

Case Report

Best Vitelliform Macular Dystrophy with Central Retinal Artery Occlusion

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Best Vitelliform macular dystrophy is a rare condition characterized by deposition of lipofuscin within the retinal pigment epithelium.

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We present a case of Best Vitelliform dystrophy with central retinal artery occlusion.

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Best's disease or Best macular dystrophy is an autosomal dominant disorder. First pedigree was described by a German ophthalmologist Dr. Franz Best in 1905¹. During its course the disease evolves gradually through five stages. This case presents the two different stages of the disease in the same patient along with the central retinal artery occlusion.

CASE REPORT

In October 2008, a 45years old (Pakistani) male presented in out patient department with sudden loss of vision in right eye two weeks earlier. While probing into the history, it was revealed that he had dimness of vision in both eyes for the last fifteen years. Two weeks back he developed sudden loss of vision in his right eye which was not associated with pain and redness of eyes. He was an average built man, non-smoker, non-diabetic, non-hypertensive and did not have any other systemic disease.

Ocular examination showed orthophoria with full extra ocular movements. His best corrected visual acuity was perception of light in right eye and 6/60 in the left eye. Intra ocular pressures were 12mm of mercury in both eyes. There was right relative afferent

pupillary defect. On slit lamp examination anterior segment was unremarkable. Pupils were dilated and fundoscopy was performed. There was a large round egg-yolk like lesion involving the whole macula in right eye. Branches of central retinal artery were thread-like and disc was pale. Left fundus showed multifocal yellowish lesion with a large central lesion of 2.5 to 3 Disc diameters at the fovea. The disc and the retinal vasculature was normal in this eye.

On the basis of history and examination, he was diagnosed a case of Best macular dystrophy (Vitelliform stage) with central retinal artery occlusion in right eye and Best macular dystrophy (Vitelliruptive stage) in left eye. EOG was not performed because of lack of electrophysiological test facilities.

Keeping in view the central retinal artery occlusion, cardio vascular system was evaluated. Complete blood count, ESR, lipid profile, ECG, Echocardiography and carotid Doppler were all normal (fig.1). As there is no treatment available for Best's disease and the central retinal artery occlusion was few weeks old, the patient was reassured and called after six months for follow up.

After six months, patient's best corrected visual acuity was 6/18 in the right eye and it was only on the temporal side. Vision in the left eye was unchanged. On fundoscopy Pseudohypopyon stage was seen in the right eye. Foveal area was clear of the lipofuscin pigment and Cilio-retinal artery could be seen supplying the macular area (fig. 2). This finding was consistent with the improvement in visual acuity. In the left eye it was Vitelliruptive stage as before (fig. 3).

DISCUSSION

Best Vitelliform macular dystrophy is one of the rare fundus dystrophies. Inheritance is autosomal dominant with variable penetrance. Five stages of the disease has been recognized².

Stage 0: Pre-Vitelliform

Stage 1: Pigment mottling at macula

Stage 2: Vitelliform with egg yolk macular lesion

Stage 3: Pseudohypopyon when part of lesion gets absorbed

Stage 4: Vitelliruptive with a scrambled egg appearance.

EOG is sub normal in all these stages.

Adult onset foveomacular Vitelliform dystrophy in contrast have smaller lesions, present late in life and do not demonstrate similar stages. Morphologically both these diseases have similarities but they are generally considered different entities³.

This patient was an unusual case in two aspects; visual acuity and association of this disease with central retinal artery occlusion. Visual acuity in Best Vitelliform degeneration is generally good in the Vitelliform stage. It is only with the onset of Vitelliruptive stage when the vision starts to deteriorate. This occurs due to retinal pigment epithelial atrophy. According to Fishman GA⁴ and colleagues, fall in visual acuity is more in patients above 50 years of age. Our patient had only perception of light in right eye (even with the Vitelliform stage) at the time of presentation. Disc pallor with thread like arterioles indicated old central retinal artery occlusion. Once the lipofuscin started to absorb from the foveal area the visual acuity improved because of the presence of cilioretinal artery. Vision in the left eye with vitelliruptive stage remained 6/60.

Data available so far suggests that Best Vitelliform degeneration is an isolated disease with no systemic associations. However, there is a case report⁵ in which



Fig. 1: Carotid Doppler right common carotid artery, showing normal flow.

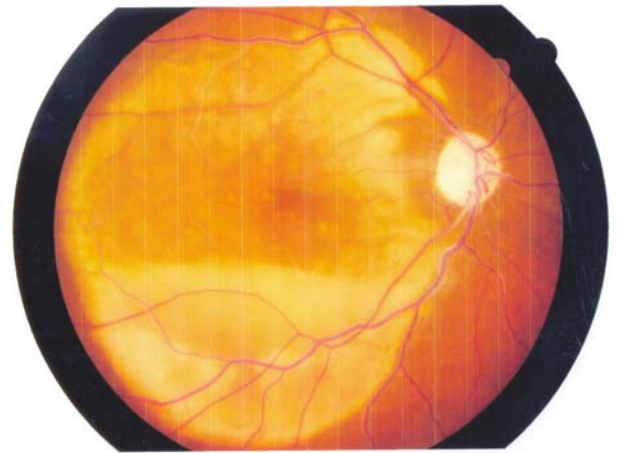


Fig. 2: Pseudohypopyon stage with white pale disc six months after the initial presentation.

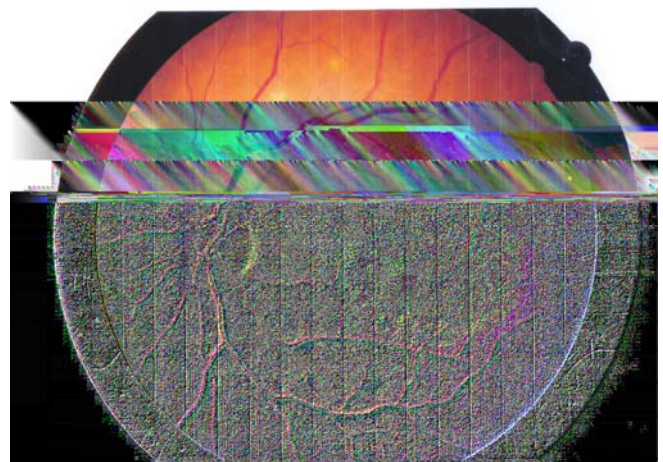


Fig. 3: Vitelliruptive stage in left eye.

Adult onset foveomacular Vitelliform dystrophy was associated with Neurofibromatosis type 1. Similarly, there are few case reports of retinal folds, central serous chorioretinopathy and choroidal neovascularization being associated with adult onset Vitelliform degeneration but not with the juvenile type.

This disease has a wide range of expressivity and Electro-oculography is the diagnostic test in doubtful cases. Nobel KG⁶ presents a study in which two cases of choroidal neovascularization and central choroidal dystrophy later turned out to be Vitelliform macular dystrophy on EOG. Similarly, lesions in Best's disease are usually single and central but there are few reports which describe multiple peripheral lesions⁷ outside the macula and posterior pole. Due to the diverse clinical presentations of this disease, there are many single case reports found in literature. Our case is another unique addition.

CONCLUSION

Our patient had an unusual presentation of Best Vitelliform degeneration with central retinal artery occlusion. To the best of our knowledge this is the first report. On cardio vascular investigations no cause of artery occlusion was found. How is it related to the central retinal artery occlusion needs to be established by further studies.

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Every case of acute glaucoma is not angle closure

Every case of raised IOP with a clear cornea and deep AC is not primary open angle glaucoma. Determine the underlying cause of glaucoma and give appropriate treatment.

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