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Multimodal imaging of adult vitelliform foveomacular dystrophy (case report)

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Abstract: *Adult foveomacular vitelliform dystrophy belongs to the heterogeneous group of Pattern Dystrophies. Multimodal imaging is therefore essential for the diagnosis and follow-up of this condition.*

Keywords: Ophtalmology, retina, foveomacular dystrophy

Introduction
Adult-onset foveomacular vitelliform dystrophy (AOFVD) is a rare, slowly progressive, bilateral and symmetrical macular disorder responsible for visual blurring with mild metamorphopsia.[1] Although inherited in an autosomal dominant pattern with variable expression and incomplete penetrance, AOFVD can also be sporadic with no evidence of familial transmission.

Case report:
We report the case of an 82-year-old female patient with a history of well-balanced type 2 diabetes, no family history of retinal disease, who presented with decreased visual acuity with progressive bilateral metamorphopsia.

Ophthalmological examination of the right eye revealed a visual acuity (MAVC) of 0.6 P4, a cortico-nuclear cataract, and a rounded yellowish macular lesion 1/3 papillary diameter on the fundus (fig1). Examination of the left eye revealed a visual acuity (MAVC) of 0.5 P4, a cortico-nuclear cataract, and a macular lesion similar to that found in the right eye. These macular lesions are hyperautofluorescent (fig2). On macular OCT (fig3), there is a voluminous deposit of bilateral vitelliform material between the retinal pigment epithelium and the photoreceptor layer, with loss of the foveolar funnel. These lesions are hypofluorescent early on, surrounded by a hyperfluorescent ring, and become progressively impregnated later on (fig4); early and late hypocyanescence is observed on ICG (fig5). Color vision and EOG are normal.

Discussion :
First described by Gass in 1974, vitelliform foveomacular dystrophy is a condition that is generally asymptomatic before the age of 50, and is usually discovered by chance on routine fundus examination [2]. Clinically, it manifests as mild metamorphopsia with generally preserved visual acuity, with a symmetrical, round or oval, bilateral yellowish central macular lesion one-third to one-half of a papillary diameter in fundus examination. On OCT, it is characterized by a dome-shaped deposit of central sub-retinal material between the photoreceptor layer and the retinal pigment epithelium (RPE). These vitelliform lesions are hyperautofluorescent, and hypofluorescent initially with late impregnation. When performed, the ICG shows a central

hypocyanescent image, helping to rule out a possible neovascular complication in AOFVD. The electro-oculogram and global electroretinogram are generally normal. At present, there is no treatment available for AOFVD, and patients present with preserved vision with a slowly progressive decline. However, the evolution can be dramatically unfavorable towards photoreceptor atrophy or choroidal neovascularization.

Conclusion :

Adult foveomacular vitelliform dystrophy belongs to the heterogeneous group of Pattern Dystrophies. Multimodal imaging is therefore essential for the diagnosis and follow-up of this condition.

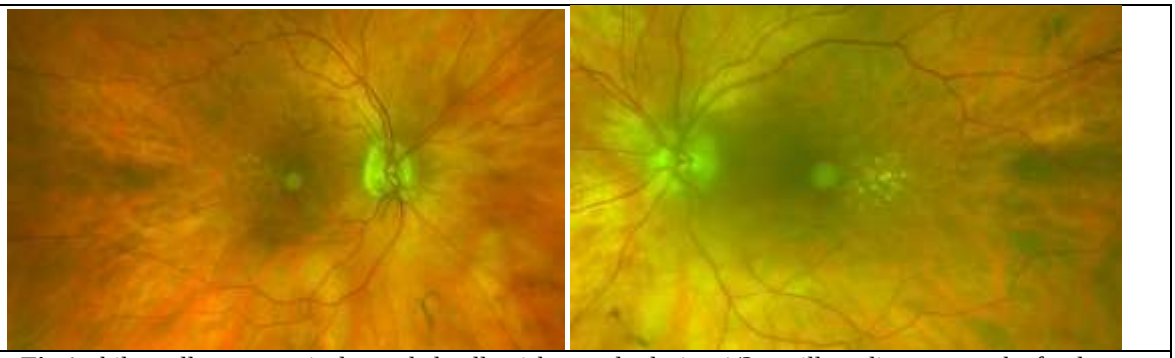

Fig 1 : bilaterally symmetrical rounded yellowish macular lesion 1/3 papillary diameter on the fundus

