollon et al. 1991 [28], the Nagel anomaloscope and the newly designed computerised colour vision test described below. Visual acuity (using an EDTRS letter chart), refraction and direct ophthalmoscopy were also performed at the same visit. Visual field assessment was also conducted on a number of subjects using a standard 30-2 program on the Humphrey visual field analyser.

2.3. Design of the computerised colour vision test

In its underlying principles, the present test resembles the test described by Mollon and Reffin [29] and Regan et al. [30]. The stimuli were generated by a State machine G8 graphics card and an Acorn A5000 computer and were presented on a graphics monitor (Sony GDM-1936). The system was calibrated using a Pritchard PR650 telespectroradiometer, and a Minolta CS-100 chroma-meter. The basic system had a resolution of 8 bits per gun, but the chromatic resolution was increased by 'dithering' [31]: alternating pixels were set to different chromaticities chosen by the computer to produce the closest chromaticity to that desired.

To determine the patient's chromatic discrimination, four discs are presented on a 2 cd/m^2 neutral background in a diamond-shaped array; each subtends 4° at

Table 2
Results for DOA subjects

F/ID	Age	AO	Sex	VA	VF	PV-16	Nagel	M-R test	Ellipse (A,B) θ
B-DB	31	7	M	R 0.1	ST	Two inversions	(18–58) −0.039	T(3),D(3),P(1)	(12.9, 59.3) 100.9
				L 0.125	ST	Two inversions	(11–63) −0.128	T(2),D(1),P(2)	(15.6, 36.1) 105.2
C-JC	67	39	M	R 0.125	—	Tritan	(0–73) −0.438	T(5),D(5),P(—)	(93.5, 113) 108.0
				L 0.2	—	Anarchic	(0–73) −0.386	T(—),D(6),P(—)	(93.5, 109.3) 144.9
D-DB	66	30	F	R 0.125	—	Anarchic	(0–73) −0.006	T(—),D(—),P(—)	(60.8, 137.1) 92.5
				L 0.125	—	Anarchic	(0–73) −0.124	T(—),D(—),P(—)	NA
D-JB	36	10	F	R 0.32	G+ST	Anarchic	(0–60) −0.084	T(—),D(4),P(—)	(53.2, 108.8) 103.9
				L 0.32	G	Anarchic	(0–62) −0.094	T(—),D(6),P(—)	(54.5, 115.9) 105.3
K-JK	61	8	F	R 0.2	S+IN	Anarchic	(0–73) −0.069	T(—),D(5),P(4)	(47.7, 81.6) 124.4
				L 0.16	—	Anarchic	(0–73) −0.101	T(—),D(7),P(—)	(47.7, 135.6) 105.5
K-SM	58	7	F	R 0.32	G+CI	Anarchic	(0–73) −0.031	T(—),D(—),P(—)	(48.5, 144.1) 95.3
				L 0.2	—	Anarchic	(0–73) −0.010	T(—),D(—),P(—)	(63.1, 137) 94.2
K-JJ	38	4	F	R 0.06	CC	Anarchic	Not performed	T(—),D(—),P(—)	(67.5, 110.6) 115.8
				L 0.06	—	Anarchic	Not performed	T(—),D(—),P(—)	(56.4, 130.4) 98.8
M-DTa	55	5	M	R 0.04	—	Tritan	Not performed	T(3),D(6),T(3)	(58.2, 96.9) 113.7
				L 0.04	—	Protan	Not performed	T(4),D(5),P(—)	(73.0, 117.2) 113.9
M-JT	29	11	F	R 0.05	—	Anarchic	Not performed	T(—),D(—),P(—)	(84.7, 110.3) 136.9
				L 0.05	—	Anarchic	Not performed	T(—),D(—),P(—)	(90.1, 126.1) 111
M-JW	34	3	F	R 0.08	G	Pass	Not performed	T(2),D(3),P(2)	(36.7, 74.4) 121.5
				L 0.06	—	Pass	Not performed	T(2),D(2),P(2)	(39.0, 79.6) 121.3
M-SM	33	5	F	R 0.16	G+CI	Anarchic	(0–73) −0.052	T(5),D(6),P(—)	(61.2, 120.2) 116.3
				L 0.1	—	Anarchic	(0–73) −0.061	T(3),D(—),P(—)	(47.2, 79.4) 133.1
M-DTb	27	1	F	R 0.05	G+S	Protan	Not performed	T(3),D(—),P(—)	(45.4, 83.6) 173.5
				L 0.05	—	Protan	Not performed	T(4),D(—),P(—)	(31.7, 99.9) 169.5
O-JW	43	25	F	R 0.8	N	Pass	(36–42) −0.024	T(2),D(1),P(0.5)	(7.7, 9.9) 111.8
				L 0.8	—	One crossing[a]	(38–42) −0.053	T(5),D(2),P(2)	(8.2, 16.4) 89.1
Q-JS	29	4	F	R 0.2	G+CC	Anarchic	(0–73) −0.136	T(4),D(7),P(5)	(38.0, 87.2) 138.5
				L 0.8	—	Four inversions	(37–42) −0.024	T(2),D(1),P(2)	(11.9, 14.4) 86.3
Y-JM	36	5	M	R 0.25	—	Scotopic	(0–73) −0.357	T(—),D(—),P(—)	(51.1, 81.6) 10.4
				L 0.2	—	Scotopic	(0–73) −0.392	T(—),D(—),P(—)	(69.7, 82.2) 177.9
Z-FB	39	2	M	R 0.04	—	Protan	Unreliable	T(4),D(—),P(—)	(20.7, 65.2) 176.4
				L 0.125	—	Protan	(0–73) −0.178	T(2),D(—),P(—)	(22.9, 78.3) 168
Z-M B	41	9	M	R 0.05	—	Anarchic	(0–73) −0.079	T(—),D(—),P(—)	(54.1, 117.3) 117.9
				L 0.05	—	Refused	Not performed	T(—),D(—),P(—)	(70.5, 90.5) 174.3

