Case Report

Follow-up of a Case of Vitelliform Macular Dystrophy Over an 8-year Period

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Abstract

Purpose: To show the follow-up of a case of vitelliform macular dystrophy with morphological and visual functional tests over an 8-year period.

Methods: Retrospective review of medical records. The morphological examination included color photography, fluorescein angiography, and ocular coherence tomography (OCT). The visual functional tests included visual acuity, electro-oculogram (EOG) and multifocal electroretinography (mfERG). The patient was observed for 8 years, from 2003 to 2011.

Results: During the follow-up, the improvement of sensory retinal detachment and reduction of yellow-white deposit were observed with color photography and fluorescein angiography. OCT revealed a decrease in sensory retinal detachment and subretinal hyper-reflective deposits; both of these morphological changes were correspondent. Visual acuity was maintained throughout the follow-up. The Arden ratio of EOG was decreased. The amplitudes of mfERG were decreased but slightly increased during the follow-up.

Conclusion: The retinal morphological changes and visual function slightly improved in this case of vitelliform maculopathy. The prognosis is good. (***Eye Science 2014; 29: 165–169***)

Keywords: vitelliform macular dystrophy; follow-up; morphology; function

Introduction

Vitelliform macular dystrophy, an autosomal dominant hereditary disease, was first described by Best in 1905, and so it is also known as Best disease. Recently, an autosomal recessive form of this disease was also reported in the literature[1]. The primary location of the disease is in the retinal pigment epithelium. The pathohistology shows that the pigment epithelium, especially in the fovea, accumulates excessive lipofusinoid material, which is then discharged from the degenerative RPE deposited between the Bruch membrane and the RPE in the fovea, resulting in previtelliform damage. The further development of the lesion then can cause damage to the photoreceptor. The accumulation of debris from the photoreceptor outer segment causes the typical vitelliform damage. Gass divided the disease into several stages: the previtelliform stage, vitelliform stage, vitelliruptive stage, serous detachment stage, atrophic stage, and choroidal neovascularization and cicatricial stage[2].

One case of vitelliform was followed up with morphology and visual function tests over an 8-year period.

Case report

A 13-year-old male (born in 1990) went to our center in June, 2003 complaining of vision blurring, color vision defect, and dismorphopsia in left eye for the previous 6 months. He had previously been diagnosed with hereditary macular disease (Best disease) at an Indonesian eye center and a Singapore eye center. He declared that he had no other eye disease and no animal contact. The temporal side of the left eye had been bruised by a basketball 6 months previously. His mother said that one of her brothers suffered from hereditary macular disease. No other family members were affected. Some

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medicines, such as centrum, inosine, carotene, and lutein were occasionally taken.

A physical examination determined that the visual acuity of his right eye was 1.0 and the visual acuity of the left eye was 0.06 and 0.1 with litter hole. The Goldmann intraocular pressure was 15 mmHg in both eyes. The cornea and lens were clear. The ophthalmoscope examination of the right eye showed that the border of the optic papillary was clear with a reddish color. The retinal vessels were normal. About 2 PD round damage appeared in the macula with bright reflections and normal fovea reflections. The left eye was the same as the right eye with semi-round yellowish subretinal damage and pigment proliferation in the macula. The retina above the damage was brightly reflective and the fovea reflection disappeared (Figure 1).

The follow-up of the patient lasted for 8 years, from June 2003 to Aug. 2011. During the follow-up, 7 color fundus photographs, 5 fluorescein angiographs, 7 OCT examinations, and 2 multifocal electroretinograms were obtained.

