Retinal complications of optic disc drusen

KAZIMIERZ RUBINSTEIN and MUMTAZ ALI
From the Birmingham and Midland Eye Hospital, Birmingham

SUMMARY Following reports by Sanders et al. and Wise et al. of optic disc drusen associated with retinal bleeding, an additional 19 cases were studied and followed up for 1 to 10 years (average 5 years). Four of them showed haemorrhages at the optic disc and its borders only, but 15 showed extensive retinopathy with macular involvement. In 3 cases retinopathy was serous and not haemorrhagic. Apart from cases in which bleeding was limited to the disc and its immediate vicinity the main clinical features of the syndrome were as follows: (1) the presence of optic disc drusen; (2) extensive retinal haemorrhages; (3) intraretinal extravasation of serum with or without the presence of blood; (4) elevated mounds lifting the retina and causing striae radiating from the mounds towards the macula and beyond; (5) pigmented disturbance of the macula or a wider area of papillo-macular bundle after the resolution of acute stages which, while often clinically severe, is compatible with recovery of normal or near normal vision. It is proposed that the syndrome be designated the ‘optic disc drusen retinopathy.’

The association between optic disc drusen and retinal haemorrhages involving the macula was first described as a clinical syndrome 10 years ago by Sanders, Gay, and Newman. Of the 8 cases presented, 3 showed haemorrhages around the optic disc only, as had been described occasionally in the past by several authors, but the remaining 5 showed a new clinical association. In their perusal of the literature Sanders et al. traced only 9 sporadic references to bleeding associated with the presence of disc drusen, starting with Gifford’s report. All these cases were concerned with the bleeding at or in the immediate vicinity of the optic disc, some with vitreous haemorrhage but none with a more extensive retinal or macular involvement.

Henkind, Alterman, and Wise reported 4 cases of retinal haemorrhages associated with drusen, and an additional 4 cases were reported by the same workers 2 years later. Of their 8 cases 5 had retinal involvement while 3 presented with haemorrhages round the optic disc only. Of the 2 cases reported by Brodrick, one was affected by pseudoxanthoma elasticum, a disease typically connected with retinal haemorrhages, and the second case showed disc splinter haemorrhages alone. Of the 6 cases reported by Ostradovec and Vladykova only 3 presented with disc haemorrhages the other 3 showed gross venous abnormalities of the disc and the retina. This brings to 11 the number of cases on record of disc drusen retinopathy and to 19 the number of cases in which only the disc and its immediate vicinity were affected.

Patients and methods
The clinical material we review consists of 19 personal cases collected during the past 10 years. There were 10 males and 9 females. The age of the patients was as follows: 12–25 years, 8 cases; 26–40 years, 3 cases; 41–60 years, 7 cases; over 60 years, 1 case. The mean age was 38.5. The right fundus was involved in 12 cases, the left in 10 cases (3 cases showed bilateral involvement). Follow-up time was between 1 year and 10 years (mean 5 years), but this figure is based on an analysis of 16 patients only; 3 patients for various reasons could not be followed up. The follow-up included a repeated thorough medical, neurological, ophthalmic, and photographic examination, as well as laboratory tests, particularly aimed at the possible existence of immunological, parasitic, and haematological disease. None was demonstrated in any patient. Fluorescein angiography was done in all cases; the presence of drusen was sometimes revealed in the fellow eye only by angiographic evidence. In all cases but one the presence of drusen of the optic disc,
buried or adjoining, was demonstrated in the fellow eye.

In 4 cases the optic disc alone and its immediate vicinity were affected by haemorrhages. In the other 15 cases extensive retinal involvement was present.

According to the clinical manifestations at the time of first attendance the cases will be described in the following order: group I. 4 cases (1–4), where only the disc and its immediate vicinity were affected by haemorrhages (Fig. 1); group II. 9 cases (5–13), where the retina was dissected by haemorrhages extending from the optic disc (Figs. 2, 3, 4, 5, 6); group III. 3 cases (14–16), where the retina was dissected by serum dextravasation (without erythrocytes) extending from the optic disc towards the macula (Fig. 7); and group IV. 3 cases (17–19), where a late burnt-out clinical picture was seen with pigment disturbance and scarring of the retina (Figs. 8 and 9). All affected eyes showed disc drusen of various shapes and locations.

