The perifoveal vasculature in albinism

Z. GREGOR

From Moorfields Eye Hospital and Institute of Ophthalmology, London

SUMMARY The perifoveal vasculature was studied in a series of patients with oculocutaneous tyrosinase-positive albinism. Fluorescein angiographic studies show a normal distribution of the major retinal blood vessels and in some cases of the capillaries in the macular area of these patients.

Albinism is a congenital heritable hypomelanosis that is limited to the eye (ocular albinism) or involves the eye and integument (oculo-cutaneous albinism) and in which the basic defect is a partial or total reduction of melanin deposition on melanosomes (Fitzpatrick and Quevedo, 1972).

Oculocutaneous albinism has been divided into 2 forms on the basis of the ability of melanocytes in hair bulbs to synthesise melanin when incubated in solutions containing tyrosine (Witkop *et al.*, 1961). These 2 forms, tyrosinase-negative and tyrosinase-positive oculocutaneous albinism, are each inherited as autosomal recessive traits resulting from the presence of 2 non-allelic genes (Witkop *et al.*, 1970).

The ocular manifestations of albinism have been extensively studied since the early description by Nettleship (1876), but there have been few reports of the histological features of these eyes. Fritsch (1906) reported on the fovea of a Negro albino as having an 'unusual structure', while Elschnig (1913) described 'the complete absence of the fovea' as the most important histological finding in the retina of the young albino, a finding confirmed by others (Nettleship, 1906; Velhagen, 1917). Usher (1920) concluded that the chief cause of defective vision and nystagmus in the albino was the imperfectly developed or absent fovea, and Gilbert (1921) showed that the appearance of the foveal cones was similar to that of cones in the periphery of the fundus. O'Donnell *et al.* (1976) found a lack of foveal differentiation in eyes from a patient with oculocutaneous albinism. Although they could not detect a fovea in an eye from an ocular albino, the maximal thickness of 8 rows of ganglion cells in the posterior retina suggests an element of retinal specialisation.

The normal fovea is surrounded by a perifoveal vascular arcade arising equally from the superior

and inferior macular branches. The central area has been described as avascular (Duke-Elder, 1961), although capillaries of small calibre but of considerable length may be demonstrated there by fluorescein angiography (Lowenstein, 1942; Yeoung *et al.*, 1973; Bird and Weale, 1974). The difference between the macular and non-macular retinal circulation may be 'physiologically significant' (Henkind, 1975). Henkind studied the development of the avascular zone in experimental animals and suggested that it arises by capillary closure after birth, is independent of vision, and that the vessels occasionally seen traversing this zone are due to persistence of fetal vasculature.

In 3 out of 6 young people with 'total albinism' Ichikawa (1913) followed the macular vessels to the fovea, where they appeared 'to cross each other in a

Fig. 1 Patient 3. Fluorescein angiogram of the left eye showing normal distribution of major retinal vessels. Visual acuity was 6/24. Moderate mottling of pigment epithelium

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disorderly fashion'. However, Falls (1951) examined groups of patients with X-linked ocular albinism and carriers of this condition and found no abnormality of the major retinal vessels.

It is the purpose of this study to demonstrate the vascular architecture in the macular region of patients with oculocutaneous albinism and to compare it with that in normal patients.

Patients and methods

Seven patients with tyrosinase-positive oculocutaneous albinism were examined. Their ages ranged from 11 to 33. Four were female and 3 were male. Five were Caucasian and 2 were Negroes. Their visual acuity ranged from 6/12 to 6/60 and all had moderate to severe nystagmus.

Full ophthalmological examination was performed, and this included indirect binocular ophthalmoscopy, slit-lamp biomicroscopy with Hruby lens, colour stereophotography, and fluorescein angiography; 5 ml of 20% sodium fluorescein was injected into the antecubital vein, and photographs were taken at 1-second intervals with a Zeiss (Oberkochen) Fundus camera on Ilford FP4 film during the initial transit of dye through the retinal blood vessels. Ilford ID 11 solution was used for developing and a contrast-reducing mask for printing.

