Atypical minimal peripapillary choroidal colobomata

SOHAN SINGH HAYREH AND JAMES F. CULLEN

Department of Ophthalmology, The Royal Infirmary and University of Edinburgh

Many reports of colobomata of the choroid, involving the peripheral choroid, are available in the literature, but no case of a localized congenital choroidal defect in the peripapillary region has been reported. A case of such colobomata present on both sides is reported here which, since coming under observation 10 years ago, has presented a considerable diagnostic problem to different ophthalmologists. This is because the colobomata in this case are associated with oedema of the optic disc on both sides and a serious type of retinal detachment involving the macular region and extending on up to the disc margin on one side.

Case report

A girl, then aged 101/2 years, was first seen at the Eye Department of The Royal Infirmary, Edinburgh, on March 12, 1962, complaining of blurring of vision in the right eye of 3 weeks' duration after an attack of influenza. She had been seen on March 7, 1962, by her optician, who discovered visual acuities of 6/60 in the right eye and 6/9 in the left. He commented that when he had previously seen her on September 8, 1960, the visual acuity in both eyes was 6/9.

When examined on March 12, 1962, the visual acuities were 6/36 in the right eye and 6/6 in the left. Both optic discs showed a moderate degree of oedema. No other ocular abnormality was detected. The visual fields showed no abnormality. The patient was immediately referred for neurological examination. A thorough neurological and general physical investigation revealed no abnormality.

On follow-up, the right visual acuity after 5 days improved to 6/18, after 4 weeks to 6/12, and after 3 months to the normal. The patient experienced no other physical symptoms, apart from intermittent headaches. The appearance of the fundi did not alter until the beginning of January, 1964, when the vision in the left eye blurred.

On February 13, 1964, she was found to have visual acuities of 6/6 in the right eye and 6/36 in the left. The optic discs were unchanged and no other fundus lesion was recorded. A dense centro-caecal scotoma was detected in the left visual field. At this time it was considered that the defective vision in the left eye was most probably due to retrobulbar neuritis which might also previously have caused the defective vision in the right eye.

On follow-up, the left vision and the field defect did not improve. She was first seen by one of us (JFC) in August, 1964, when the above-mentioned clinical findings were confirmed and a diagnosis of bilateral drusen of the optic discs was suggested. The patient was also found to have bilateral enlargement of the blind spots. She was followed up every 6 months and the fundus was photographed at various visits, starting from February, 1964. When seen on December 22, 1966, she was found to have developed an inferior nasal field defect extending from 15° down to the periphery in the left eye, in addition to the centro-caecal scotoma (Fig. 1). This further loss of the left visual field was thought to be due to the progressive nature of the drusen, because the fundus revealed a changing pattern in the white lesions seen around the optic disc in that eye. These visual field
Defects in the left eye have not altered since then. Neither the eyes nor the optic discs have subsequently shown any change. The present visual acuities are 6/5, N5 in the right eye and 6/36, N8 in the left.

Recently the patient was further investigated by stereoscopic fundus photography and intravenous fluorescein fundus angiography. At the same time, all the previous fundus photographs since 1964 were compared and analysed to assess the fundus lesions. This revealed the following extremely interesting findings which had not been detected earlier.

**Left eye**

On comparing the colour fundus pictures from February, 1964, to October, 1970, it was discovered that in February, 1964, the patient had a serous retinal detachment in the macular region extending up to the optic disc (Fig. 2, overleaf). A rounded white choroidal defect was seen temporal to the optic disc. In addition, white deposits (the so-called “hard exudates”) were seen in the retina, mainly around the margin of the optic disc.

By August, 1964, the white deposits were more extensive and were encircling the optic disc on the nasal side, above, and supero-temporally, with a few deposits in the infero-temporal part of the retina (Fig. 3, overleaf). In December, 1965, the white deposits were thinning, though these were still evident all around the optic disc and, by September, 1967, most of them had cleared (Fig. 4, overleaf). In January, 1969, the deposits had nearly all gone apart from a few tiny white spots.

