Vitelliform macular dystrophy of late onset

ROBERT A. WIZNIA,¹ BARBARA PERINA,¹ AND KENNETH G. NOBLE²

From the ¹Retina Service, Section of Ophthalmology, Department of Surgery, Hospital of St Raphael, New Haven, Connecticut, and the Department of Ophthalmology and Visual Science, Yale University School of Medicine, New Haven, Connecticut; and the

²Department of Ophthalmology, New York University School of Medicine, New York, New York, USA

SUMMARY A family with vitelliform macular dystrophy is presented in which the proband became symptomatic at age 51 rather than during the more typical first or second decade. Elderly patients with vitelliform macular dystrophy may have clinical findings resembling age-related degenerative choroidopathy of the macula.

Vitelliform macular dystrophy or Best's disease is a dominantly inherited fundus dystrophy with macular findings demonstrating irregular penetrance and variable expressivity. It is often difficult to establish the diagnosis from clinical examination alone, since the disease has a wide range of expressivity with polymorphous appearance of the fundus.¹ A severe depression of the light peak/dark trough ratio of the electro-oculogram (EOG) is the most constant feature of the disease² and may be found even when the macula has a normal appearance.

The typical age of onset is in the first and second decade with the chief complaint being a decrease in visual acuity. Since Best's vitelliform dystrophy typically has its onset in childhood and adolescence, reports of patients first presenting with symptoms in their fifth decade and older are infrequent. These have been limited to the affected individual with no discussion of other family members^{3,4} or to patients who were studied after younger family members had been detected with the condition.^{2,5}

A family with vitelliform macular dystrophy is presented in which the proband became symptomatic at age 51 and was found to have a unilateral typical 'egg yolk' macular lesion.

Patients and methods

The proband presented with a history of difficulty in reading small print. The results of his examination prompted evaluation of all available family members—a total of 7 patients (Fig. 1). The patient's father was deceased, but had no history of visual difficulties. One sister, age 56, refused examination.

All patients underwent a complete ophthalmological examination including Amsler grid testing, Goldmann visual field testing, colour vision testing with the American Optical-HRR and City University tests, and slit-lamp biomicroscopy with Goldmann

Fig. 1 Genetic diagram detailing the inheritance pattern in the reported family. Case numbers are shown.

Vitelliform macular dystrophy of late onset

contact lens. Fundus photography and fluorescein angiography were performed with the Zeiss fundus camera in all cases with macular pathology. All patients had electro-oculogram testing with the LKC systems dual channel electro-oculograph; all patients had their pupils fully dilated with 1 drop of tropicamide 1% and 1 drop of phenylephrine 10%. After pretesting bright light adaptation for 5 minutes, testing was performed at one-minute intervals for 20 minutes in the dark and then at one-minute intervals for 15 minutes with the Ganzfeld background light and room light both on. Light peak-dark trough ratios were then calculated. Normal readings in this laboratory for patients of 40 years of age or less are 2.00 or higher and for patients older than age 40 are 1.90 or higher. Definitely abnormal readings are 1.80 or less for both groups.

Case reports

Case 1. The proband first presented with symptoms in April 1979 at age 51. This white man reported difficulty with reading small print, particularly with the right eye for several months. Reports of previous examinations by other ophthalmologists in 1968 and 1972 were unremarkable. In 1975 his visual acuity was reported to be 6/6 OU, although a small, flattened retinal pigment epithelial detachment was present superior to the right macula. No vitelliform lesion was noted. The left fundus was within normal limits.

When first examined by us in April 1979 best corrected visual acuity was RE 6/15+2 and LE 6/7.5. Colour vision testing was normal in both eyes. Amsler grid testing of the right eye demonstrated metamorphopsia centrally within 3° of fixation. No abnormality was noted with the left eye. A round yellow lesion just smaller than one disc diameter was present at the level of the retinal pigment epithelium in the right macula (Fig. 2). The left fundus was unremarkable. Characteristic blockage of choroidal fluorescence by the vitelliform disc in the right eye was noted on fluorescein angiography (Fig. 3). The LP/DT ratio of the electro-oculogram (EOG) was RE 1.50 and LE 1.57.

These findings were felt to be compatible with Best's vitelliform macular dystrophy and prompted examination of other family members.