Fig. 2 Patient 1. Fluorescein angiogram of the right eye showing normal distribution of perifoveal capillaries. Visual acuity was 6/24. Minimal mottling of pigment epithelium

Results

The retinal pigment epithelium screened the underlying choroid to a variable degree. The screening appeared normal in 3 patients, imperfect in 2, and was virtually non-existent in the remaining 2 patients. The major retinal vessels appeared normal in all patients.

Fluorescein angiography was performed in all 7 cases. The retinal pigment epithelium appeared abnormal in all the patients. This abnormality ranged from scattered small transmission defects in 4 patients to larger patches of background fluorescence in 1 patient. In the remaining 2 patients there was so little pigment present in the pigment epithe-

Fig. 3 Patient 4. Fluorescein angiogram of the right eye showing the arrangement of the perifoveal capillaries. Small vessels are crossing the macular area forming direct anastomoses with superior and inferior vessels. Visual acuity was 6/18. Moderate mottling of pigment epithelium. (A) Direct print from the negative. (B) Positive print made with contrast-reducing mask

Table 1

Patient	Sex	Age	Corrected visual acuity		Biomicroscopic findings		Angiographic findings		
			Right	Left	Distribution of perimacular vessels	Macular reflex	Pigment epithelium	Capillary arcade	Vessel wall
1	M	18	6/24	6/24	Normal	Absent	Minimal mottling	Absent	Normal
2	F	27	6/36	6/18	Normal	Absent	Minimal mottling	Impossible to assess due to nystagmus	
3	F	34	6/24	6/9-3	Capillaries crossing foveal areas	Present	Moderate mottling	Present	Normal
4	M	37	6/18	6/24	Capillaries crossing foveal areas	Absent	Moderate mottling	Present	Normal
5	M	13	6/24	6/36	Normal	Absent	Moderate mottling centrally absent in the periphery	Present	Normal
6	F	20	6/36	6/36	Normal	Absent	Almost absent	Impossible to assess due to choroidal fluorescence	
7	F	18	6/36	6/36	Normal	Absent	Absent	Impossible to assess due to choroidal fluorescence	

limum that the retinal vessels were difficult to distinguish. The calibre of the retinal vessels was within normal limits, and there was no leakage or staining of the vessel wall.

In all 7 cases the major branches of the retinal vessels followed their normal course with respect to the macular area (Fig. 1). The perifoveal vasculature could be satisfactorily analysed in 4 patients. In 2 of these it appeared normal (Fig. 2). In the other 2 cases there were small vessels crossing the macular area and forming direct anastomoses with superior and inferior vessels (Fig. 3). The remaining 3 cases were difficult to examine owing to nystagmus in 1 patient and to the transmitted choroidal fluorescence in 2 patients (Table).

Comment

The results suggest that the macular area in a group of 7 patients with tyrosinase-positive oculocutaneous albinism does differ from the surrounding retina. In all these patients the major retinal vessels respected the macular region. This is not in agreement with Ichikawa's statement (Ichikawa, 1913). Having examined Ichikawa's original fundal drawings, our impression was that even these are within normal limits.

The findings that in some of our patients the central area was crossed by capillaries is unusual, though it has been described in people with normal pigmentation and normal vision (Bird and Weale, 1974).

The size of the central avascular area is variable

(Eisler, 1930). Lowenstein (1942) and Yeoung (1973) demonstrated a capillary-free area which measured 0.165 to 0.07 mm respectively. The variability in size does not appear to prejudice foveal function (Yeoung, 1973). Bird and Weale (1974) showed that capillaries can cross the central foveal area in patients with normal visual acuity, and similar findings were obtained by Shikano and Shimizu (1968).

In conclusion, we suggest that there is a degree of retinal specialisation in the macular area of the patients examined by us. This may have important implications for the future treatment of patients with albinism.

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