F/ID, family/subject code; AO, age of onset of symptoms of DOA; VA, visual acuity (Snellen fraction); VF, visual fields; S, superior field defect; IN, inferior-nasal field defect; CI, with central island of sparing; CC, centro-caecal scotoma; G, generalised loss; ST, superior temporal field defect; N, normal; PV-16, enlarged FM D-15.

[a] Subject made one crossing from chip 5 to 15.

Nagel anomaloscope: the values for matching range are enclosed in brackets. Also given (outside brackets) is the gradient of the brightness settings of the 589-nm primary. A deuteranope should have a gradient of around 0, and a protanope a gradient of around −0.384. The (unrelated) patients C-JC and Y-JM displayed protanopic brightness matches, recalling the patients described by Aulhorn and Gruetzner: such patients could simply be suffering from a concomitant X-linked colour deficiency or could represent a distinct phenotypic form of DOA.

M-R, Mollon-Reffin test: numbers in brackets represent the least saturated chips that the subject could discriminate for the protan (P), deutan (D), and tritan (T) axes. A dash indicates that the subject failed to discriminate the most saturated chip.

Ellipse: as for Table 1.

1 m, and is separated by 2.5° from adjacent discs. On each presentation, one of the discs differs in chromaticity from the remaining three (which remain neutral in hue; $u' = 0.211$, $v' = 0.474$ or $x = 0.333$, $y = 0.333$): the subject's task is to determine which disc differs from the others in colour. An oddity task of this kind has the advantage that it is cognitively one of the easiest tasks for a patient to perform. The luminance of each disc is set randomly to a value lying between 6 and 26 cd/m^2, and the chromaticity of the test disc is varied so that colour discrimination is probed along 20 axes spaced 18° apart in CIE 1976 L*u*v* space. The program uses a staircase procedure to determine the minimum saturation at which the coloured disc can be discriminated from the neutral discs. The threshold data, when plotted as points in the CIE 1976 u' v' chromaticity diagram, may be fitted with a least-squares ellipse centred at the chromaticity coordinates of the neutral discs, as described by Regan et al. [30]. In addition, we estimated the achromatic area of the chromaticity diagram by calculating the area of the polygon obtained by joining the points representing the chromaticity coordinates of the threshold values in the CIE 1976 u' v' chromaticity diagram. As Regan, Reffin & Mollon

