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Best Macular Dystrophy in a Nigerian: A Case Report

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Key Words

Best macular dystrophy · Choroidal neovascular membrane · Retina · Nigerian

Abstract

Best macular dystrophy is reported to be rare in Africans. It is a hereditary disease that starts in childhood and progresses through some stages before visual symptoms occur. This case report presents a 43-year-old Nigerian with the disease and stresses the importance of regular eye exams of patients and relatives to detect changes such as choroidal neovascular membrane amenable to treatment.

Case Presentation

A.T., a 43-year-old Nigerian lady, presented to the Eye Clinic of the University College Hospital, Ibadan with gradual blurring of vision in both eyes for 4 years. Two of her siblings were reported to have similar complaints. Ocular examination showed unaided visual acuity of 6/12 OU corrected to 6/9 in both eyes. Near vision was normal. The anterior segment examination was not remarkable. IOP was 13 mm Hg in both eyes. Dilated funduscopy showed a well circumscribed cystic fovea lesion with a fluid level and an atrophic center in the right eye, while the left eye showed a scar in the macula ([fig. 1](#) and [fig. 2](#)). The systemic examination was not remarkable. An assessment of Best macular dystrophy was made. The patient was prescribed with spectacles, counseled and placed on close observation with Amslers grid for the development of choroidal neovascular membrane that will require urgent treatment.

Discussion

This is probably the first case of Best macular dystrophy to be reported in Ibadan, Nigeria and possibly Sub-Saharan Africa to the best of the author's knowledge. The disease is a hereditary macular dystrophy first described by Best in 1905 [1]. It has also been reported in Caucasians and Asians [2], the other cases seen in African Americans were associated with sickle cell trait [3]. The disease starts in childhood with a

characteristic macular lesion resembling an egg yolk. It progresses through some stages before visual symptoms become apparent. [Table 1](#) describes these stages [\[4\]](#).

Pathophysiology

The pathophysiology of Best's disease is explained by abnormality in the retinal pigment epithelium (RPE) with resultant abnormal ionic transport leading to the accumulation of lipofuscin in the RPE cells and sub-RPE space in the macular area. Degeneration of RPE cells can occur [\[5, 6\]](#).

Vision is good in the early stages of the disease. The vitelliruptive stage may herald visual deterioration which becomes worse in the atrophic stage due to the presence of choroidal neovascular membrane. Our patient presented with the early stages of the disease, hence the good visual acuity. The patient will be under observation and follow-up so as to detect changes amenable to treatment. Choroidal neovascular membrane (CNVM) from Best's disease has been reported to respond well to intravitreal antiVEGF [\[7, 8\]](#). Fundus fluorescein angiography is essential in confirming the presence of CNVM and should be done when patient presents with sudden deterioration in vision. If a CNVM develops, then a corresponding area of hyperfluorescence with leakage will be found.

Since the disease is an autosomal dominant disorder, other family members will benefit from regular fundus examinations, and Amsler's grid is a valuable tool for monitoring central vision. An electrophysiologic test such as electrooculogram (EOG) is specific for confirming the presence of the disease in relatives even in the absence of clinical signs and symptoms. A severe decrease occurs in light response, reflected by an Arden (light-peak/dark-trough) ratio of 1.1–1.5 (the normal Arden ratio is 1.8) [\[9\]](#). The full-field electroretinogram (ERG) result is normal in Best's disease. A focal ERG or multifocal ERG, concentrating on macular function, reveals abnormal function corresponding to the area of anatomical disruption [\[10\]](#).

Conclusion

This is a report of Best macular dystrophy in Ibadan, Sub-Saharan Africa. Regular eye exams are advised for families with the disease. Other so-called rare ocular diseases may not be so after all.

Table 1. Stages of Best macular dystrophy (modified) [4]

Stage of disease	Findings	Expected visual acuity
I Pre-vitelliform	Speckled fine pigmentary disturbance in the macula is seen	6/6
II Vitelliform	Egg-yolk lesion composed of a round, homogeneous, opaque yellow lesion with discrete margins measuring approximately 1 disc diameter in size	6/6–6/12
III Pseudo-hypopyon	The yellow material within the vitelliform cyst develops a fluid level, resulting in the appearance of a pseudohypopyon	6/6–6/36
IV Vitelliruptive/atrophic	Advanced disease with an atrophic macular pigment epithelium (stage IVa), fibrous scarring (stage IVb), or subretinal neovascularization (stage IVc)	6/6–6/60, CNVM may be associated with VA less than 6/60

Fig. 1. Right eye of patient with the pseudohypopyon stage of Best's disease.

Fig. 2. Left eye of patient with the atrophic stage of Best's disease.

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