Fig. 1 Case 1. (a) Fundus at first attendance. (b) Six months later. (c) Fluorescein angiogram at first attendance. (d) Six months later.
Retinal complications of optic disc drusen

Case reports

Case 1
A 25-year-old man, an insurance representative, was first seen in June 1973. Nine days previously he noticed some blurring of vision in the left eye. 2 days later a floater was noticed too. His visual acuity was 6/5 each eye. His left eye showed a radiating halo of 'deep' haemorrhage around a swollen disc with a beady crescent of fresh looking blood at its temporal edge. Red-free photography located all these haemorrhages in front of the pigment epithelium and some even in front of retinal vessels. Fluorescein angiography showed lumpy staining of the optic disc. During the following 3 months the vivid marginal blood disappeared and the main haemorrhagic halo faded. Two months later all haemorrhages were gone. When examined last in 1981 (8 years follow-up) the disc looked flat but without physiological cupping. There were no further episodes of blurred vision, which was 6/5 each eye (Fig. 1).

Case 2
A 48-year-old engineer was examined in February 1977 by an optician who tested him for reading glasses. Visual acuity was 6/5 each eye and there were no symptoms. The left optic disc had a hazy outline and seemed large. At the 12 o'clock meridian there was a fine meshwork of just visible vessels and a small haemorrhage at the edge of the disc on the 3 o'clock meridian. Four months later a string-like horizontal

Fig. 2 Case 6. (a) Fundus at first attendance. (b) Four weeks later. (c) Fluorescein angiogram at first attendance.
haemorrhage was seen across the nasal half of the disc, and a second round, dark haemorrhage was present at the 6 o'clock meridian. Vision was not affected. Six months later only the deep, round haemorrhage below the disc was present. Red-free photography placed this haemorrhage in front of the pigment epithelium. Six months later no haemorrhages were present. The disc outline was deformed by drusen, but its margins were now clearly defined. The fundus remained unchanged until 1980, when the patient was last seen.

**CASE 3**
A 55-year-old man was first seen in October 1973 complaining of gradual loss of clear vision in the left eye for the previous 2 months. He could see 6/18 with this eye; the other eye could see 6/9 (corrected). Both eyes showed high mixed astigmatism. The fundi showed swollen optic discs and abnormal, crowded vessel entry. Both fundi showed extensive drusen in the vicinity of but not in contact with the optic disc. The left disc showed a small haemorrhage at its nasal side. Neurological examination proved negative and remained negative for all 8 years of observation. In June 1974 the fundus showed a 'deep' haemorrhage all round the disc, especially vivid at its temporal margin. Vision was unchanged. In August 1974 the haemorrhages were remarkably diminished and in December quite gone. Eight months later a deep diffuse haemorrhage recurred at the temporal margin of the disc. The right disc now also showed a deep marginal haemorrhage at its temporal margin, and fluorescein angiography showed diffuse leakage of the dye towards the macula. In March 1976 there was still a rim of haemorrhage in the same location, while the originally affected left eye showed no haemorrhages. In October 1977 both discs were free of haemorrhages and looked much flatter. Both showed limited fluorescein leakage towards the macula. Thereafter, up to 1981, there were no haemorrhages. Visual acuity was 6/12 right eye, 6/18 left eye. Both discs were clearly defined.

**CASE 4**
A 34-year-old man was seen in 1959. He was neurologically diagnosed as suffering from tuberculous sclerosis. His face showed marked sebaceous adenoma. The vitreous of his left eye showed a small globule of an astrocytoma and an optic disc distorted and obliterated by a large, scaly, sharp-edged conglomeration of drusen. At the lower edge of this drusen mound retinal haemorrhages were present.

**CASE 5**
A 42-year-old typist was seen first in April 1971 with a complaint of failing sight in the left eye for the previous 2 months and a distortion of central vision for the previous week. Vision was 6/18 with this eye and 6/5 with the other. The left fundus showed a somewhat disfigured optic disc clearly affected by buried drusen. About half way towards the macula there was a wide crescent of retinal haemorrhage, and splashes of small haemorrhages were seen along the lower temporal vessels. Red-free photography showed all these haemorrhages to be intraretinal in front of the pigment epithelium. Fluorescein angiography showed early fluorescence from the marginal disc drusen, expanding to engulf the optic disc and towards the macula in the early venous stage. During the following 4 months all haemorrhages were absorbed, but a dark crescentic new haemorrhage appeared close to the lower temporal quadrant of the disc. Visual acuity was 6/12. This took 18 months to absorb, with vision improved to 6/6. When last seen in 1981—10 years after the onset—the optic disc looked flat, and on its temporal side was a round disc-sized area of fine thinning of the pigment epithelium. Between the two, at the disc edge, was a sharp splinter of black pigment. The macula seemed normal. Vision was 6/5 each eye.

