AUTOSSOMAL RECESSIVE BESTROPHINOPATHY: A CASE REPORT

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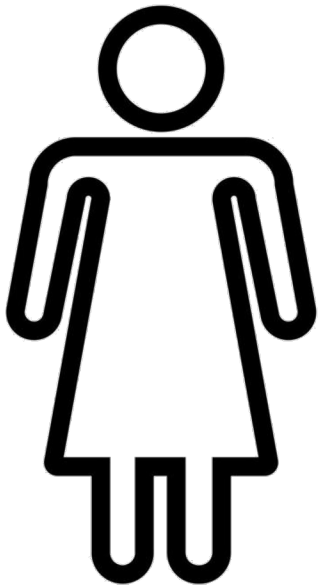
PURPOSE

- To report the case of Autossomal Recessive Bestrophinopathy

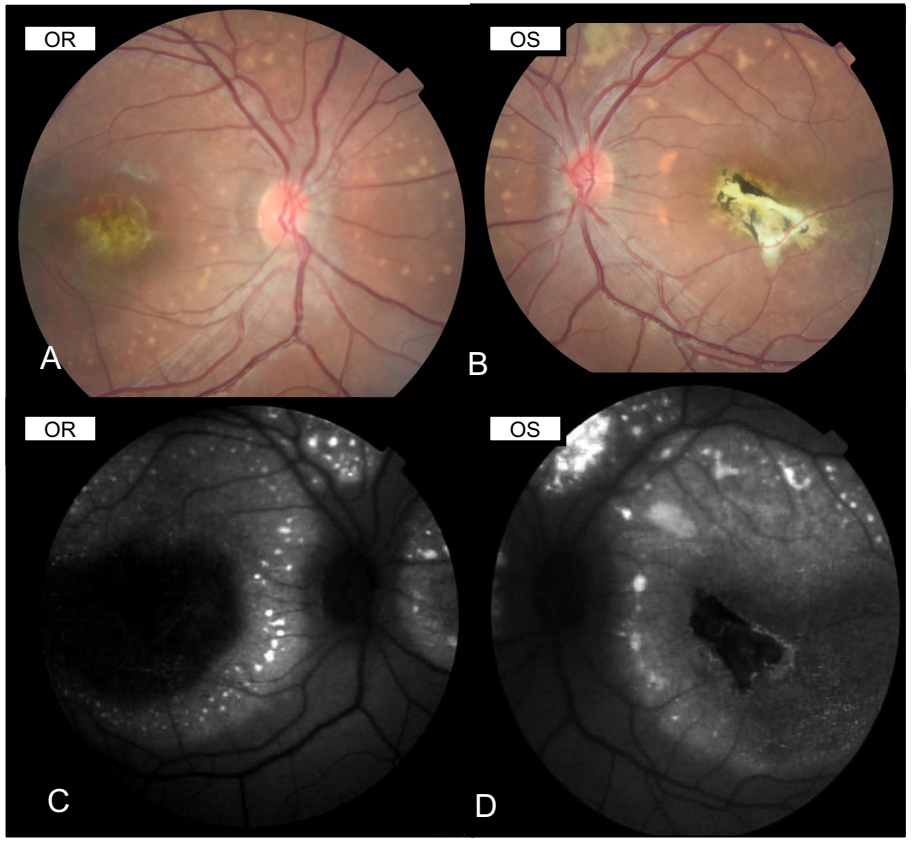
CASE REPORT



- Female patient, 18 years old, admitted to the Altino Ventura Foundation complaining of low visual acuity in both eyes since childhood, partially resolved with the use of glasses, but with progressive worsening. Her family history and personal history were unremarkable.

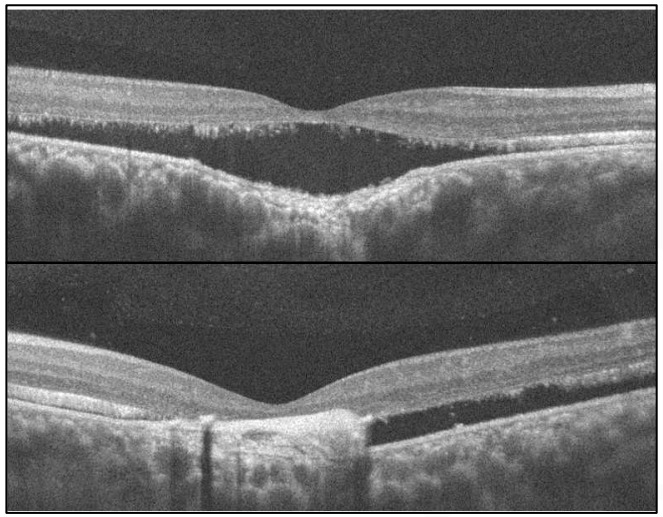
- Best corrected visual acuity (BCVA): 20/200 in the right eye (OR) and 20/100 in the left eye (OS). Similar fundoscopy in both eyes showing yellowish subretinal deposits and widespread, shallow subretinal fluid, with peripapillary sparing. The left eye also showed a yellowish subretinal lesion with mobilization and pigment at the edges (subretinal neovascular membrane appearance-SRNVM) in the foveal region measuring around 2 disc diameters.

Figure 1



A: Retinography of OR; **B:** Retinography of OS; **C:** Autofluorescense retinography of OR; **D:** Autofluorescense retinography of OS

Figure 2



A: OCT- B-Scan of OR; **B:** OCT- B-Scan of OS

- Retinography was performed (Figure 1 A and B), Optovue® optical coherence tomography (OCT- B-Scan) in AO, OCT of both eyes showing the presence of subretinal fluid, in OD there is focal excavation of the choroid and in OE subretinal hyperchromic material suggestive of SRNVM (Figure 2 A and B).
- The Autofluorescense retinography of both eyes shows multiple subretinal points of hyperautofluorescence in the topography of the vascular arcades and a region of hypoautofluorescence in the macula, with peripapillary sparing (Figure 1 C and D).

CASE REPORT

- A loading dose of Intravitreal aflibercept injection in both eyes was performed, which resulted in a partial reduction in subretinal fluid but no improvement in BCVA.

- In view of the multimodal study of the imaging exams, the main hypothesis is Recessive Bestrophinopathy and electro-oculogram, electroretinogram and genetic panel were requested to complement the diagnostic investigation.

DISCUSSION

ARB is characterized by multifocal yellowish subretinal deposits, choroidal neovascularizations and shallow subretinal fluid, probably resulting from loss of retinal adhesion to the RPE, [1,2,3] common features in both eyes of the patient reported. Birtel et al. highlighted in their article that peripapillary preservation in fundus autofluorescence imaging is a consistent and characteristic finding in patients with ARB [3], which was also observed in the present report. Multimodal retinal imaging can guide doctors towards a diagnosis and consider genetic testing, a tool that is not yet widely available in the public health system.

References

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