

OPHTHALMOLOGICAL ALTERATIONS IN A PATIENT WITH ALPORT SYNDROME – CASE REPORT

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47º BRAVS Meeting/ Retina 2023

April 28th to May 1st

PURPOSE

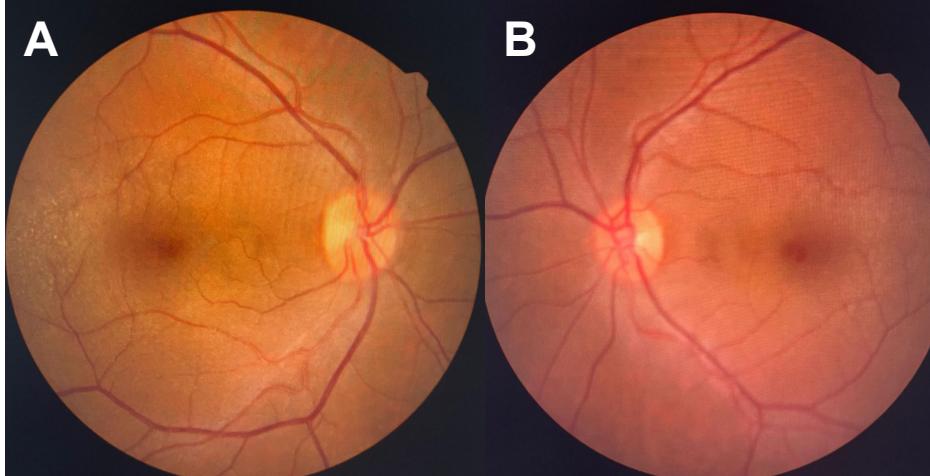
- To report a case of ocular lesions in a patient with Alport syndrome.

CASE REPORT

- Male patient, 34 years old;
- Hypertensive;
- Kidney transplantation seven years ago and hypoacusis;
- Low visual acuity (VA) in both eyes (OU) for years;
- Ophthalmological examination: OU better visual acuity (BCVA) = 20/40;
- Biomicroscopy: alteration of the lens with anterior lenticone and oil drop sign.

CASE REPORT

➤ Retinography



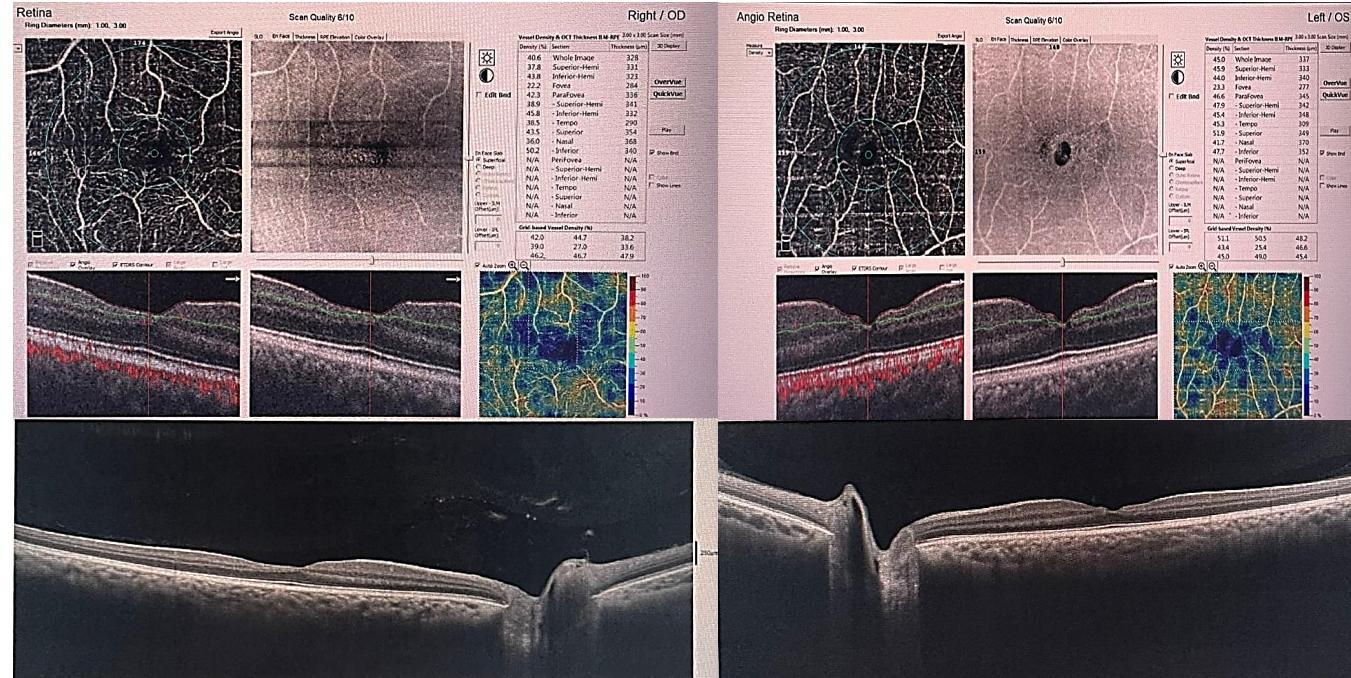
Right (A) and left eye (B)

➤ Retinal mapping:

- Optic disc and physiological papillary cupping;
- Arteriolar attenuation with increased dorsal arteriolar reflex;
- Altered foveal contour;
- Retinal spots in the mid-periphery.

CASE REPORT

➤ The optical coherence tomography (OCT) of the macula



Right eye (A - B) left eye (B - D).

- Thinning of the temporal retina and a fingerprint sign in some radial sections in OU.
- OCT with angiography (OCTA): increase in FAZ.
- Diagnosis: Alport's Syndrome.

DISCUSSION

- The case showed that Alport syndrome could often present with significant ocular alterations, highlighting the importance of ophthalmological evaluation for an early diagnosis.

REFERENCE

- 1.Ghadiri NJ, Stanojcic N, Raja M, Burton BJ. A triad of retinal signs in Alport syndrome: The 'stair-case' fovea, choroidal thinning and peripheral schisis. Eur J Ophthalmol. 2019 Jul;29(1_suppl):10-14.
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- 3.Cicinelli MV, Ritter M, Ghossein C, Aschauer C, Laccone F, Nagel M, Schmidt-Erfurth UM, Jampol LM, Gill MK. The spectrum of internal limiting membrane disease in Alport syndrome: A Multimodal Imaging Study. Retina. 2022 Feb 1;42(2):274-282.