**CASE 6**
A woman part-time cleaner aged 25 was first seen in April 1980 when she complained of light flashes to the right of the fixation point and a hazy film affecting the right eye for the previous few days. Her vision was 6/5 each eye. The right fundus showed a large, grey swollen drusen body at the upper temporal edge of the disc and extensive retinal haemorrhages between the disc and the macula. Fluorescein angiography showed a strong, early glow of the disc extending towards the macula. Four weeks later all haemorrhages were gone, as were her symptoms. A glistening, yellowish, elevated crescent of drusen remained at the upper temporal edge of the disc. The macula showed fine pigment stippling. The eye remained unchanged for the remaining 12 months of follow-up (Fig. 2).

**CASE 7**
A 12-year-old schoolboy was first seen in August 1971 complaining of blurred vision in the left eye. Vision was 6/36 with this eye and 6/5 with the other. The left optic disc was fringed temporally by a whitish, elevated drusen body which showed a crescent of haemorrhage convex towards the macula. Striate oedema streamed from the drusen body.

Fig. 3 Case 7.
Retinal complications of optic disc drusen

toward and across the macula, which was spattered by sub-neuroepithelial deposits. Two months later vision was 6/12 and the fundus was free of haze. The disc adjacent to the drusen body was better defined and smaller. Some neuroepithelial detachment with posterior yellow dusty deposits persisted. Fluorescein angiography showed late but increasing staining of the drusen body but not of the disc. Five months later visual acuity was 6/6. Three years later (1975) vision was 6/12. The macula showed pigmentary disturbance. The drusen body was oval, white, with black granular deposits, and seemed separated from the disc. The patient then moved away, but to our recent inquiry (1981) replied that he had had no further trouble with his eyes and that he could see well (Fig. 3).

Case 8
A 52-year-old housewife was first seen in March 1976. She noticed gradual deterioration of vision in the right eye for the previous 3 months. Her vision was 6/36 in this eye and 6/4 in the other. The fundus showed the optic disc to be deformed by multiple buried drusen. Close to the large drusen located in the temporal area of the disc, but separated from it, there was a slaty, small lump of pigment. There were a few small retinal haemorrhages within the lower half of the arcuate vessel area. The macula showed a round swelling of serous detachment with a cluster of brush-like white exudates in the nerve fibre layer and some depigmentation. Red-free photography showed the peripheral haemorrhages to be intraretinal but the slaty mound near

Fig. 4  Case 8. (a) Fundus at first attendance. (b) Fluorescein angiogram. (c) Fluorescein angiogram of the other eye.
the disc subretinal. Fluorescein angiography showed a strong penetration of the dye from the glowing disc towards the macula, and several drusen round the disc margins which were not seen ophthalmoscopically. There were no recurrences during the following 5 years. The slaty mound became a whitish thin scar with scattered pigment dots; the macula now showed extensive pigment scatter and depigmentation. Vision returned to 6/12.

The other eye showed—from her first attendance—an exuberant multiple small drusen of the optic disc, distorting its outline, particularly along the superior temporal vessels, where a prominent separate drusen body was placed far from the normal disc territory and bordered nasally by a small haemorrhage. Fluorescein angiography showed a diffuse pooling of the dye over all the area and the disc. When it was seen 5 years later (1981) there was a marked increase in size and number of these drusen. Her visual acuity was 6/6 (Fig. 4).

CASE 9
A healthy girl of 23 was first seen in February 1976 complaining of distortion of vision in her right eye, which had started 2 months previously. During the previous 4 weeks the distortion disappeared and her vision became very poor, but it had been improving since then gradually. Visual acuity was 6/12 with this eye and 6/4 with the other. The right fundus showed a pink disc with filled physiological cupping. At its temporal side there was an elevated crescentic roll of greyish colour, a small intraretinal haemorrhage bordered it inferiorly, and another small haemorrhage was seen nearer to the macula. The macula was elevated by a disc-sized serous detachment with a sprinkling of flecked, dust-like, yellow deposits, and retinal striae ran across it from the disc drusen towards the periphery. Fluorescein angiography showed intensive glow of the drusen body, which involved even in later stages only the temporal part of the disc. It spread, however, towards the macula and along the superior temporal arcuate vessels. For the next 3 months there was a gradual drying of the fundus and vision improved to 6/9. When seen in 1981—5 years after onset—visual acuity was 6/5; the macula showed a very delicate stippling only. The disc drusen body was shrunk, with patchy dense pigmentation (Fig. 5).