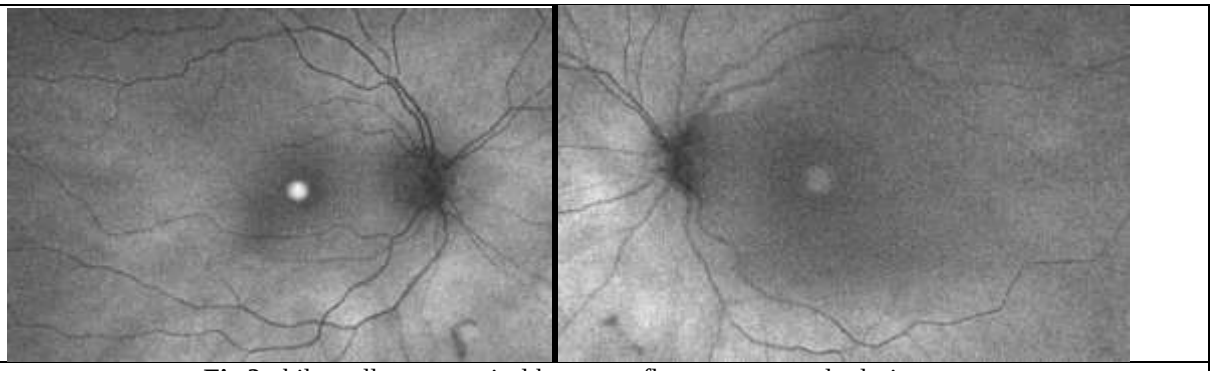

Fig 2 : bilaterally symmetrical hyperautofluorescent macular lesions

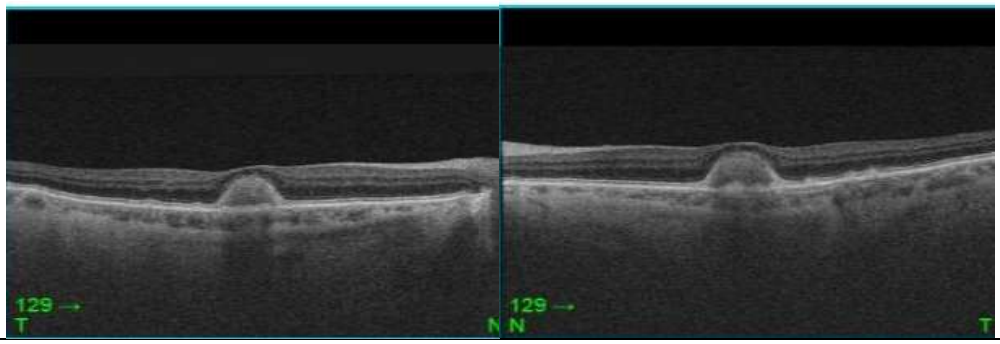

Fig 3 : B-Scan macular OCT showing a voluminous deposit of bilateral vitelliform material located between the retinal pigment epithelium and the photoreceptor layer, with loss of the foveolar funnel

