

“A Case Report on Best’s Vitelliform Macular Dystrophy.”

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Abstract : Best’s disease also termed as vitelliform macular dystrophy is an autosomal dominant disorder which classically presents in childhood with the striking appearance of a yellow or orange yolk like lesion in the macula. Dr Franz Best, a German ophthalmologist, described the first pedigree in 1905.

A 23year old male with complain of gradual reduction of vision distortion of images and presence of blind spots in the centre of his visual field in his both eyes from last 7 years presented to the out patient department with no similar family history. His visual acuity on snellen’s chart was found to be 6/24 B/E with improvement upto 6/18P in his right eye and no improvement in left eye on refraction. Fundus examination showed well circumscribed around 1.5 disc diameters in size yellowish lesion at fovea B/E with abnormal EOG, hyperfluorescent well circumscribed lesion at fovea on FFA and subretinal mound on OCT B/E all pointing towards the diagnosis of Best’s vitelliform macular dystrophy B/E.

Keywords : Best’s disease, EOG, FFA, metamorphopsia, OCT

I. Introduction

Macular dystrophies are arbitrarily classified on the basis of their tissue of origin into those originating from nerve fibre layer, photoreceptors and retinal pigment epithelium (RPE), RPE alone, Bruch’s membrane and choroid. Vitelliform dystrophy of the fovea is a separate entity among the inherited macular dystrophies. It was first reported by Adam [1] and the first pedigree was described by Dr Franz Best, a German ophthalmologist, described the first pedigree in 1905. Best’s disease also termed as vitelliform macular dystrophy is an autosomal dominant disorder with variable penetrance [1], involving the long arm of chromosome 11 (11q12-q13) [2]. It is a bilateral & asymmetrical disease which classically presents in childhood with the striking appearance of a yellow or orange yolk like lesion in the macula. Many individuals with Best disease initially are asymptomatic, with fundus lesions noted on examination. Visual symptoms can include decreased acuity (blurring) and metamorphopsia. These symptoms may worsen if the disease progresses to the atrophic stage.

II. Case Report

A 23year old male with history of gradual reduction of vision, distortion of images and presence of blind spots in the centre of his visual field in his both eyes from last 7 years presented to the out patient department of M.L.B Medical college, Jhansi. He gave no history of any trauma, redness, pain, watering or discharge from either eye. He was not a known diabetic, hypertensive and never suffered any chronic disorder. There was no similar history in the immediate family. On examination he was found to be orthotropic with a Snellen’s vision of 6/24 B/E with improvement upto 6/18P in his right eye and no improvement in left eye on refraction . Both eyes were externally quiet with clear corneas, clear anterior chambers, briskly reacting pupils with normal iris morphology and transparent lenses. Fundus examination of the both eyes (B/E) revealed a clear media with a normal well- defined vertically oval optic disc. The macula showed a 1 disc diameter in size in RE and 1.5 disc diameter in LE, well-circumscribed, single, yellow, sub- retinal lesion of a variable density, over which normal retinal blood-vessels traversed (Fig.1). There was no evidence of past or current inflammation and the remaining retina appeared normal. The overlying retina was normal, as was the peripheral retina. The intra-ocular pressures were normal with applanation tonometry. The patient was investigated to rule out any specific cause of sub-retinal inflammation and all systemic investigations were normal. In view of the yellow vitelliform lesions in both eyes Best’s vitelliform dystrophy was suspected and the patient was investigated with fundus fluorescein angiography (FFA), optical coherence tomography (OCT) and an electro-oculogram (EOG). FFA of B/E (Fig. 2) showed a normal optic disc, a large sub- retinal area of mottled hyperfluorescence that was well-defined and had a distinct upper level. The overlying and peripheral retina appeared normal. FAZ was still largely normal as was the central and peripheral retina. OCT (Fig. 3) showed a medium sized subretinal area at macula showing RPE disruption and thickening suggestive of pigment accumulation . There was no evidence of a choroidal neo-vascular membrane (CNVM). EOG showed light peak values of 217 uV and dark trough value of 143 uV thus giving an Arden ratio of 1.51 which was significantly below the normal of 2. Electro-retinogram (ERG) showed value of 210 uV for ‘a’ wave and a value of 410 uV for ‘b’ wave under mesopic conditions. ERG values were well within normal limits. Thus on basis of bilateral multiple well-circumscribed yellow-

orange macular sub-retinal lesions, OCT findings and revealed EOG-ERG dissociation on electro-physiology, a diagnosis of Best’s vitelliform dystrophy was made. LE probably had a more advanced disease and had reached Stage 3 or pseudohypopyon (Fig. 1 - arrow) stage while LE had a typical multi-focal vitelliform Stage 2 lesion.