CASE 10
A 60-year-old postal supervisor was first seen in April 1978. Two months previously he noticed a dense shadow in the right eye, which since then decreased in size. His vision was 6/24 in this eye: vision in the other eye was 6/6 and was normal. The right fundus showed blurred disc margins with 3 small satellite drusen and an intraretinal sheet of haemorrhage extending from the temporal disc margin towards the macula. The macula showed cystic degeneration. Fluorescein angiography showed a strong glow of the disc and the main drusen, extending towards the macula along the superior arcuate blood vessels. During the following months the haemorrhages were gradually absorbed without trace and vision improved to 6/12. Three years after the onset a small whitish linear scar was seen at the 10 o'clock meridian of the disc: the vision remained 6/12 in spite of marked pigment disturbance at the macula.

CASE 11
A 30-year-old window cleaner was first seen in November 1971 complaining of gradual loss of vision in the left eye for the previous month. It was 1/60 in this eye; vision in the other eye was 6/4. A wreath of retinal haemorrhages round the disc extended from the 10 o'clock radius to the 6 o'clock meridian. The centre of this wreath was deeply oedematous. From this swollen centre some striae stretched over the macula. Fluorescein angiography showed copious infusion of the dye into the area, extending towards the macula. Marked disturbance of the pigment epithelium affected the oedematous central area. The equatorial periphery of either eye showed many small yellowish spots which stained with fluorescence. Within 4 weeks there was considerable absorption of haemorrhages and flattening of the central oedema. A large drusen body at the temporal half of the disc came to light. All clinical investigations, particularly those concerned with parasitic infection, proved negative. During the following year there was only slow absorption of retinal haemorrhages and striae, very incomplete at the patient’s last attendance. His vision never improved, but the affected eye showed hypermetropic astigmatism—the right eye was emmetropic—and a large central scotoma in the affected eye could have been suppressive. The patient was followed up for 12 months only, when he emigrated overseas.

CASE 12
A 25-year-old waitress was first seen in February 1976 with a history of an attack of blurred vision of the right eye 6 months ago, lasting for a few weeks. The sight in her left eye became blurred again 4 months later but 2 weeks before attending here showed much improvement. Vision was actually 6/5 in each eye when examined. Both fundi showed a disfigured optic disc outline and clumpy pigmentation at the maculae. Four weeks later sight in the left eye became blurred, down to 6/36. Fluorescein angiography showed early strong glow of the optic disc and stippled pigment disturbance between the disc and the macula, which eventually was engulched from the disc effusion, and this progressed temporally to form a glowing ring round the macula. A few small haemorrhages were seen near the temporal disc edge. The vision in the left eye became 6/5 within 4 weeks, but 3 months later the vision of the right eye failed to 2/60. There was an extensive oedema of the posterior pole of the eye but no haemorrhages. Two weeks later visual acuity improved to 6/9, but vision in the left eye dropped to 5/60, and the fundus showed similar extensive oedema of the posterior pole. During the next 2 months the vision of both eyes improved to 6/5 and in the following 2 months to 6/4. Both fundi showed delicate pigment clumping at the maculae, and in the right eye a large triangular scar was present temporally to the macular area over the previous limit of oedema. There was no change in the fundi in the following 4 years.

In June 1980, however, the patient returned complaining of blurred vision in the left eye. The visual acuity was 6/5 in each eye, but on the nasal side of the left eye a prominent marginal drusen body was surrounded by a sheet-like retinal haemorrhage extending nasally. Fluorescein angiography showed massive fluorescence of the whole optic disc. In 3 months all haemorrhages disappeared. There was a marked increase in elevation of a drusen body at the 11 o’clock
Retinal complications of optic disc drusen

Fig. 5 Case 9. (a) Fundus at first attendance. (b) Five years later. (c) Fluorescein angiogram at first attendance, early stage. (d) Later stage.

meridian, and new drusen appeared at the lower nasal edge of the disc. The visual acuity of the left eye dropped to 6/6 (5 years’ follow-up).