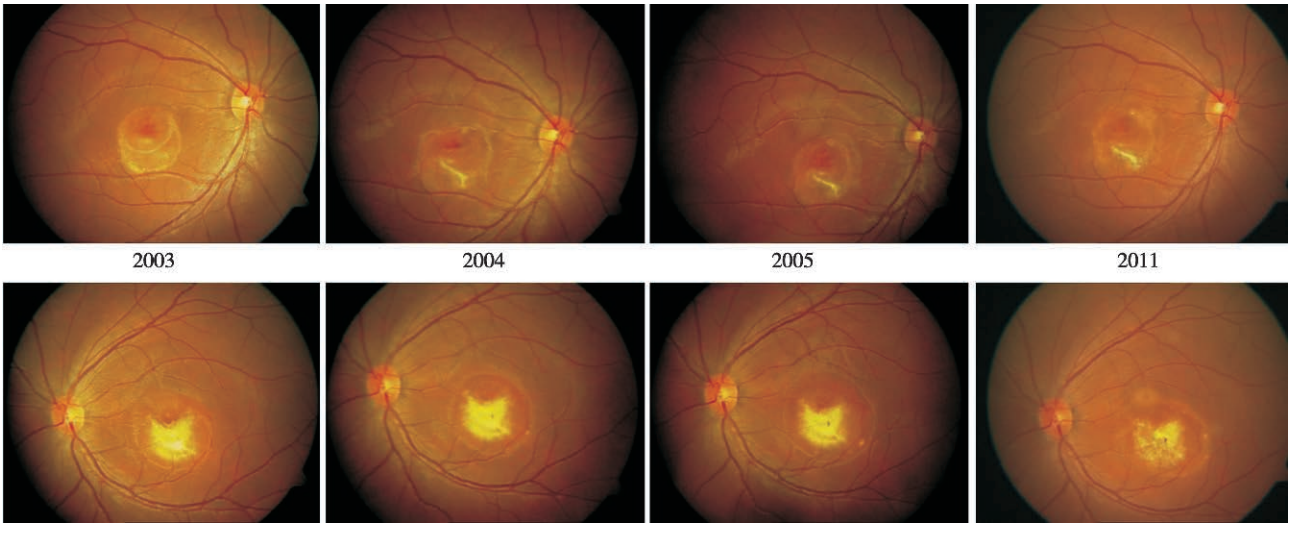

Figure 1 The fundus color photography at the initial visit, and at 1, 2, and 8 years of follow-up (the photographs of 2007, 2009, 2010 were neglected).

The follow-up of visual acuity

The patient was tested with dilated-pupil optometry examination. The right eye was corrected to 1.0 with +1.00DS+1.00DC×90°. The left eye was corrected to 0.4 with +0.75DS+1.50DC×90°. The corrected visual acuity maintained the same throughout the follow-up period.

The follow-up by color photography

At the initial visit, about 2×2PD of the sensory retinal detachment was found in macular area of the right eye. The lower-half of the damage was deposited with yellowish material. The fluid of the detachment area and the deposit were gradually absorbed during the follow-up. After one year of follow-up, one strip of proliferation existed. A 1.5×1.5PD proliferation was found in macular area of the left eye, which was reduced during the follow-up period (Figure 1).

The follow-up of fundus fluorescein angiography

In the initial visit, about 2×2PD hyperfluorescein was found in macular area of the right eye. Fluorescein leakage was found in lower-half of the lesion which was gradually reduced during follow-up. After two years of follow-up, one string of hyperfluorescein stain appeared in the lesion. In the left eye, about 2×2PD area of hyperfluorescein was found in the macula, 1.5×1.5PD area of strong fluorescein appeared in the center of the hyperfluorescein. The strong fluorescein reaction gradually decreased during follow-up. The changes in FFA corresponded with those indicated by the color photographs (Figure 2).

