Angle-closure glaucoma associated with autosomal recessive bestrophinopathy

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Key words: Angle-closure glaucoma, autosomal recessive bestrophinopathy, electro-oculography

Mutations in the *BEST1* gene have been associated with different ocular phenotypes. They affect the retinal pigment epithelium (RPE) metabolism and consequently the outer retinal function with which the RPE is intimately associated. Autosomal recessive bestrophinopathy (ARB) is one among its phenotypic variants. Here, we report the case of a patient who presented with angle closure glaucoma and was diagnosed as ARB on fundoscopy and subsequent investigations.

A 38-year-old female patient presented to us with complaints of defective vision in both eyes (BE) and with a history of one episode of vomiting. Her best-corrected visual acuity (BCVA) was 6/18 in both eyes. Intraocular pressure (IOP) was 30 and 34 mmHg in the right and left eyes, respectively. The anterior segment was unremarkable, except for a shallow chamber by Van Herick grade 2 in BE [Fig. 1]. Gonioscopy revealed occludable angles in BE. She underwent a YAG peripheral iridotomy (PI) in BE. On the subsequent visit, dilated fundoscopy was performed that revealed advanced glaucomatous optic disc cupping and yellowish–white deposits in the posterior pole in BE. She underwent retinal autofluorescence imaging and optical coherence tomography (OCT), which revealed autofluorescent subretinal deposits with subretinal fluid in BE [Fig. 2]. Electrooculogram (EOG) was performed according to the current International Society for Clinical Electrophysiology of Vision (ISCEV) standards using the Espion Red Profile Ganzfield Profile System (Diagnosys LLC, USA), which revealed characteristic absent light rise, decreased/abnormal light rise-to-dark trough ratio (LP: DT), 1.08 in the right eye and 1.18 in the left eye (normal >1.6–1.8), suggestive of Best disease. However, electroretinogram (ERG) could not be performed due to the clinical condition of the patient as control of intraocular pressure was more important. A provisional diagnosis of ARB was made in association with chronic angle-closure glaucoma because OCT and EOG were more in favor of ARB.

Discussion

Mutations identified in the *BEST1* gene that encode bestrophin-1 to date are associated with a wide range of phenotypes, including autosomal dominant Best vitelliform macular dystrophy (BVMD), adult vitelliform macular dystrophy, autosomal dominant vitreoretinochoroidopathy (ADVIRC), the MRCS (microcornea, rod-cone dystrophy, cataract, posterior staphyloma) syndrome, retinitis pigmentosa, and ARB.[1,2]

Bestrophin-1 dysfunction has been found to be associated with defective regulation of subretinal fluid reabsorption and aberrant phagocytosis of the photoreceptor discs.[3]

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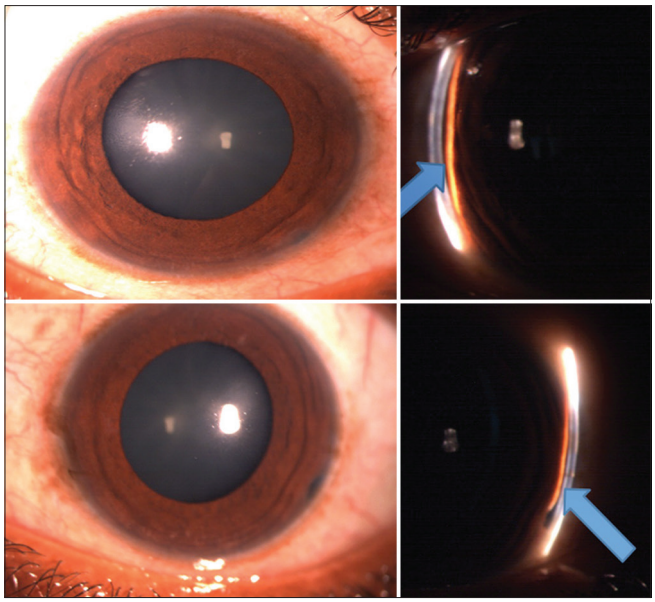

Figure 1: Slitlamp image right eye (RE) and left eye (LE) showing shallow anterior chamber (Van Herick grade 2) with patent peripheral iridotomy

BVMD is associated with vitelliform lesions at the macula due to the deposition of lipofuscin. ARB, in contrast, is present predominantly along the arcades as seen widely in our patient.[2,4-7] Best disease in addition is associated with narrow angles as in our patient, with reduced axial length, hypermetropia, and sometimes small eyes.[1,2] Angle-closure glaucoma is associated with more than 50% of ARB.

Our patient presented with acute elevation of IOP with narrow angles with advanced disc damage. Despite a YAG PI, her IOP could not be controlled with maximum anti-glaucoma medications. Her visual fields by Humphrey field analyzer (HFA) showed advanced field defect; hence, we planned a micropulse transscleral cyclophotocoagulation (MPTSCP) for BE sequentially to reduce the IOP temporarily and preserve vision, as it causes less thermal damage to collateral tissues. On subsequent follow-up, her IOP in BE was 18 mmHg on maximal medical therapy.

In conclusion, we report this clinically diagnosed ARB with angle-closure glaucoma though further studies are required to confirm. A very careful examination in these eyes is required due to their frequent association with narrow angles, which can be missed as ARB can also result in a sudden drop in vision. Hence, a thorough IOP monitoring and disc evaluation are needed as in our case who had advanced disc damage. Managing glaucoma from progressing and timely intervention to prevent vision loss from advanced optic nerve head damage are crucial.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

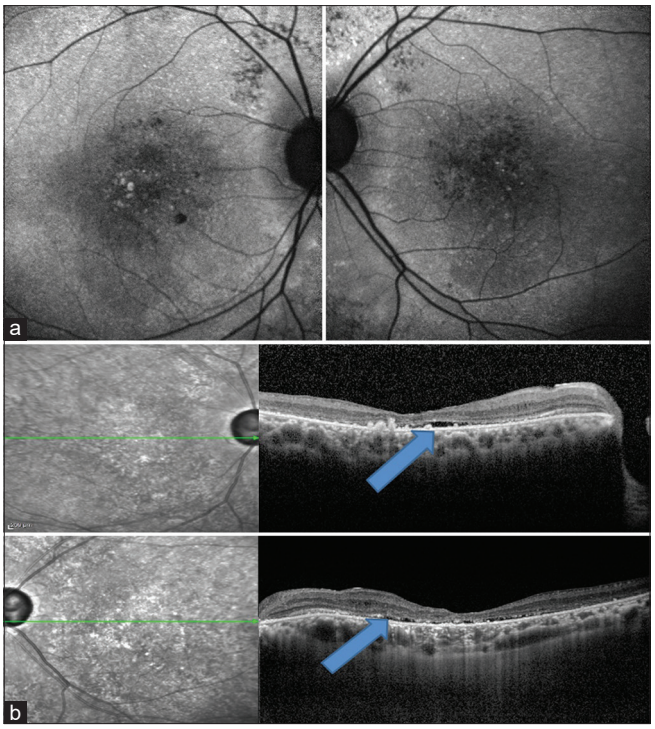

Figure 2: (a) Autofluorescent images demonstrating widespread autofluorescent lesions typical of ARB in BE. (b) Spectral domain OCT images demonstrating subretinal deposits, intraretinal and subretinal fluid in BE

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Conflicts of interest

There are no conflicts of interest.

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