Ocular findings in sickle cell-haemoglobin O Arab disease

PATRICK I. CONDON AND GRAHAM R. SERJEANT

From the Medical Research Council Laboratories, University of the West Indies, Kingston, Jamaica

SUMMARY The ocular findings in 2 patients with sickle cell-haemoglobin O Arab disease are described. One patient had proliferative sickle cell retinopathy with extensive autoinfarction of lesions. Sickle cell-haemoglobin O Arab disease must be added to the list of conditions that may be associated with proliferative sickle cell retinopathy.

Sickle cell-haemoglobin O Arab (SO Arab) disease results from the inheritance of 2 abnormal β chains, and is a rare condition resembling sickle cell-haemoglobin C (SC) disease on routine haemoglobin electrophoresis at alkaline pH. Previous observations suggested that the clinical and haematological severity was intermediate between that of SC disease and homozygous sickle cell (SS) disease (Milner *et al.*, 1970). In view of the high prevalence of proliferative sickle cell retinopathy in SC disease it was of interest to report the ocular lesions occurring in 2 patients with SO Arab disease.

Patients and methods

Approximately 2000 patients with different genotypes of sickle cell disease attend the sickle cell clinics at the University Hospital of the West Indies. Of these, 4 patients have been identified as SO Arab disease (Milner *et al.*, 1970), and the present 2 patients represent cases 3 and 4 in that report. The diagnosis of SO Arab disease was based on criteria described by Milner *et al.* (1970). Mean steady state haematological findings in the 2 patients are summarised in Table 1.

Correspondence to Dr G. R. Serjeant, MRC Laboratories (Jamaica), University of the West Indies, Mona, Kingston 7, Jamaica WI.

CASE 1

A 15-year-old black female had been followed up in the Sickle Cell Clinic for 3 years with a history of intermittent joint pains and chronic leg ulceration. She had no eye complaints, and examination showed a visual acuity of 6/6 in both eyes. Abnormalities of conjunctival vessels typical of sickle cell disease (Paton, 1961) were present, the ocular media were clear, and the optic discs and maculae normal. Peripheal venous tortuosity and arteriolar narrowing with some peripheral whiteness without pressure was present in localised quadrants (Fig. 1). A brown mottling of the peripheral retina was noted in the inferotemporal left quadrant. There was no evidence of proliferative sickle retinopathy. The patient subsequently emigrated and was lost to follow-up.

CASE 2

A 52-year-old black female has been followed up in the clinic for 10 years with a history of intermittent joint pains, leg ulceration, and gallstones. There was a history of sudden visual loss in the right eye years ago with subsequent improvement of acuity, which did not return to normal, and then vision gradually deteriorated. On examination in April 1973 visual acuity was 3/60 in the right eye and 6/12 in the left eye. Abnormalities of conjunctival vessels were present, the ocular media were clear,

Table 1 Steady-state haematological findings in the 2 patients

Patient no.	Hb (g/dl)	PCV (g/dl)	MCHC (g/dl)	RBC ($\times 10^{12}/l$)	MCV (fl)	MCH (pg)	Retics (%)	ISC (%)	Total bilirubin (mg/dl)	Serum Fe (μ g/dl)
1	8.8	0.26	34	3.12	82	28	2.0	4.2	1.2	92
2	8.4	0.27	31	3.46	76	24	6.6	3.0	1.0	74

Fig. 1 Retinal drawing of findings in patient 1. (a) Right eye—peripheral whitening between 1 o'clock and 5 o'clock (depicted by arrow heads). (b) Left eye—peripheral whitening between 8 o'clock and 11 o'clock and between 1 o'clock and 3 o'clock. Shaded area—Pigmented chorioretinal lesion

Fig. 2 Retinal drawing of findings in patient 2. (a) Right eye. (b) Left eye. Shaded areas indicate pigmented chorioretinal lesions. Dotted lines indicate occluded arterioles. Arrows depict occluded proliferative retinopathy and arrows with L indicate leaking proliferative retinopathy. Macular drusen

Fig. 3 Patient 2. Fluorescein angiogram of left superonasal periphery with leaking proliferative sickle cell retinopathy lesion

and both fundi were easily seen. Both optic discs showed physiological cupping. A mottled pigment epithelial change affected both maculae, the right more than the left, but there were no signs of macular oedema. Occluded retinal arterioles occupied the complete circumference of both eyes and extended posteriorly to the equator in places. Occluded proliferative retinopathy was noted in the superotemporal and inferonasal peripheral retina of the right eye (Fig. 2a) and in the superotemporal region of the left eye (Fig. 2b). Three

leaking proliferative lesions were seen in the nasal region of the left eye. Multiple areas of pigmentary disturbance (sunburst spots) occurred in both eyes and a posterior vitreous detachment in the right eye. Fluorescein angiography confirmed leaking proliferative sickle cell retinopathy lesions at the junction of vascular and ischaemic retina (Fig. 3).

Re-examination of the patient in February 1977 and July 1978 showed little change. The vision had deteriorated to counting fingers in the right eye and to 6/18 in the left, the macular pigment epithelial changes having progressed. Black sunburst spots increased in number in both eyes. The leakage from proliferative lesions in the left eye had diminished by autoinfarction to about 50% of that observed in 1973.

Discussion

The retinal vascular changes that occur in other forms of sickle cell disease may also be found in SO Arab disease. The first patient showed mild changes of peripheral venous tortuosity and arteriolar narrowing comparable to the findings in many patients with SS disease at this age. In the second patient moderately severe proliferative sickle cell retinopathy was undergoing autoinfarction. The gradual deterioration of visual acuity in this patient may have been caused by the progressive macular changes, and the history of sudden but transient visual loss may have represented a vitreous haemorrhage.

Although uncommon, sickle cell-haemoglobin O Arab disease must be added to the list of sickle cell conditions associated with proliferative retinopathy and possible visual impairment.

References

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