The choroidal defect and the peripapillary and macular retinal detachment showed no significant change at any time during the period of observation.

When the patient was seen in October, 1970, stereoscopic fundus photography (Fig. 5, overleaf) showed the presence of:

(a) Serous retinal detachment in the area from the temporal margin of the optic disc to the macular region and in the peripapillary region.

(b) The hole in the peripapillary choroid.

(c) Swelling of the optic disc.

Apart from that there was no significant change when compared with the pictures of January, 1969.
**FIGS 2 to 5** Left fundus showing (A) peripapillary coloboma, (B) serous retinal detachment (outlined by arrows), (C) white deposits, and oedema of the disc. Fig. 2 on 13 February, 1964. Fig. 3 on 18 August 1964. Fig. 4 on 13 April, 1967. Fig. 5, (opposite) on 7 October, 1970.

**Fluorescence fundus angiography**

During the transit of the dye, no fluorescence in the area of choroidal defect was seen, but some thinning of the peripapillary part of the pigment epithelium was present and was more marked than that on the right eye (Fig. 6a, opposite). The small white deposits, as expected, showed no abnormality on angiography. During the late phases (Fig. 6b), fluorescein diffused from the choroidal defect into the surrounding detached area, producing an abnormal fluorescence in the area of the choroidal defect, a part of the detached area of the retina temporal to the choroidal hole, the peripapillary region, and the optic disc. The peripapillary retinal detachment was least extensive in the superonasal region.
FIG. 6 Fluorescence fundus angiograms of left fundus on October 7, 1970, (a) during arterial phase; (b) during late phase (15 minutes after (a))
Right eye

On comparing the fundus pictures of this eye from February, 1964, to October, 1970, we found no significant change in the optic disc and the lesions around it. The stereoscopic pictures taken on October 7, 1970, showed a serous retinal detachment around the white circular choroidal defect, extending on to and around the optic disc (Fig. 7). The disc was swollen but less so than the left disc. Two small hyaline drusen were seen on the optic disc, one supero-nasally and the other infero-nasally.

**FIG. 7** Right fundus on October 7, 1971, showing (A) peripapillary coloboma, (B) serous retinal detachment (outlined by arrows), (C) oedema of the optic disc

*Fluorescence fundus angiography*

During the transit of the dye, abnormal fluorescence of the peripapillary region was seen, indicating a thinning of the pigment epithelium, with no early fluorescence in the area of the choroidal defect. The latter observation in both eyes confirmed that the rounded whitish spot in the peripapillary region was really a defect in the choroid and not a transparent drusen. Small white spots seen in the peripapillary region represented thinning of the pigment epithelium and again were not true drusen. During the late phases, the choroidal defect showed a marked fluorescence and the staining extended to the adjacent detached area of the retina and the peripapillary and disc regions (Fig. 8, opposite).

The holes in the peripapillary choroid on both sides were lying obliquely, with their mouths directed medially towards the optic discs. The temporal margin in both the cases was adherent to the overlying retina like a tent.

**Discussion**

This patient is interesting because she had presented a series of diagnostic problems since coming under observation 10 years ago.

1. The optic discs on both sides were found to be oedematous at her first visit to the hospital and it was thought that this was due to some intracranial cause. However, complete neurological investigations showed no abnormality. She has remained perfectly fit and healthy, in spite of having bilateral oedema of the optic disc all these years.

2. The presence of a centro-caecal defect in the left eye was at first thought to be due to a retrobulbar neuritis and, later on, to the drusen in the optic disc, although in reality the cause was an undiagnosed secondary serous retinal detachment involving the macular region and the area between the optic disc and the macula.
The white, rounded peripapillary spots on both sides were considered to be large drusen of the discs, responsible for a pseudopapilloedema on both sides. In reality, the white spots were peripapillary choroidal holes, as is demonstrated very well by stereoscopic fundus photography and fluorescence fundus angiography.

