as an association of blepharophimosis syndrome in the literature. The patient's marfanoid habitus was not associated with any other systemic or ocular features of Marfan's syndrome. The other physical findings such as low-set ears and a broad nasal bridge are well-known associations of blepharophimosis syndrome.

In our case, incomplete coloboma was evident in the left eye only. The features described in the right eye could possibly be explained as a forme fruste of an iris coloboma. However, we could not find any such description in the literature. Baraitser and Winter[6] reported a new syndrome of three cases, two of them siblings, with a combination of short stature, mental retardation, bilateral iris coloboma, ptosis, hypertelorism and a broad nasal bridge. Our case differed in having normal mental status, arachnodactyly and unilateral iris coloboma and bilateral congenital ectropion uveae. This report bore the closest resemblance to our patient.

The amblyopia observed in the left eye could possibly be explained by the hypermetropic refractive error and the asymmetrical ptosis with the compromise of the pupillary axis. That these factors predispose to the development of amblyopia in blepharophimosis syndrome was highlighted by Beaconsfield *et al.*[7] in their large series of 101 cases of the syndrome.

This atypical case of blepharophimosis syndrome with arachnodactyly and uveal abnormalities highlights one more dimension to the many associated ocular and systemic developmental anomalies which have been described in conjunction with blepharophimosis syndrome.

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Sir,

## Vitelliform Macular Dystrophy: A Cause of Macular Holes with Retinal Detachments

Vitelliform macular dystrophy, also known as Best's disease, is one of the heredo-familial macular disorders.[1] The mode of inheritance is probably autosomal dominant with irregular penetrance.[2] One of the remarkable clinical features of this disorder is the surprisingly good visual acuity, which does not correlate with the ophthalmoscopic findings. The classic ophthalmoscopic picture in vitelliform macular dystrophy is a yellow, round, slightly elevated structure resembling an egg yolk, usually found accidentally during routine fundus examination of children between 3 and 5 years old without any visual symptoms. This typical feature may not be encountered but instead other evolutionary stages may be found in which the characteristic structure either undergoes spontaneous resorption, forming the 'scrambled egg' stage with occasional cyst formation and a fluid level resembling a hypopyon giving rise to a so-called pseudohypopyon stage. Finally, the atrophic stage ensues with widespread retinal pigment epithelial (RPE) atrophy. The clinical diagnostic test in this disorder is a subnormal electrooculogram (EOG) with a light/dark ratio rarely exceeding 1.5, whereas the electroretinogram (ERG) is invariably completely normal.[2] Vision is commonly affected late in the disease unless a complication such as haemorrhage in the vitelliform structure or subretinal neovascularisation occurs.[2,3] Recent histopathological studies report flattened RPE cells with displacement of the nuclei towards the apex and diffuse deposition of abnormal lipofuscin granules.[4]

This report documents an unusual clinical presentation of vitelliform macular dystrophy: macular holes with retinal detachment.

### *Case Reports*

*Case 1.* A 35-year-old man presented with a complaint of recent onset of metamorphopsia in the right eye. He had had defective vision in his left eye 7 years earlier with no specific medical history and no history of trauma to that eye. Visual acuity was 6/12 in the right eye and light perception in the left with normal intraocular pressures and normal anterior segments on biomicroscopy. Examination of the right eye disclosed a macular lesion typical of the pseudohypopyon stage of vitelliform macular dystrophy with normal retinal periphery (Fig. 1A). The left eye showed a large atrophic macular hole with widespread RPE atrophy and long-standing total retinal detachment with retinal thinning and cysts (Fig. 1B). No other peripheral retinal breaks were present.

Electrophysiological studies confirmed the diagnosis of vitelliform macular dystrophy, the EOG showing a subnormal response in both eyes (1.39 in the right eye and 1.15 in the left eye) with a normal ERG tracing. Fluorescein angiography revealed bilateral widespread RPE atrophy as evidenced by the window defects throughout the phases of the angiogram (Fig. 2), in addition to a small leaking hyperfluorescent spot in the right eye denoting subretinal neovascularisation.