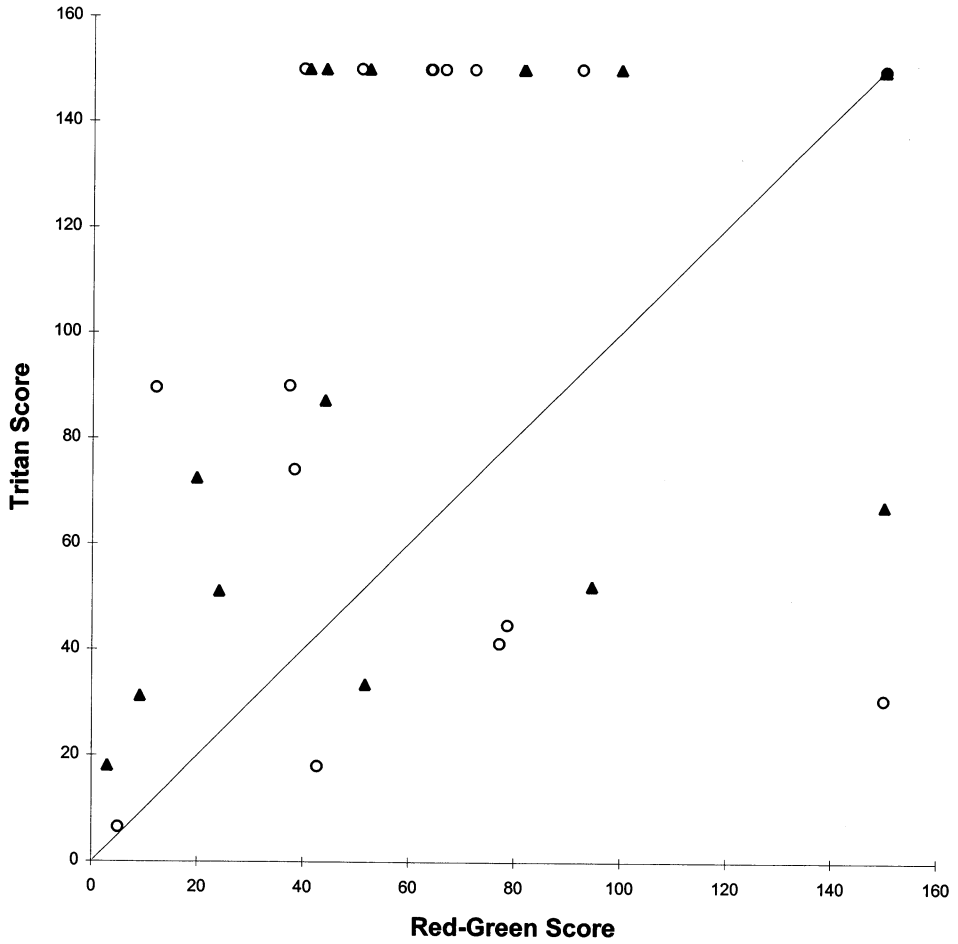

Fig. 1. Tritan versus red-green discrimination in thousandths of CIE 1976 u' v' units. Open circles represent data for the right eye, and closed triangles for the left eye. Tritan discrimination is obtained by averaging the values of threshold for the 270° and 288° axes (0°/180° correspond to the u' axis, and 90°/270° correspond to the v' axis). The values for red-green discrimination were obtained by averaging thresholds for the 342°, 0° and 18° axes. The chromaticity of the screen phosphors enabled greater tritan excursions; for the purposes of this graph the maximum tritan threshold has been limited to 150 (0.15 CIE 1976 u' v' units). The control group had a mean tritan score of 4.71 with a standard deviation of 1.87. The mean value for the red-green score was 3.29, with a standard deviation of 1.28.

note, the origins of this form of colour vision assessment can be traced to the 19th century: Kolbe published discrimination ellipses for daltonians in 1881 ([32]).