Both the optic discs, which were considered to have pseudopapilloedema initially, have been found by fluorescence angiography actually to be oedematous.

The white deposits (the so-called "hard exudates") in the peripapillary region on the left side were confused with multiple small drusen, and their normally changing pattern over the years was considered to be an unusual change undergone by the drusen.

The peripapillary choroidal hole in the left eye has resulted in secondary serous retinal detachment of the area, extending from the optic disc to the macula, which is responsible for the centro-caecal field defect. The cause of the sectoral infero-nasal field defect in that eye is still obscure and may be due to an interference with the blood supply to the optic disc in that region, as a result of the prolonged oedema of the optic disc. There is no evidence of the involvement of the nerve fibre layer of the retina to account for this defect.

The presence of a serous retinal detachment involving the macular region, similar to the one seen in the left eye of the present case, is a well-known occurrence in about a third of the reported cases with pits (holes) in the optic disc (Kranenburg, 1960; Sugar, 1962, 1964, 1967; Ferry, 1963; Gass, 1969; Gordon and Chatfield, 1969; and many others). The mechanism of production of the serous retinal detachment in the two types does not seem to be identical. Fluorescence fundus angiography in the present case with peripapillary choroidal holes has shown a profuse leakage of fluorescein from the choroid at the margins of the holes, which extends in both eyes into the subretinal fluid adjacent to the hole and in the left eye to that in the macular region. This indicates that the subretinal fluid is derived from the choroid around the hole. Similar fluorescence angiographic studies have been performed by one of us (SSH) in four patients with pits (holes) in the optic disc.

In these cases, the pit in the optic disc was associated in all cases with a crescent along the margin of the disc in the same sector as the pit which is also seen in the pictures of
Sugar (1967), Gass (1969), and Gordon and Chatfield (1969). On fluorescence fundus angiography, the crescent showed fluorescence during the very early phase of the transit of the dye, at which time the pit in the disc was non-fluorescent. During the late phases, however, there was fluorescence around the pit (Fig. 9). This has also been the experience of other authors (Gass, 1969; Gordon and Chatfield, 1969). The fluorescein did not leak from either of these sites into the subretinal fluid in the area of retinal detachment in the 15 to 30 minutes of the injection of the dye.

These angiographic findings indicate:

1. In the area of the crescent the only abnormality seen is the absence of the pigment epithelium and it is not a congenital crescent (vide infra).

2. It is highly unlikely that the subretinal fluid is derived from either the crescent or the pit, unless one were to consider the slight possibility that the leak of the fluid from these sites is so slow that it is not apparent on fluorescence angiography.

The source of the subretinal fluid in the central serous retinal detachment associated with pits in the optic disc has excited considerable controversy. Kranenburg (1960) believed that the temporally located pits are associated with anomalous papillomacular nerve fibre bundles which result in a "congenital locus minoris resistentiae" and are responsible for the detachment. Zimmerman (cited by Ferry, 1963) considered traction upon the macula to be responsible for the macular detachment. Sugar (1967) postulated a mechanical leak of fluid from the pit into the subretinal space because he had noticed in one particular case a channel between the pit and the elevated macula (Sugar, 1964). Babel (1967) and Farpour and Babel (1968) described the presence of vascular abnormalities around the pit on fluorescence angiography and believed that to be responsible for the macular lesion. Regenbogen, Stein, and Lazar (1964) speculated the central serous retinal detachment in these cases to be most probably due to subretinal-subarachnoid communication and the subretinal fluid being the cerebrospinal fluid. Gass (1969), on histopathological and fluorescence angiographic studies, also considered this as a possibility. This is because histopathological studies in the optic disc pit have revealed that the pit is formed by rudimentary retinal tissue associated with irregular glial elements and remnants of nerve fibres and pigment epithelium; these usually dip down into the intermediary tissue near the point of junction of the lamina cribrosa with the scleral promon-
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... and extend downwards towards the vaginal sheath, with defective lamina cribrosa in the region of the pit (Coats, 1908; Lauber, 1909; von Szily, 1913; Seefelder, 1915; Hagedoorn, 1928; Calhoun, 1930; Jaensch, 1931; Ferry, 1963; Badtke and Bauermann, 1964; Gass, 1969). This indicates that the pit in the optic disc and the hole in the peripapillary choroid seen in the present case are really not identical lesions. This is further confirmed by the dissimilarity of fluorescence angiographic findings between the two types of hole; hence the pathogenesis of the macular serous detachment in the two conditions does not seem to be identical. In the peripapillary choroidal holes in this patient it is due to the leakage of fluid from the choroid via the hole into the subretinal space.