CASE 13
A 16-year-old girl in the 5th month of pregnancy was first seen in January 1976 complaining of loss of sight in the left eye for the previous 4 weeks. She could only count fingers with this eye; vision in the other eye was 6/4. The fundus showed an optic disc without clear margins and deep central white cupping. Its temporal edge was peaked towards the macula and showed strands of pigmentation. At the 3 o’clock meridian a square pigmented mound was seen and a separate plum coloured globule of blood nearly touching the macula, which showed a small, separate haemorrhage and oedema. Fluorescein angiography showed a strong progressive stain from the disc towards the macula. The fundus periphery and equator showed numerous spots characteristic of histoplasmosis, some with a pigmented core and some yellow and unpigmented. The other eye showed the same kind of peripheral spots and a similar disc, but no bleeding or pigment disturbance. During the following 2 years the
vision slowly improved to 6/18 and the areas of pigmentation became much lighter in colour. In particular the para-macular mound changed its shape into a doughnut-like pigmented ring with a red centre. This slowly became white, and the ring became broken up into small pigment clumps showing against the white background. The 'histoplasma' spots never changed. Extensive and repeated laboratory investigation, particularly for parasitic infection, were negative (Fig. 6).

CASE 14
A 47-year-old electrician was first seen in December 1975 complaining of blurred vision in the right eye for the past 3–4 months, vision was 6/9 in this eye and 6/5 in the other.

The fundus showed an optic disc with softly defined edges and no physiological cupping. Above and temporally a little distance from the disc edge there was an isolated, sharply defined, small whitish drusen body. The central retina and vicinity of this drusen body showed a dusting of the posterior retinal layer with numerous yellowish spots. Fluorescein angiography showed buried drusen distorting the outline of the disc as they took the dye. Only in one direction was there an expansion of the disc glow, temporally towards the macula. When the patient was last seen in 1981—6 years after onset—vision was 6/5 with each eye. The isolated drusen body was only marginally smaller and sharper in outline. Punctate yellowish dust could still be seen in the macular area (Fig. 7).
Retinal complications of optic disc drusen

CASE 15
A 33-year-old housewife was first seen in April 1975 with a 2-week history of disturbance of the upper visual field of the left eye. On examination her vision was 6/6 in this eye; the other eye 6/5, but there was some oedema of the macula. Her symptoms disappeared during the following 6 weeks. In July 1976 she reported back as an emergency because of loss of sight, again in the left eye, which on examination could see 6/24 part. The optic disc showed marginal drusen along its temporal margin, and fluorescein angiography showed a signet-like extension of optic disc glow towards the macula. The vision returned to 6/5 in 4 weeks. In January 1977 she again returned complaining of blurred vision of this eye, and its vision was 6/9 only, while the right eye could see 6/5. A similar picture was seen with the ophthalmoscope and by fluorescein angiography as that seen previously. The condition settled within a few weeks, and when examined last in December 1980 she could see 6/6 right eye, 6/5 left eye. Apart from distortion of the optic disc margin there were no traces of past episodes.

CASE 16
A 67-year-old retired man was first seen in November 1976. Four months previously he noticed blurred vision in the right eye, which after a while started clearing. Vision in the right eye was 6/9, in the left 6/5. The right fundus showed a somewhat elevated whitish crescent at the temporal edge of the disc which showed no physiological cupping. A widespread, delicate stippling of the retina of the posterior pole was noted. Fluorescein angiography showed early glow at the temporal edge of the disc, extending signet-like towards the macula. When he was seen last in 1981—4 years' follow-up—both eyes could see 6/5. The right macula showed fine stippling.

CASE 17
A 62-year-old man, first seen in February 1976, was referred from an optician to whom he went for a replacement of reading glasses. His vision was 6/6 right eye and 6/9 left eye. There was no history of any previous eye trouble. The discs were pink and showed multiple marginal drusen. The macular areas showed irregular scarring of pigment dystrophy bridging from the temporal disc margins towards the maculae. The picture was practically identical in each eye. Fluorescein angiography showed large irregular areas of pigment dispersal within the papillomacular bundle, with pigment scattering reaching from the disc to the macula. Drusen were clearly outlined all round the discs. The

Fig. 7 Case 14. (a) Fundus at first attendance. (b) Fluorescein angiogram.

Fig. 8 Case 17.
macular function was retained at 6/6, 6/9 vision level for the following 5 years of observation (Fig. 8).