The follow-up of OCT

At the initial visit, about a 3 mm area of sensory

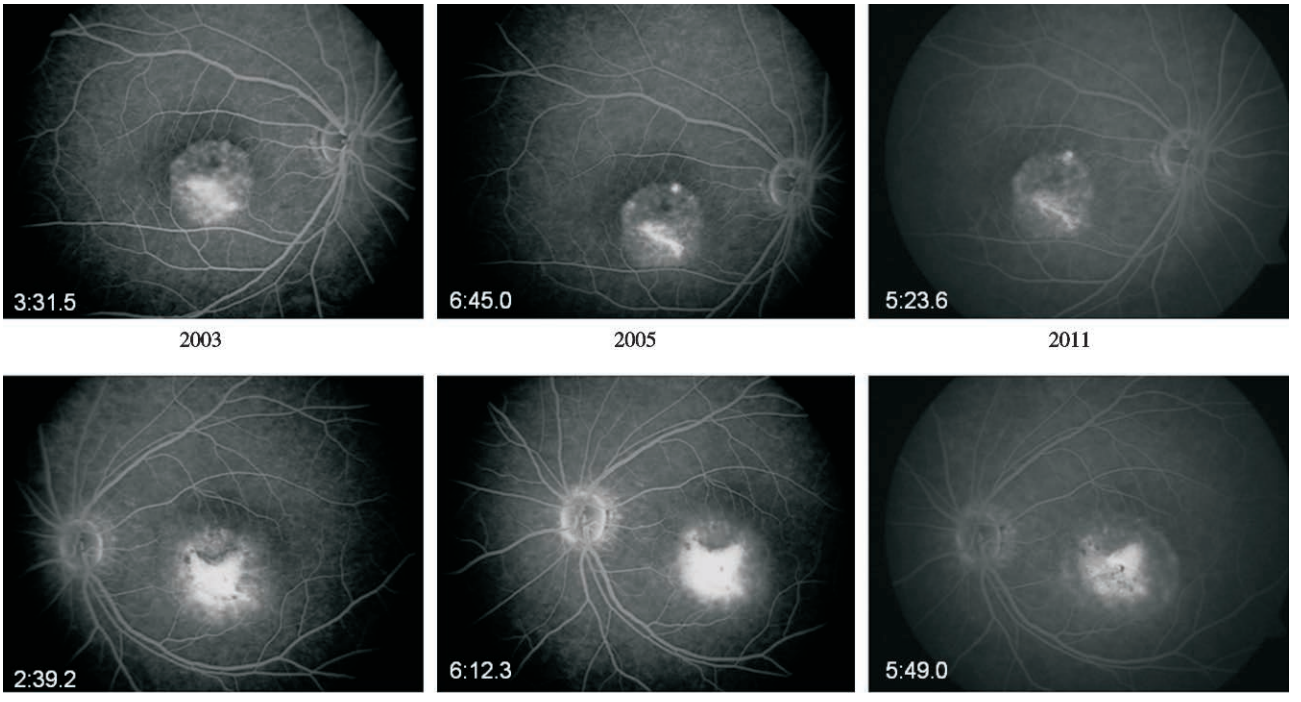

Figure 2 The fundus fluorescein angiography at the initial visit and after 2 and 8 years of follow-up (the photographs from 2007, 2009 were not included)

retinal detachment was found in the macula of the right eye. Some slightly reflective material appeared on the surface of the RPE. The sensory retinal detachment became smaller during the follow-up. In the left eye, highly reflective material appeared in the lower part of the fovea and sensory retinal detachment in the upper half of the fovea. The retinal detachment became smaller during the follow-up (Figure 3).

The test of EOG

In 2003, the patient was tested with EOG, which showed that the Arden ratio was dramatically decreased (Figure 4 and Table 1).

The follow-up of multi-focal electroretinography

The mfERG was tested in 2005 and 2011. The amplitudes of the mfERG had increased somewhat (Figure 5).

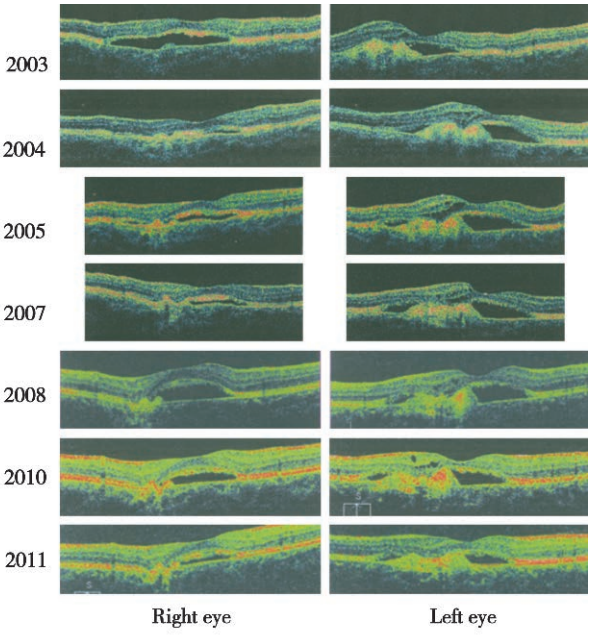

Figure 3 OCT in the initial visit and follow-up with down-up scan (the photographs of 2005 and 2007 were 5 mm scanning, the other photographs were 6 mm scanning). All of the scans passed through the fovea.