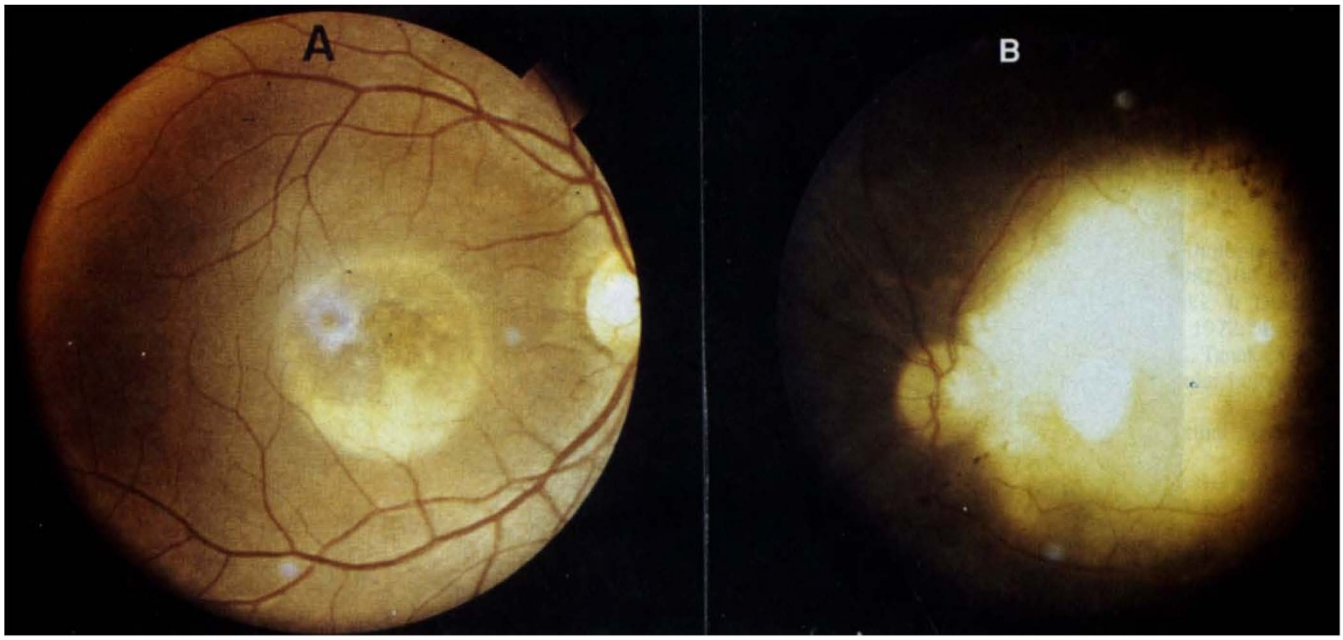


**Fig. 1.** *Case 1. (A) Right eye showing typical pseudohypopyon stage. (B) Left eye with a large macular hole and retinal detachment. There is widespread retinal pigment epithelial (RPE) atrophy involving the whole central area.*

In view of the poor state of the left retina, no attempt to repair the retinal detachment was made. The small subretinal neovascular membrane in the right eye was followed up for 4 months with no decline in vision.

*Case 2.* In November 1989 a 19-year-old woman was referred with a history of defective vision in both eyes dating from 2 years earlier and a diagnosis of bilateral idiopathic macular degeneration. Her general medical history was unremarkable. Visual acuity was 6/36 in the right eye and 6/60 in the left eye, with normal intraocular pressures and unremarkable anterior segments on biomicroscopy. Ophthalmoscopy of the right eye revealed a white scar with surrounding pigmentation in the macular area (Fig. 3A). The left eye showed a large macular hole with a yellowish colour in the adjacent area which was flat with no evidence of subretinal fluid (Fig. 3B). The posterior hyaloid membrane was attached. Electrophysiological studies confirmed the diagnosis of vitelliform macular dystrophy. The EOG measured 1.41 in the right eye and 1.25 in the left. The ERG was normal. The patient was discharged from follow-up.