The white deposits (the so-called "hard exudates") seen in the left eye of this patient represent degeneration of the retinal tissue secondary to the retinal detachment around the optic disc and macular region and, as expected, have regressed over the period from 1964 to 1970.

The oedema of both optic discs is due to seepage of the fluid from the area of the peripapillary choroidal hole into the loose prelaminar tissue and into the circumpapillary subretinal space, as shown on stereoscopic fundus and fluorescence fundus examinations. The seat of oedema in this case is thus almost identical to that observed in oedema of the disc due to other causes (Hayreh, 1969). Thus, both discs have a true oedema (not pseudopapilloedema), which has persisted unchanged since it was first noticed in 1962. Presumably the patient has had oedema of the discs all her life.

The bilateral identical peripapillary choroidal holes seen in this patient represent, in our view, atypical choroidal colobomata. In a coloboma of the choroid the edges are usually clear cut and frequently pigmented; the choroidal defect is directly associated with an absence of Bruch’s membrane and the overlying pigment epithelium of the retina (Duke-Elder, 1964). The overlying retina may be normal (Pause, 1878; Bach, 1898) although it is usually malformed.

The typical choroidal colobomata are situated in the region of the embryonic fissure, i.e. below and slightly medially, because these are due to disturbance in the closure mechanism of the fissure. The defect is usually bilateral (Møllenbach, 1947). The position of the embryonic fissure is not always absolutely constant (Mann, 1921) and is not specific or localized to any particular part of the vesicle (Beckwith, 1927). This so-called rare atypical colobomata might, in fact, be typical (Duke-Elder, 1964).

In order to understand the true nature of the choroidal holes seen in the present case, it is necessary to consider other similar congenital defects involving the optic disc, peripapillary region, and macular region.

The presence of a pit or hole in the optic disc is a well-known occurrence and its association with a serous retinal detachment of the macular region has already been discussed. The pit in the optic disc is regarded by most authors as a minimal coloboma in an atypical position. Kranenburg (1960) found the pit to be situated temporo-inferiorly in 46 per cent., mid-temporally in 26 per cent., supero-temporally in 4 per cent., mid-superiorly in 3 per cent., mid-nasally in 4 per cent., nasally in 3 per cent., infero-medially in 7 per cent., and centrally in 4 per cent. The high incidence of temporal location of the pit with no other evidence of coloboma of disc or choroid suggests the possibility of its being unrelated to closure of the embryonic fissure (Badtke and Bauermann, 1964). However, a case of Gass (1969) showed the pit of the disc with choroidal coloboma in the same eye and along the same axis.