3. Results and discussion

The results for the control and DOA groups are summarised in Tables 1 and 2. For the DOA group, visual acuity ranged from 0.8 (6/7.5) to 0.04 (6/150). Even those subjects with severely reduced visual acuity found the test simple to perform.

The mean achromatic area for the DOA group, in square thousandth CIE 1976 u' v' units, was 20920 and the standard deviation 14612. By comparison, the mean achromatic area for the control group was 110.0 and the standard deviation 63.49.

Fig. 1 shows the mean of the patients' thresholds for the two axes closest to a tritan line, and the mean of the thresholds for three axes close to the protan and deutan lines. As a general rule, tritan discrimination was poorer than red-green discrimination when expressed as CIE 1976 u' v' units. However, no patient displayed a truly isolated loss in tritan discrimination when assessed with the complete battery of tests: although B-DB and O-JW (left eye only) had elongated tritan ellipses, they also made red-green errors on the Mollon-Reffin test, and B-DB had a widened Rayleigh match. As might be expected from previous reports of the condition, we observed patients whose ellipses were well aligned to different classical confusion axes, as can be seen from Table 2.

For the averaged data from the right and left eyes, there was no significant correlation between achromatic area and age (Spearman $r = 0.266$). Similarly, the

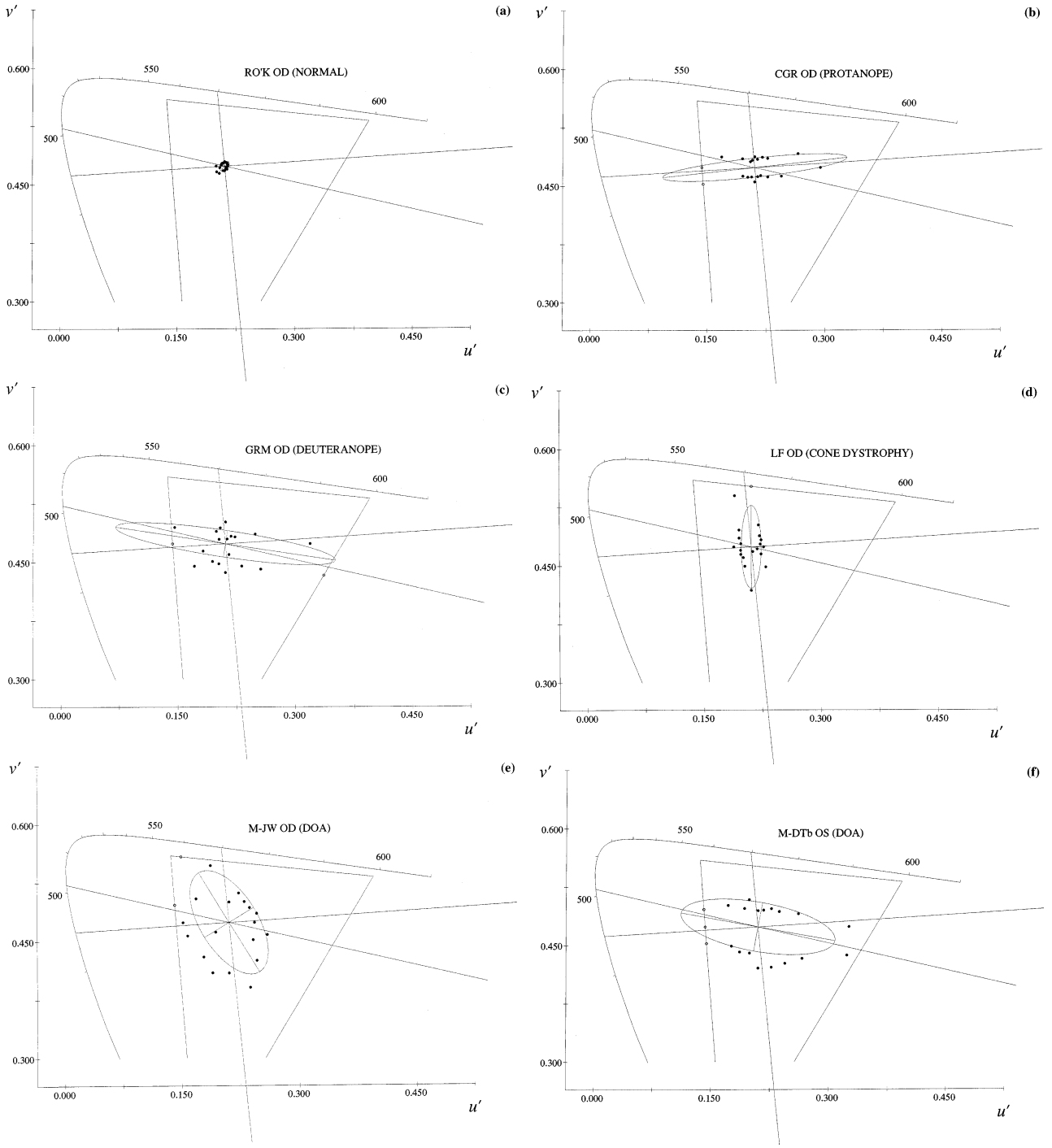

Fig. 2. Thresholds plotted in the CIE 1976 u' v' chromaticity diagram for: (a) a normal subject; (b) a protanope (CGR, aged 42year); (c) a deuteranope (GRM, aged 62 year); (d) a progressive cone dystrophy patient with an acquired tritan defect (LF, aged 22year); (e) and (f) two female cousins, M-JW and M-DTb, with DOA; and (g) and (h) the right and left eyes of patient Q-JS. The incomplete triangle represents the limit of the gamut that could be produced by the monitor. Open circles indicate that the threshold exceeded that which could be produced by the monitor. The line closest to the horizontal represents the protan line, the line closest to the vertical represents the tritan line and the line rotated slightly clockwise from the protan line represents the deutan line. Ellipse dimensions are (12.3, 118.3) 6.2 for subject CGR, (17.1, 142.0) 171.3 for subject GRM, and (12.8, 53.5) 90.2 for subject LF. Details of ellipse dimensions for the remaining subjects can be found in Tables 1 and 2.

Spearman r values for visual acuity and age and for achromatic area and visual acuity were not significant (-0.219 and 0.323, respectively). Our sample consists mainly of those with poor acuity and this may have reduced the correlation coefficient for achromatic area versus acuity. It is warranted to note that the three eyes (from two patients) with the best acuity (0.8 or Snellen 6/7.5) also had the best colour discrimination, in that