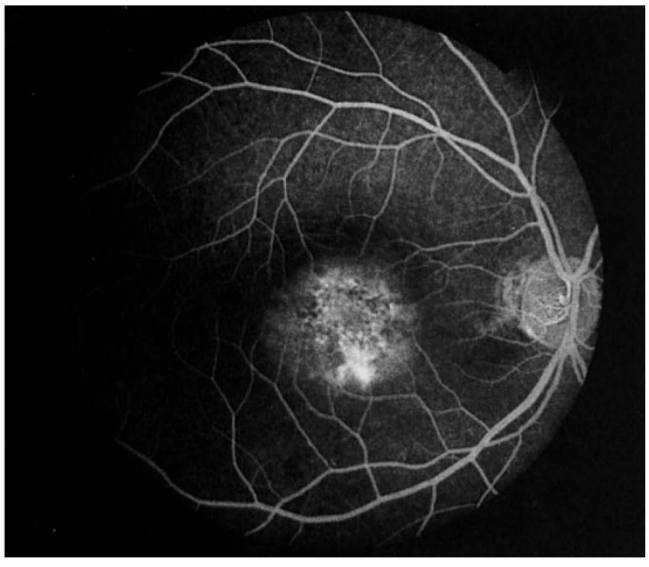

**Fig. 2.** *Case 1. Fluorescein angiogram of the right eye showing central RPE atrophy with an area of leakage inferiorly caused by a subretinal neovascular membrane.*

In December 1991, the patient presented with recent deterioration of vision in her left eye. Visual acuity remained 6/36 in the right eye but had dropped to hand movements in the left. The fundus appearance of her right eye was virtually unchanged (Fig. 3C), but the left eye showed a central retinal detachment extending beyond the temporal vascular arcades (Fig. 3D).

A pars plana vitrectomy with endodrainage and air–fluid exchange was planned but the patient declined surgery and defaulted.

*Case 3.* A 15-year-old boy was referred with the diagnosis of left retinal detachment due to a macular hole. The visual disturbances had started 6 months earlier. Visual acuity was 6/9 in the right eye and hand movements in the left eye with normal intraocular pressures and unremarkable anterior segments. Fundus examination of the right eye disclosed a typical yellow egg-yolk lesion characteristic of vitelliform macular dystrophy. The left eye showed a large macular hole with surrounding RPE atrophy and a total retinal detachment and no peripheral breaks. The posterior hyaloid membrane was intact. Electrophysiological tests showed the characteristic features of the disorder.

A pars plana vitrectomy with endodrainage and air–fluid exchange was carried out with face-down positioning of the patient post-operatively. This initially flattened the retina but subretinal fluid began to accumulate on ambulation. Positioning the patient face-down for