Discussion

The typical vitelliform macular dystrophy is usually seen at 3-15 years of age and then scars form. It is an autosomal dominant hereditary disease with the following clinical characteristics of the disease were as follows: ① autosomal dominant inheritance, ② the characteristic lesions in the macula were round or oval round, with clear boundaries, 0.5–4 PD size located on the retinal pigment epithelium. ③ It was often accompanied by hyperopia. ④ Fundus fluores-

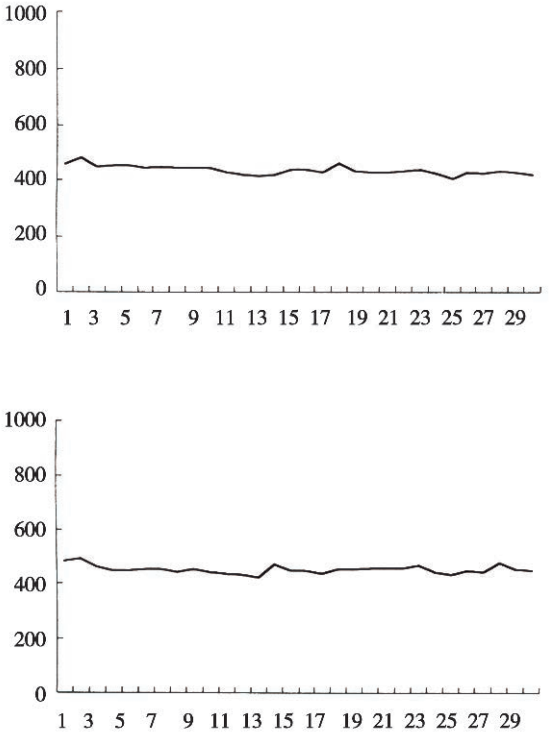

Figure 4 EOG at the initial visit. Abscissa: minutes; ordinate: μV (Upper: right eye; Lower: left eye)

Table 1 The results of EOG

	Dark-trough Time (min)	Light-peak Time (min)	Dark-trough Potential(V)	Light-peak Potential(V)	Arden ratio
Right eye	10	8	416	430	1.03
Left eye	10	10	426	480	1.23

cein angiography showed hypofluorescence from the vitelliform damage, depigmentation, and atrophy. Some cases could accompany choroidal neovascularization. ⑤ The characteristic EOG change might appear in the previtelliform stage.

The Arden ratio was less than 1.5. This case was found in a 13-year-old. One of the brothers of his mother supposedly had a similar disease. The symmetrical macular damage, the accompanying hyperopia and changes of visual electrophysiology, and better visual acuity were correlated with the above characteristics. The patient could be diagnosed with vitelliform macular dystrophy although he lacked the typical vitelliform lesion. The disease was in a stage between sensory retinal detachment stage and the cicatricial stage.

The patient was followed up using morphological and electrophysiological tests. The color photography of his right eye showed that the sensory retinal de-