**CASE 18**
A 17-year-old girl attended an endocrinological clinic in her childhood because of a suspicion of retarded growth, and the idea was entertained that she might be suffering from a tumour of the upper brain stem. All investigations including a CT scan were negative, and no endocrine symptoms were discovered over more than 10 years of neurological follow-up. It was only in 1980 that, because of complaints of headaches, she was again seen by neurologists, and an abnormality of her right fundus was noticed; the disc was suspected of showing oedema. Electrodiagnostic tests and the electoretinogram were all negative, and her visual fields were full. On ophthalmic examination the patient's visual acuity was found to be 6/24 right eye, 6/9 left eye. The right fundus showed marked distortion and drusen of the optic disc, a granular track of pigment dispersion between the disc and the macula, and atrophic spotty and patterned macular dystrophy. The left fundus showed a small disc surrounded by dark crescents and feathery outline, typical of drusen, which were confirmed by fluorescein angiography. It showed a normal macula. The picture is interpreted as a quiescent, late manifestation of drusen of the optic disc connected with haemorrhagic or serous infiltration of the retina.

**CASE 19**
A 53-year-old man was seen in 1977 after an abnormality of the right fundus was noticed by an optician. His vision was 6/9 in right eye, 6/5 in the left. The fundus showed buried drusen of the optic disc and pigment clumping area from the disc towards the macula, which was involved. Fluorescein angiography showed strong fluorescence of the disc, extending some distance towards the macula signet-like. An area of depigmentation was seen near and below the disc edge. The other disc showed buried drusen but a normal macula (Fig. 9).

**Comments on case reports**

**GROUP I**
The 4 cases in this group showed haemorrhages of the optic disc and its immediate vicinity only. Case 4 was seen only once by consultation. His clinical notes were lost, and only the photographic evidence of his adenoma sebaceum and intraocular features of tuberous sclerosis were preserved. The fundus is similar to that described by Gifford, but no mention of an underlying general disease was mentioned in that report. The other 3 cases had very slight symptoms or no symptoms at all, with visual acuity 6/5 in cases 1 and 2, 6/9 in case 3. They had recurrences, case 3 for 3 years, each eye being affected at different times, and recovering within 8–12 months each time. In no case did the blood break through the internal limiting membrane into the vitreous, as described by others, and in no case was the sight permanently diminished by the episodes.

Extensive neurological investigations were all negative, and within a follow-up time of 8 years in 2 cases, and 4 years in the third, no general health problems arose.

The haemorrhages in these patients look darker and they were considered to be 'deep'. This is an optical illusion. By the use of the slit-lamp and red-free filters for observation and photography they can be proved to be superficial, in the nerve fibre layer or intraretinal. In our case 1 some were demonstrated in front of the retinal vessels but still behind the internal limiting membrane (Fig. 1).

**GROUP II**
Nine cases in this group (5–13) constitute the classical symptomatic haemorrhagic retinopathy with macular involvement. Apart from 2 patients who attended within a week because of flashes of light and some distortion of vision, most patients attended with a history of 2–4 months' deterioration of sight, which at this stage varied between 1/60 and 6/5 (average 6/18). Apart from the patient with 1/60 vision, of the other 8 cases 5 recovered their sight to 6/5 level and none to worse than 6/18. The visual recovery was usually parallel with the absorption of haemorrhages. The absorption was complete in 3–4 months, and this applied to recurrences as well, of which there were 1 in case 5, 5 in case 12, and 3 in case 15.

Case 5 was particularly interesting because, while the original fundus picture was classical for this group, with bright red extensive haemorrhages, the recurrence 4 months after the first attack took the shape of a 'deep' disc margin bleeding only—similar to the
Retinal complications of optic disc drusen

Group I pattern—and this took all of 18 months to be absorbed without disturbing the recovery of visual acuity, in contrast with a few months’ absorption of haemorrhages in other patients in this group.

Case 12 was also interesting. While the left eye was originally affected—with a recurrence—by a serious extravasation only, the last attack took the shape of classical sheath haemorrhages. Until that event the diagnosis was very tentative. Case 12, with cases 11 and 13, merits special notice. These 3 cases showed peripheral foci of choroidal scarring—‘histoplasma’ spots—and case 12 was observed to develop scarred areas through stages of localised oedema. All investigations, particularly aiming at parasitic infection, were negative. ‘Histoplasma’ spots in the affected eye (case 7) were described by Wise et al. It may be of interest to note that the average age of these patients with additional ‘histoplasma’ changes was distinctly lower than overall, 23.5 as opposed to 38.5 (Figs. 2–6).

Group II

In cases 14, 15, and 16 the intraretinal extravasation was serous and not haemorrhagic. Case 12 combined both. All patients in this group attended with complaints of blurred vision for the previous 4 to 6 months, and the recovery took a few months, just as in the cases of group II. Case 15 had 2 recurrences; the visual impairment was 6/9 (recovering to 6/5), 6/24 (recovering to 6/5), and 6/9 (recovering to 6/5). At the macula, or in a more extensive papillomacular bundle area, signs of schisis were present, with dusting by white specks of posterior retinal lamina. In all cases fluorescein angiography showed a characteristic signet-like stain of the disc with its drusen extending towards the macula.