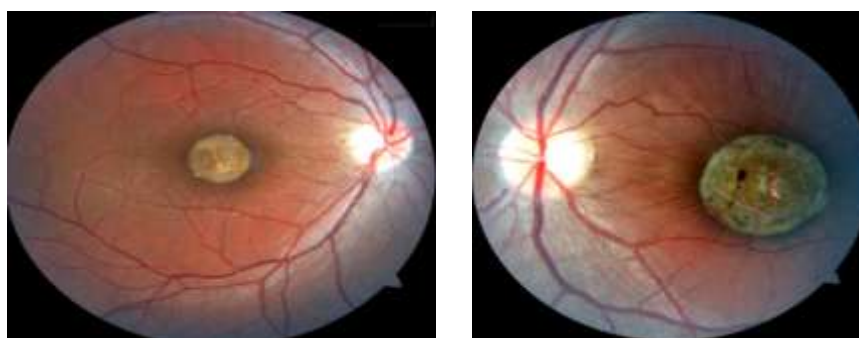


Figure 1. Fundus picture of right eye and left eye respectively.

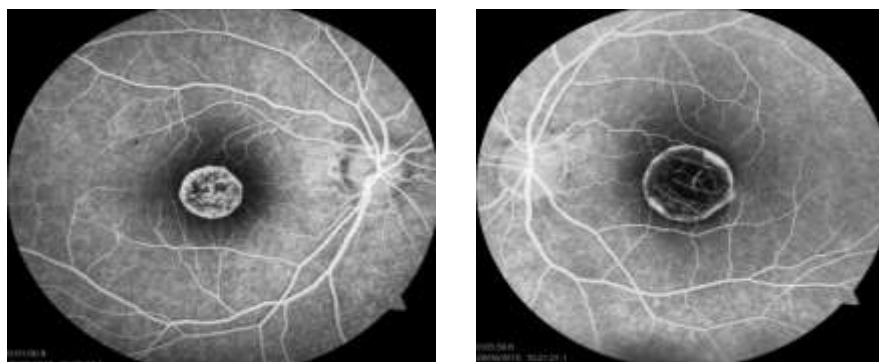


Figure 2. Fundus fluorescein angiography of right eye and left eye respectively.

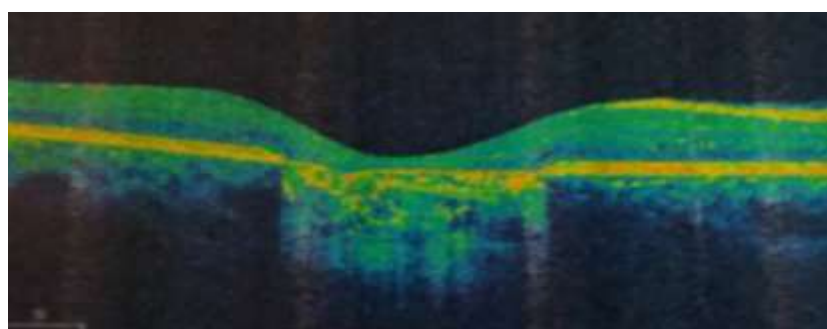


Figure 3. Optical coherence tomography(OCT)

III. DISCUSSION

Best’s disease is one of the rare an autosomal dominant macular dystrophy of variable penetrance in which lipofuscin granules accumulate in retinal pigment epithelial cells^(3, 4). It typically affects young patients in whom a macular lesion gradually evolves through several characteristic stages⁽⁵⁾.

Dutmann classified the evolution of Best disease in different stages⁽⁶⁾ which has been modified by Mohler and Fine in 1981:

- Stage 1:** Normal fundus, abnormal EOG
- Stage 2:** Pre-Vitelliform stage
- Stage 3:** Vitelliform stage ("sunnyside-up egg yolk")
- Stage 4:** "scrambled egg " stage
- Stage 5:** Cyst stage
- Stage 6:** " Pseudohypopyon" stage
- Stage 7:** Atrophic stage

In Best’s disease abnormality is in the RPE, as noted on histopathology and electrophysiology testing. Lesions are restricted to the eye without any systemic associations. Petrukhin et al ^[7], identified the retina-specific gene and designated it as the VMD2. The protein encoded by this gene was proposed to be called bestrophin. Weingeist et al ^[8], reported an abnormal accumulation of lipofuscin granules within the RPE. The RPE have degenerative changes in some cases and there may be secondary loss of photoreceptor cells.

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 reduce. This occurs due to retinal pigment epithelial atrophy. According to Fishman GA ⁽⁹⁾ and colleagues, fall in visual acuity is more in patients above 50 years of age.

This disease has a wide range of state and in doubtful cases Electro-oculography is the diagnostic test which is abnormal in all stages. Patients usually maintain visual acuity better than 6/12 through the vitelliruptive stage ⁽⁵⁾. It is in the atrophic stage that visual acuity can deteriorate to the level of legal blindness because of retinal pigment epithelial cell and photoreceptor disruption and death. Similarly, lesions in Best’s disease are frequently single and central but there are few reports ⁽⁴⁾ which describe multiple peripheral lesions outside the macula and posterior pole.

There is no effective treatment available to slow the progression of Best’s disease. Antioxidant supplementation might have a theoretical benefit, given the role of free radicals in the formation of lipofuscin ⁽¹⁰⁾. The patient has to be observed for development of CNVM that may have to be treated using photodynamic therapy or intra- vitreal anti-vascular endothelial growth factors.

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