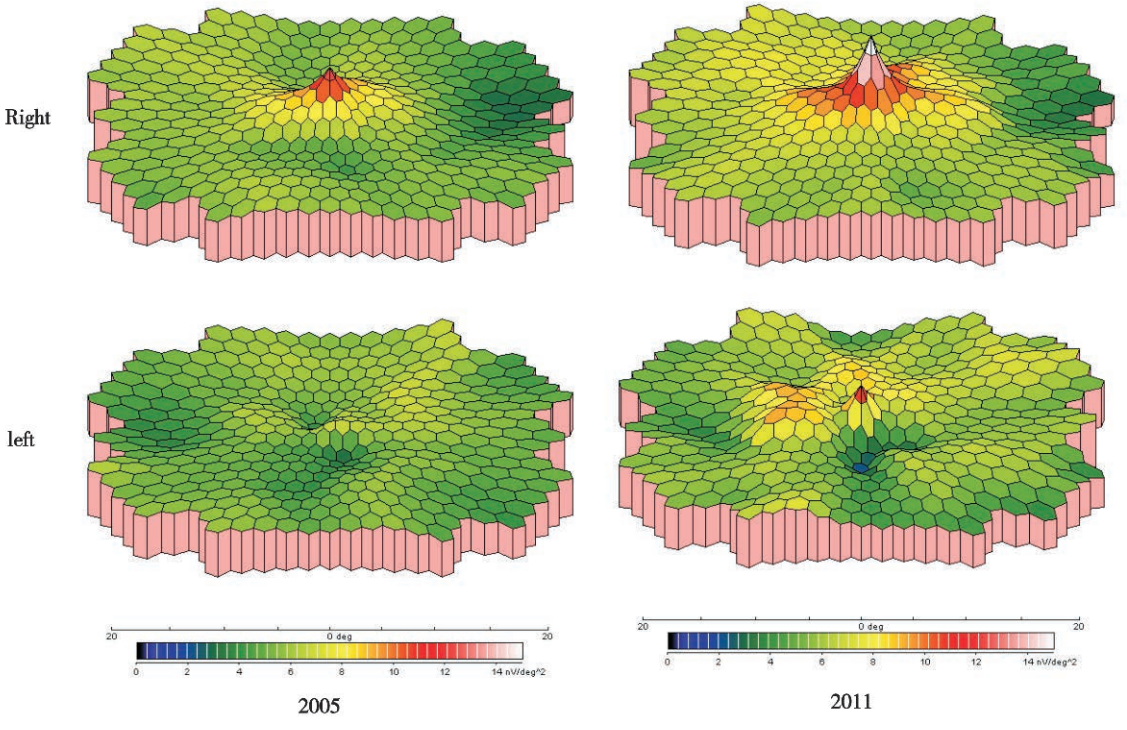

Figure 5 The mfERGs in 2, 8 year follow-up. Upper: right eye; lower: left eye

tachment of the macula was improved and the subretinal deposit was gradually absorbed. The damage stayed stable after 1 year of follow-up. The scar on the left eye was reduced during the 8 year follow-up. The fundus fluorescein angiography showed similar changes.

The OCT changes in Best disease have been reported in the literature[3-4]. The changes include serous retinal detachment, hyperreflective interface between the inner section and outer section, hyporeflection corresponding to the inner nuclear layer, and hyperreflection between the hyporeflective outer nuclear layer and hyperreflective RPE[3]. Kay found that the vitelliform damage consisted of materials above the RPE and below the tip of the outer segment. Some patients had fiber nodes under the RPE[4]. This patient had a similar clinical appearance. The follow-up showed that the serous retinal detachment was gradually absorbed and that the hyperreflective material was gradually reduced, which corresponded with the FFA observations. OCT and FFA were important in monitoring the progress of the disease.

Most patients maintain good visual acuity. In the whole disease process, one eye often preserves better visual acuity. The patient may have accompanying hyperopia, strabismus, and amblyopia. The functional abnormality, especially the visual electrophysiology, may appear several years prior to the appearance of macular disease with the ophthalmoscope. A characteristic change is the reduced Arden ratio, but some patients may have near normal EOG[5]. The Ganzfeld ERG was normal but the focal ERG was abnormal. The amplitude may be decreased even when the visual acuity is still good. The EOG of this patient showed a dramatically decreased Arden ratio. His visual acuity remained stable during the 8 year follow-up. The mfERG showed a slight amplitude increase. These results demonstrated that the visual function may be preserved for a long time.

Because the incidence of vitelliform macular dystrophy is low, long period follow-up of the disease is rare. The follow-up of this case may aid us in understanding the course and the prognosis of this disease.

References

1 Bitner H, Mizrahi-Meissonnier L, Griefner G, et al. A homozygous frameshift mutation in BEST1 causes the classical form of Best disease in an autosomal recessive mode. Invest Ophthalmol Vis Sci, 2011, 52 (8): 5332–5338.

2 Agarwal A. Gass' Atlas of macular diseases. Elsevier, Saunders, printed in China, 2012, PP240–252.

3 Xu H, Ying L, Lin P, et al. Optical coherence tomography for multifocal vitelliform macular dystrophy. Optom Vis Sci, 2013, 90(1): 94–99.

4 Kay CN, Abramoff MD, Mullins RF, et al. Three-dimensional distribution of the vitelliform lesion, photoreceptors, and retinal pigment epithelium in the macula of patients with best vitelliform macular dystrophy. Arch Ophthalmol, 2012, 130(3): 357–364.

5 Low S, Davidson AE, Holder GE, et al. Autosomal dominant Best disease with an unusual electrooculographic light rise and risk of angle-closure glaucoma: a clinical and molecular genetic study. Mol Vis, 2011, 17: 2272–2282.