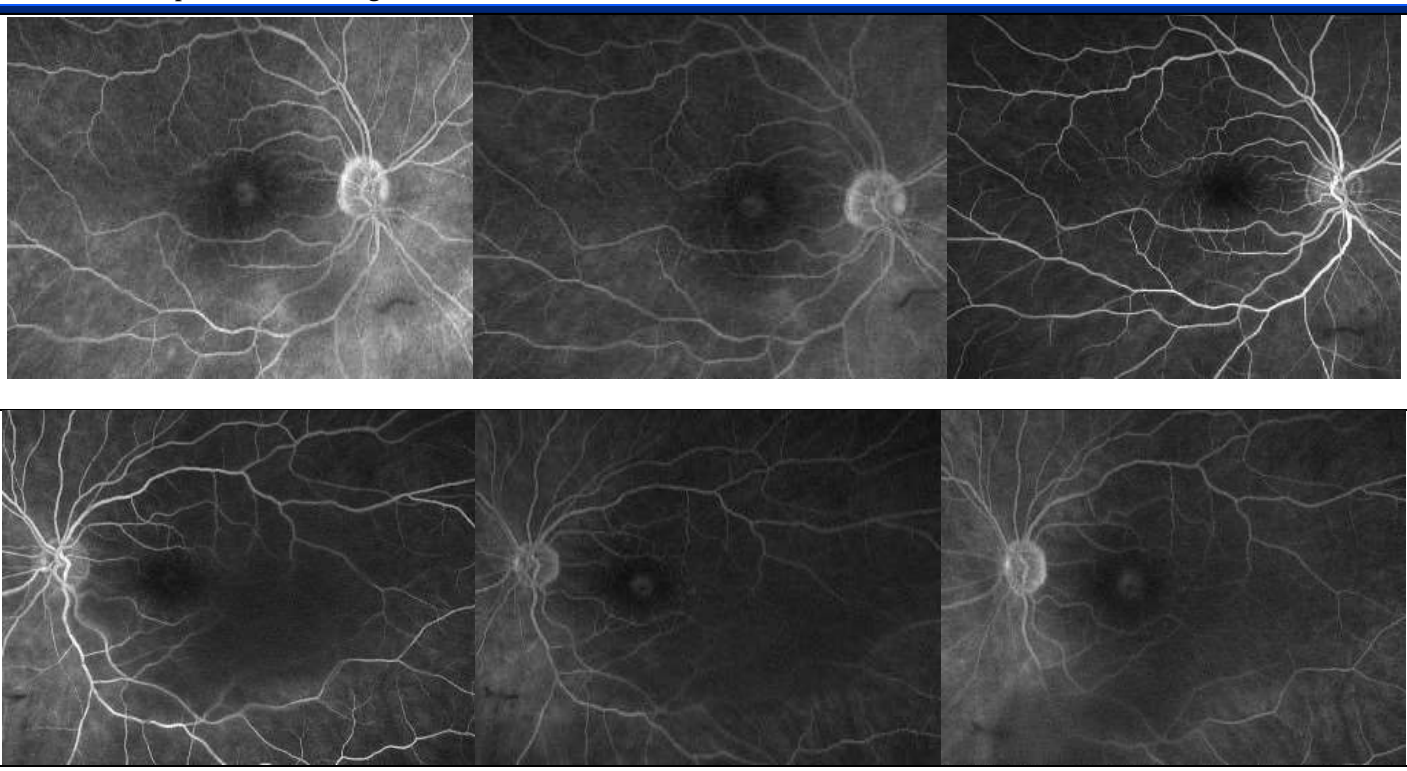

Fig 4 : Fluo Angio showing an initially hypofluorescent bilateral and symmetrical macular lesion, surrounded by a hyperfluorescent ring, with progressive impregnation at late time.

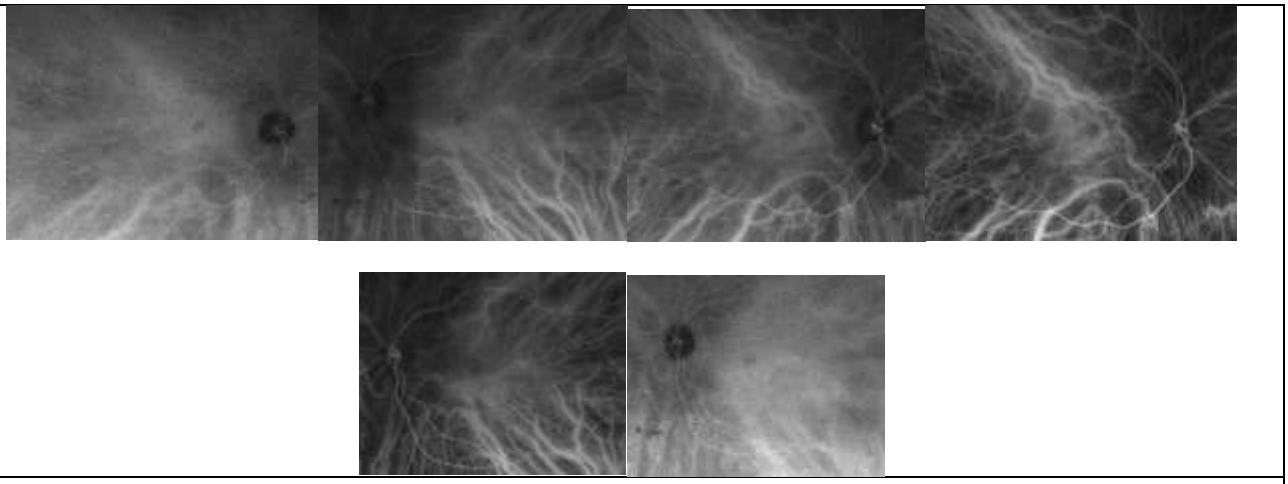

Fig 5 : Angio ICG sequences showing a bilateral, symmetrical hypocyanescent macular lesion at early and late times.

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