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Medical Science

MACULAR HOLE IN A YOUNG MALE FROM A FAMILY OF BEST'S DYSTROPHY

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ABSTRACT The occurrence of macular hole with Best's dystrophy is an extremely rare finding with very few cases reported in the literature so far. This case is with typical Best's dystrophy in one eye and a macular hole in the contralateral eye. Different stages of Best's dystrophy were noted in this family. The patient's father had undergone the usual course of the disease but the young patient presented with macular hole. This is probably first family with unusual stage of Best's disease being reported from India.

KEYWORDS :

Case Report:

A 38 year old male presented with complaints of progressive diminution of vision in both eyes since 2 years. There was loss of central vision in LE over last 2 months. The father also had history of defective vision in both eyes for many years. There were no other significant systemic complaints. There was no significant drug history. His general examination was within normal limits. Visual acuity in RE was 6/24 and 2/60 in LE. The fundus examination revealed absence of foveal light reflex with vitelliform stage (an egg-yolk like appearance of 1.5-disc diameter at the macular region) of Best's dystrophy in the RE[Figure 1(a)] and full thickness macular hole in the LE[Figure 1(b)]. Further, electrooculogram(EOG) was performed to confirm this diagnosis.

Electrooculogram showed diminished response in both the eyes. Arden's ratio was 1.3 in RE and 1.1 in LE. This implied that significant amount of functional impairment occurred at the vitelliform stage itself, however its progression to hole formation was unclear. OCT showed an optically empty space between retinal pigment epithelium(RPE) and the neurosensory retina in RE[Figure 2] with full thickness macular hole in the left[Figure 3]. There was no surgical intervention due to poor visual prognosis and full thickness hole of long standing duration.

As Best's dystrophy is autosomal dominant, the patient's family members were examined. The father had atrophy stage of Best's in both eyes[Figure 1(c)] and elder brother had previtelliform stage of Best's in both eyes[Figure 1(d)]. The two sons of the patient, one 10 year old and other 12 year old were also examined. They did not have any positive fundus findings then.

Discussion:

Best disease, also termed vitelliform macular dystrophy, is typically 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[1]. The lesion evolves through several stages over many years, with increasing potential for adverse visual outcome. A hallmark of the disease is a markedly abnormal electrooculogram in all stages of progression and in phenotypically normal carriers[2].

Lesions in Best disease are restricted to the eye. No systemic associations exist. Abnormalities in the eye result from a disorder in the retinal pigment epithelium. A dysfunction of bestrophin results in abnormal fluid and ion transport by the RPE[4]. Lipofuscin (periodic acid-Schiff [PAS] positive) accumulates within the RPE cells and in the sub-RPE space, particularly in the foveal area. The RPE appears to have degenerative changes in some cases, and secondary loss of photoreceptor cells has been noted[5]. Breakdown of RPE/Bruch's membrane can allow choroidal neovascularization to develop as a late complication. The mechanism that leads to macular hole is unclear.

The incidence of best dystrophy in Indian families is very rare. There are no statistics available on the actual incidence in India. There is mention of only one case of a giant macular hole with Best's disease in a young boy from India so far[3].

In this report, we have seen different stages of the Best's disease in the same family. The father and elder brother had undergone a usual course of the disease but our patient presented with macular hole which is an unusual occurrence. This is probably the first family with unusual stage of Best's disease being reported from India.

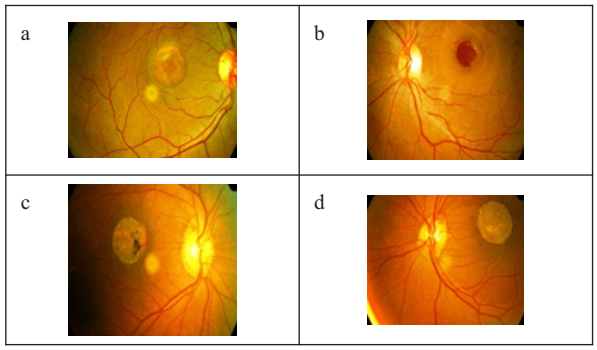

Figure 1: (a) Patient's RE: Vitelliform Stage of Best's (b) Patient's LE:Macular hole (c) Patient's father: RE:Atrophy stage of Best's (d)Patient's brother: Previtelliformstage of Best's.

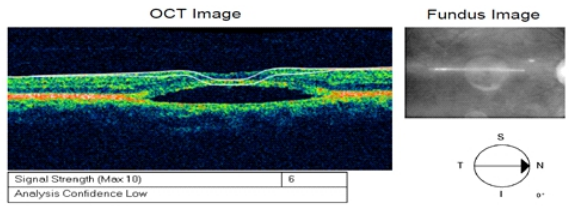

Figure 2: OCT showing an optically empty space between retinal pigment epithelium(RPE) and the neurosensory retina in RE

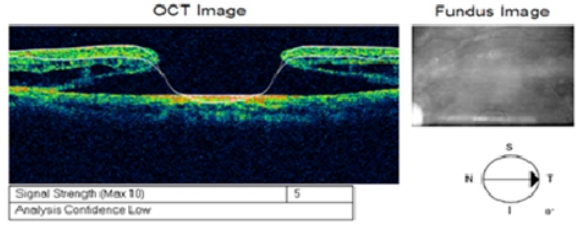

Figure 3: OCT showing a full thickness macular hole in the left eye

References:

1) Best F. Ubereine hereditare Maculaaffektion. Beitrage zur Vererbungslehre. Augenheilkd. 1905. 13:199.
2) Pinckers A, Cuypers MH, Aandekerk AL. The EOG in Best's disease and dominant cystoid macular dystrophy (DCMD). Ophthalmic Genet. 1996 Sep. 17(3):103-8.
3) Mandal S1, Basu S. Giant macular hole with Best's disease in a young boy. Clin Exp Optom. 2012 Nov;95(6):643-5. doi: 10.1111/j.1444-0938.2012.00755.x. Epub 2012 Jun 24.
4) Weingeist TA, Kobrin JL, Watzke RC. Histopathology of Best's macular dystrophy. Arch Ophthalmol. 1982 Jul. 100(7):1108-14.
5) Leu J, Schrage NF, Degenring RF. Choroidal neovascularisation secondary to Best's disease in a 13-year-old boy treated by intravitreal bevacizumab. Graefes Arch Clin Exp Ophthalmol. 2007 Nov. 245(11):1723-5.