The mean age of the patients was 49, considerably higher than overall for the other groups (Fig. 7).

Group IV

Three patients in this group were 17, 53, and 62 years of age, with visual acuity 6/24 6/9, 6/6 6/9, and 6/5 6/9 respectively in the right eye. They were asymptomatic. Disc drusen were bilateral and showed a typical signet-like extension towards the macula on fluorescein angiography. There was a marked contrast between the degree of preserved good vision and extensive pigment dystrophy affecting the papillomacular bundle area. Case 7 of the St Louis series and case 6 of the New York series showed similar fundus changes. A similar contrast was seen in most cases of groups II and III, where the function of the macula seems to have recovered in time in spite of marked pigment thinning or clumping (Figs. 8 and 9).

Discussion

The purpose of putting on record this clinical material is to add a considerable number of cases to those hitherto reported. The opportunity to follow them for a period of up to 10 years (average 5 years) helped to establish the natural history and the character of the syndrome. We propose to call it the Optic Disc Drusen Retinopathy. The manifestations of the syndrome areas follows.

Drusen

Drusen of the optic disc which may be ‘buried’ and clinically manifest only as a filling of physiological cupping and an impression of a small pink dark circled optic disc, but where fluorescein angiography reveals their presence. The drusen may affect the marginal area of the disc, clinically deforming it. The drusen may also seem to be separated from the edge of the disc, although usually close to it, but fluorescein angiography in these cases demonstrates a plateau of staining, involving both the drusen and the disc.

Haemorrhages

Haemorrhages start always at the edge of the disc or an extending drusen. They may be limited to the disc surface (splinter haemorrhages) or its rim and immediate vicinity (group I of our series). When located at the rim they seem dark red and used to be described as ‘deep’ haemorrhages. They can be proved by the use of red-free photography to be quite superficial (intra-retinal) and some were demonstrated to be placed in the nerve fibre layer, in front of the retinal blood vessels. The darker colour and slowness of absorption of some ‘deep’ haemorrhages may signify their location posterior to the neuroepithelial layers of the retina, but still in front of pigment epithelium. A fine meshwork of disc surface vessels may accompany them. They may be recurrent, but they do not permanently interfere with visual acuity. They may rupture through the internal limiting membrane into the vitreous.

A second type of haemorrhage while starting at the edge of the disc extends from there temporally in bright red sheets and splashes. The haemorrhage may originate at a drusen body located at the nasal edge of the disc and then it extended into the nasal retina. The resolution is identical with the temporally located haemorrhages, but the macula is clearly not involved. In all other cases the haemorrhage even if not quite reaching the macula is preceded by an area of fluid dissecting the retina.

These haemorrhages can be proved to be intraretinal by slit lamp observation or red-free photography. A change in the character of the haemorrhage from this type to the ‘deep’ disc edge type was observed.
on recurrence. The absorption of this 'deep' haemorrhage takes many months, while the peripheral haemorrhages absorb within a 2–4 months period.

**SEROUS EXTRAVASATION**

The haemorrhages dissecting the retina from the direction of the disc towards the macula as a rule do not reach the macula itself. Their advancing edge is, however, preceded by serous effusion which does. Optic disc drusen maculopathy may present without haemorrhages, but with serous extravasation only. It causes similar visual symptoms and it takes a similar amount of time to regress. The signs of retinal schisis may be very clear in these cases and the fluorescein angiography shows the usual disc drusen pattern. The change from the serous type of retinopathy to the classical haemorrhagic type was observed in a recurrence.

We propose that the vascular extravasation could either be severe causing bleeding, but in less compromised circumstances only the serum may leave the blood vessels.

**RETINAL MOUNDS**

The degree of macular oedema and the oedema of the drusen themselves may be severe in some cases, throwing the retina into striate folds and tenting it from the drusen across the macula towards the periphery. The drusen in these cases are very elevated and situated over the disc edge or even seemingly outside its outline. As the resolution progresses these drusen, often covered initially by haemorrhages, settle, shrink, and eventually lose their slaty colour. They either become whitish ovoid nodules sprinkled by black pigment spots or form solid greyish-yellow scars. In spite of being usually placed in the papillo-macular bundle area at the temporal side of the disc they cause no nerve bundle scotoma. It must have been the early clinical stage of such a case which led to the erroneous diagnosis of a melanoma in Sanders et al.’s report on case 3.