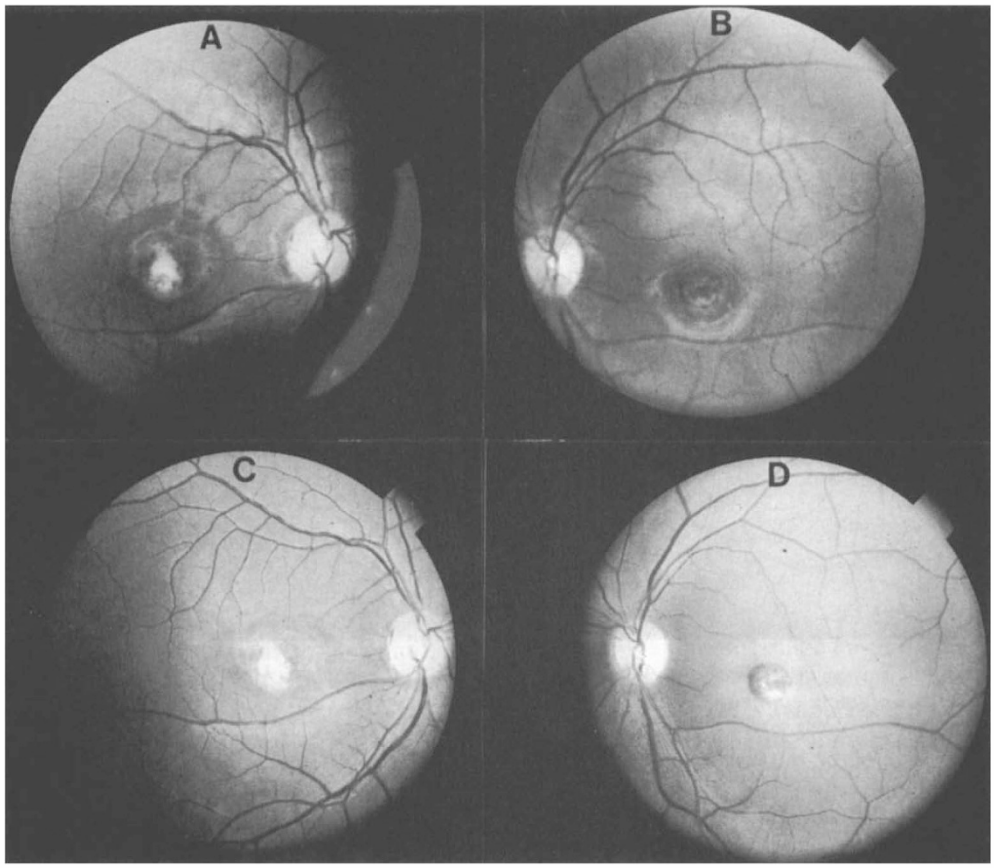


**Fig. 3.** *Case 2. (A) Right eye at first presentation with central white scar and surrounding pigmentation. (B) Left eye at presentation showing a macular hole. The surrounding retina is attached. (C) Right eye 1 year later showing no change in appearance. (D) Left eye with central detachment 1 year later.*

a few hours resulted in complete absorption of the subretinal fluid. Laser photocoagulation was then immediately applied to the margin of the macular hole with proper positioning of the patient to achieve internal tamponade of the retina. The retina remained attached for 6 months and vision improved to 6/60.

## *Discussion*

Full-thickness macular holes complicated with retinal detachment have been reported in association with degenerative myopia,[5,6] following blunt trauma[7] and sometimes complicating retinal vascular diseases. Macular hole and retinal detachment is an extremely rare presentation in vitelliform macular dystrophy. A single similar case was reported in the ophthalmic literature in 1985 by Schachat *et al.*[8] This report describes and documents 3 cases with this rare presentation. All 3 cases demonstrated a rather typical lesion in the other eye and electrophysiological tests confirmed the clinical diagnosis of vitelliform macular dystrophy.

The pathogenesis of macular holes in these cases gives rise to many speculations. Rupture of the cyst in the vitelliform stage may be a probable mechanism. On the other hand, in the end-stage form, the retina becomes atrophic with widespread RPE atrophy – a condition simulating the lesions of degenerative myopia. Some investigators have suggested that a lowered function of the RPE decreases the pumping effect of these cells and impairs the dehydrating mechanism of the subretinal space, finally weakening the adherence of the sensory retina to the retinal pigment epitheleium.[9] Thus in the presence of widespread RPE dysfunction the chances of a macular hole progressing to retinal detachment is definitely increased, as has been the experience with highly myopic eyes. This is also the case in vitelliform macular dystrophy, where there is widespread RPE dysfunction as evidenced by the subnormal EOG. Moreover, the macular area usually shows significant RPE atrophy as demonstrated by fluorescein angiographic studies.[2] Although macular holes in emmetropic eyes are not known to develop retinal detachment,[6] all 3 cases in this report were emmetropic and nevertheless progressed to retinal detachment in a very similar fashion to highly myopic eyes with macular holes. This comfirms that RPE dysfunction probably plays a key role in the development of retinal detachment in macular holes.

Interestingly, case 1 presented with a subretinal neovascular membrane in the other eye, another rare feature of this disorder.[3]

The management of case 3 resembles that described by Schachat *et al.*[7] However, in cases with widespread central RPE atrophy similar to case 1, a pars plana vitrectomy

with a permanent internal tamponade such as silicone oil may be required.

Vitelliform macular dystophy should be considered in the differential diagnosis of causes of macular holes, particularly in young non-myopes. Macular holes in these cases resemble those in myopic eyes and may develop retinal detachment. The key to the diagnosis is the fundus appearance of the fellow eye and the characteristic results of electrophysiological tests. These cases may not respond to simple vitrectomy and air–fluid exchange due to the widespread atrophy of the underlying RPE, and a more aggressive approach may be required.

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