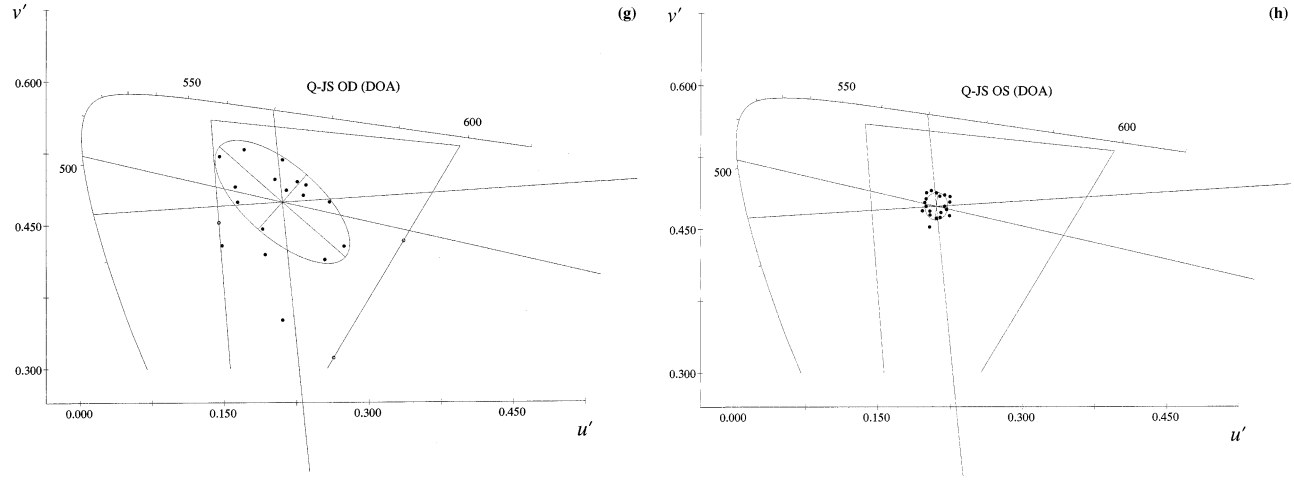

Fig. 2. (*Continued*)

they had the smallest achromatic areas and were also the only eyes with normal Rayleigh matches. However, these eyes did not display perfectly normal colour vision. They exhibited both tritan and red-green errors when tested with the Mollon minimal test and with the SPP plates. On the computerised test, O-JW's red-green scores were 4.9 and 2.9, and her tritan scores were 6.5 and 15 for the right and the left eyes, respectively. The red-green and tritan scores for Q-JS's left eye were 9.3 and 10.4 (the mean red-green score for normals was 3.29, and the standard deviation 1.28; the mean for the tritan score was 4.71, and the standard deviation 1.87).

Of particular interest are two cousins (M-JW and M-DTb) of the same family who displayed a marked difference in residual colour discrimination. As can be seen in Table 2, these patients had a similar level of visual acuity. It is interesting that M-DTb's sister (M-SM) displayed a similarly oriented ellipse to that of her cousin M-JW. The differences observed between M-DTb and other members of her family are unusual: most subjects within families M and K showed an agreement in results. Congenital colour deficiency is unlikely to be the cause of the differences observed between M-DTb and her cousin M-JW, as both subjects are female. The ellipses obtained for these subjects are plotted in Fig. 2e, f. One DOA subject displayed an asymmetry in colour vision between the right and left eyes (Fig. 2g, h). This patient also displayed an asymmetry in acuity: acuity for the right eye was 0.2 (6/30), and for the left, 0.8 (6/7.5).

It is unlikely that variations amongst our patients arose from differences in fixation due to the presence of centro-caecal scotomata. Chromatic discrimination is reduced only very slightly at 10° eccentricity in one normal observer tested (MPS, male aged 24 year): taking the mean of the four quadrants tested (nasal, temporal, superior and inferior) the achromatic area increases by 0.13 log unit. When fixation is 20° from the centre of the screen, then the achromatic area increases by 0.7 log unit. Beyond 20°, there is a marked increase in chromatic threshold. In view of the fact that those patients using eccentric viewing/eccentric fixation did not appear to be using extra-macular fixation when assessed ophthalmoscopically, one may safely assume that they used an area within about 10° of the fovea to fixate the test targets.

Does a genetic heterogeneity underlie the phenotypic variations in the colour vision seen in DOA? Our finding of differences within family M suggests that some of the variation is not genetic in origin; and molecular genetic studies have consistently linked DOA to chromosome 3q28. However, the condition might yet prove to be heterogeneous at the molecular level: in much the same way as the various mutations of the rhodopsin gene give rise to clinically distinct forms of autosomal dominant retinitis pigmentosa [33], there may be a variety of mutations of a single gene all giving rise to DOA. If such genotypic variations were identified, it might prove possible to correlate them with the variations in colour deficiency in DOA.

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