Case 2. The proband's mother, 79 years old, white, was asymptomatic. Her best corrected visual acuity was RE 6/9 and LE 6/15. Funduscopy examination of both eyes revealed retinal pigment epithelial rarefaction and atrophy and drusen in the macula and immediately surrounding region (Fig. 4). EOG studies demonstrated the LP/DT ratio to be RE 1.12 and LE 1.11.

The clinical findings point to the marked variability

Fig. 2 A round yellowish one disc diameter lesion is noted in the right macula of case 1.

in the fundus picture of the vitelliform macular dystrophy, particularly in elderly patients, where the evolution of egg yolk lesions to an atrophic picture may occur.

Case 3. A 47-year-old white female, a sister of the proband, had visual acuity of 6/9 in both eyes. Funduscopy examination was normal in both eyes, but the EOG testing demonstrated subnormal LP/DT ratios, RE 1.25 and LE 1.14.

This case demonstrates that an abnormal EOG can

Fig. 3 There is characteristic blockage of choroidal fluorescence by the vitelliform disc in the right macula of case 1.

4a

4b

Fig. 4 a: Right eye. b: Left eye. The fundus of both eyes of case 2 resembles findings typically noted in age-related degenerative choroidopathy.

be a genetic marker for vitelliform macular dystrophy in the absence of clinical findings.

Case 4. A 53-year-old white male, a brother of the proband, was asymptomatic. Visual acuity was 6/7.5 in both eyes. Funduscopy examination and EOG testing (RE 2.42 and LE 2.26) were within normal limits.

Case 5. A 46-year-old white female, a sister of the

proband, was asymptomatic. Visual acuity was 6/6 in both eyes. A normal fundus examination was noted. EOG testing demonstrated LP/DT ratios of RE 2.40 and LE 2.50.

Case 6. A 20-year-old white female, the daughter of the proband, was also asymptomatic. Visual acuity was 6/6 in both eyes. Funduscopy examination was normal. LP/DT ratios on the EOG were RE 2.50 and LE 2.26.

Case 7. A 19-year-old white male, the son of the proband, was asymptomatic. Visual acuity was 6/6 in both eyes. Funduscopy examination was normal. EOG testing revealed LP/DT ratios of RE 2.20 and LE 2.33.

Discussion

Vitelliform macular dystrophy is generally regarded as a disease with onset of symptoms in childhood and adolescence. We have presented a family in which the proband first noted visual problems at age 51. The proband in our family also demonstrates that the typical egg yolk lesion may develop late in life rather than its being a congenital or early childhood finding.

It is important to examine all family members to determine all individuals at risk. Members of our family with the disease had either no disturbance or only minimal visual problems pointing to the mild form of the condition affecting them. The EOG is a selective and sensitive indicator of this disease.⁶ It established case 3 as positive for the disease despite a normal fundus examination. Vitelliform macular dystrophy can be accepted as the diagnosis in our family because of the autosomal dominant inheritance pattern and the presence of an abnormal EOG in all affected individuals.

In vitelliform dystrophy the macular lesion may be unilateral² as found in case 1. The marked variability in funduscopy appearance attributed to vitelliform dystrophy is demonstrated by case 2 in particular. This unusual presentation in an older individual shows that vitelliform can mimic age-related degenerative choroidopathy (senile disciform macular dystrophy).

References

- 1 Noble KG, Scher BM, Carr RE. Polymorphous presentations in vitelliform macular dystrophy. Subretinal neovascularization and central choroidal atrophy. *Br J Ophthalmol* 1978; **62**: 561–70.
- 2 Deutman AF. *The Hereditary Dystrophies of the Posterior Pole of the Eye*. Springfield: Thomas, 1971: 198.
- 3 Sorr EM, Goldberg RE. Vitelliform dystrophy in a 64 year old man. *Am J Ophthalmol* 1976; **82**: 256–8.
- 4 Krill AE. *Krill's Hereditary Retinal and Choroidal Disease*. Hagerstown: Harper and Row, 1977: 2: 665.
- 5 Vine AK, Schatz H. Adult-onset foveomacular pigment epithelial atrophy. *Am J Ophthalmol* 1980; **89**: 680–91.
- 6 Cross HE, Bard L. Electro-oculography in Best's macular dystrophy. *Am J Ophthalmol* 1974; **77**: 46–50.