The congenital crescent (conus) around the optic disc in the peripapillary region (unlike the acquired crescent, e.g. in myopia, etc.) has been considered to be due to
defective closure of the embryonic cleft (Liebreich, 1859; Schnabel, 1874; Fuchs, 1882). According to Mann (1957), it is produced primarily by a neural ectodermal defect through failure of the pigment epithelium to reach the site of implantation of the optic stalk as a result of aberrant differentiation of any of the various parts of the secondary optic vesicle at the 8 mm. stage. Since the choriocapillaris and the pigment epithelium are essentially interdependent in their development (Redslob, 1925), failure of the two layers together is to be expected. Microscopically in these cases there is an absence of the pigment epithelium, Bruch’s membrane, and nuclear layers of the retina and choroid; the floor is formed by the sclera with normal nerve fibres of the retina over it. Vossius (1885) found the congenital crescent to be situated below the optic disc in 67 per cent., nasally in 8·1 per cent., infero-nasally in 7·3 per cent., superiorly in 4·5 per cent., supra-temporally in 7·2 per cent., and infero-temporally in 5·4 per cent.

Macular colobomata are common and with rare coincidental developmental anomalies. These are frequently bilateral (de Wecker, 1872; Schmidt-Rimpler, 1880; Fuchs, 1882; Dor, 1888; Bock, 1893; Kastalsky, 1898; Kimpel, 1898; Funciuss, 1913; Lemke, 1913; Helmch, 1920; Schott, 1921; Bär, 1925; Shoji, 1926; Przybyslka, 1926; Clarke, 1927; Davenport, 1927; Pierce, 1932). It is considered that a vast majority of these are secondary to intrauterine inflammations; toxoplasma infection is perhaps the most common type and they may also be due to syphilis and tuberculosis. However, some may be due to an ectodermal developmental defect, since no evidence of inflammation has been seen on histopathology in some cases, while an hereditary tendency in some and the presence of coincident developmental aberrations in others suggests a genetic cause (van Duyse, 1898; Parsons and Coats, 1906; Seefelder, 1911).

Rarely, atypical colobomata have been described in all directions, e.g. colobomata extending from the optic disc to the macula (Nuel, 1885), superotemporally (Lang, 1886; von Hippel, 1901), and temporally (Roschowski, 1906).

The above discussion leads us to the suggestion that the peripapillary choroidal holes seen in this patient are most probably atypical minimal colobomata of the choroid, involving the choroid, Bruch’s membrane, and the pigment epithelium. In the retina the overlying nerve fibres are not involved. The situation of the holes temporal to the optic disc in both eyes is unlike that of typical colobomata, but the presence of the pit of the optic disc and the conus on the temporal side was noted, as mentioned above. This temporal location may be due to an ectodermal defect in this region in either (a) an atypically situated or accessory embryonic fissure (van Duyse, 1920; Caspar, 1925; Finlay, 1930; Badkhe, 1957–8), or (b) the pluripotential cells of the optic cup. In the latter event, the development of the pigment epithelium or its pigment may be inhibited and with it the development of the choriocapillaris; or a glial overgrowth may destroy the pigment epithelium and produce aplasia of the choroid (van Duyse, 1920); or there may be an imbalance of growth between the two layers of the optic cup associated with faulty differentiation of the tissues (Klien, 1959) or metaplasia. Treacher Collins (1927) thought that atypical coloboma was the result of a failure in the development of a part of the chorio-capillaris with consequent aplasia of the choroid and defect in the pigment epithelium.

It may be argued that the choroidal holes seen in the present case could be the result of an intrauterine inflammatory process similar to that seen in macular colobomata. The presence of normal nerve fibres over the holes would tend to contradict such a hypothesis.
In conclusion, we feel that in this case these peripapillary choroidal holes are a form of minimal atypical choroidal colobomata. Their association with chronic oedema of the optic discs and serous retinal detachment of the adjacent areas extending to the macular regions may constitute a syndrome which has not been described before.

Summary

A case of bilateral atypical colobomata of the peripapillary choroid, associated with oedema of the optic discs and serous retinal detachment around the colobomata, extending in one eye to involve the macular region, is presented. The various signs led to erroneous diagnoses at different stages, e.g. papilloedema of intracranial origin, retrobulbar neuritis, pseudopapilloedema, and drusen of the optic disc. The pathogenesis of the lesions is discussed and compared with that of the optic disc pit (hole).

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