**‘HISTOPLASMA’ SPOTS**

Although not a regular feature of the syndrome they are worth noting. In 2 patients the equatorial areas of both fundi showed a scatter of small, round, yellowish spots, some with central pigment clumps. In one of them a slaty, irregular tract of pigmented scar was seen in the temporal periphery. In another case small choroidal scars developed during the follow-up. The process was observed as starting with focal retinal oedema without signs of inflammation. Similar ‘histoplasma’ spots were described by Wise et al.³ in their case 7. It is tempting to consider the whole optic disc drusen retinopathy syndrome as a manifestation of parasitic infestation, but in neither of these cases, or indeed in any reported cases, were the results of specific tests abnormal. The significance of 4 such affected cases among those on record is unclear.

**DIFFERENTIAL DIAGNOSIS**

The cases of optic disc drusen retinopathy confined to bleeding at the optic disc only (group I) require differentiation from papilloedema; with the use of fluorescein angiography this has ceased to be a major problem.

The cases presenting an acute picture with retinal haemorrhages require differentiation from disciform senile degeneration, parasitic infestation, and melanoma. The differentiation from disciform degeneration is important in respect of prognosis. Optic disc drusen retinopathy is likely to resolve within a few months with restoration of vision (Fig. 10), while Bruch's membrane degeneration leads to serious loss of sight. It is the demonstration of drusen and the absence of subretinal vessels which are crucial. The age of the patient is a much less valuable help in diagnosis. Patients over 60 are among those affected by optic disc drusen retinopathy, while patients of any age may develop disciform response foci.

*Fig. 10 Visual acuity changes, groups I, II, and III (including 4 recurrences).*
Retinal complications of optic disc drusen

Parasitic infestation should be accepted only if appropriate laboratory tests prove positive. To mistake a pigmented posthaemorrhagic mound for a malignant tumour would nowadays be unlikely.

The bloodless forms of the syndrome should be kept in mind in cases of spontaneous macular retinoschisis, and here the use of fluorescein angiography with its typical signet-like staining of drusen, which may not be obvious clinically, is helpful. Even the development of pigmentary macular changes is compatible in this type of maculopathy with preservation of good vision. The burnt-out, late picture of optic disc drusen retinopathy may help in the diagnosis of some macular degenerations, especially if unilateral, irrespective of age. Finally, the very crucial characteristic of the optic disc drusen retinopathy is the fact that it is a self-limiting disease with a good visual prognosis.

Histopathology

Sanders et al. reported histological findings of a 12-year-old boy's eye which was enucleated on the suspicion of a malignant-melanoma-like nodule situated near the temporal edge of the optic disc (case 3). The histological section showed a haematoma located between the neuroepithelial retina and pigment epithelium which seemed intact. The section also showed large drusen of the affected disc.

Our clinical observations clearly fit the histologically demonstrated localisation of the haemorrhage in front of the epithelium. They place the haemorrhages and serous extravasation certainly in front of the pigment epithelium and probably wholly intra-retinally—between the internal and external limiting membranes. In some cases pigment epithelium showed in later stages patchy depigmentation, but there was never any evidence of blood vessels invading under the retina through Bruch's membrane. This fundamental difference between optic disc drusen retinopathy and those connected with Bruch's membrane insufficiency could explain the very different prognosis and self-limiting, benign course of optic disc drusen retinopathy.

On pathogenesis and aetiology of optic disc drusen retinopathy we can only offer speculation. Drusen of the optic nerve are always located in front of the lamina cribrosa. The area is supplied by small vessels from the ciliary circulation, and it is from these vessels that the extravasation must come. Whether it is because of mechanical injury to the vessels by sharp edges of the calcified drusen, or their pressure on the venous outflow, whether the congestion and subsequent bleeding are caused by choking of the optic disc by orthograde axoplasmic neuronal flow and mitochondrial debris we do not know.

The second unknown is related to the way in which the blood specifically finds its way to dissect the retina or its interface with pigment epithelium. The movements of blood would then have to be through the junctional tissue of Kuhnt, where the retinal layers end at the margin of the optic nerve. In this respect there seem to be some similarities between disc drusen retinopathy and retinopathy connected with optic disc pits.

An increased awareness of the syndrome would certainly make a larger number of cases available for study. An extended follow-up time, combined with a donor eye index, should in time yield necropsy material for electron microscopy of the crucial junctional area between the retina and the optic disc.

References