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An Unusual Incidental Finding of Drusen in a 32-year-old Woman: A Case Report and Brief Review of Literature

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

The different appearances of drusen in young subjects could be explained by a different structure and origin. These conditions are closely related to AMD, but their relationship has not been clearly established. We describe cuticular drusen, colloid drusen and dominant drusen .We report a case of macular colloid drusen in a 32 years old woman , incidentally finded , with no complications ; an ophthalmological workup was performed. There was no intervention required and the patient will be monitored for complications. Generally, patients are managed conservatively with observation, unless choroidal neovascularization develops. In this case, anti-VEGF injections have been shown to improve vision and resolve subretinal fluid. Another treatment option is the use of lasers to eliminate the sub-retinal fluid.

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1. INTRODUCTION

Drusen are small, yellowish-white deposits visible at the back of the eye. Predominantly lipid-based, they are extracellular deposits accumulated between the pigment epithelium and the collagen layer of the bruch's membrane. They usually appear after the age of 50 and represent the early stages of age-related macular degeneration (AMD) or age-related maculopathy. In some cases, drusen may be present before the age of 50.

2. CASE PRESENTATION

We report the case of a 32-year-old woman, radiology technician, thyroidectomized on levothyroxine, anemic on iron supplementation. No clinically relevant personal history was reported; no risk factors such as smoking, dyslipidaemia, obesity, and hypertension were reported. There was a family history of AMD (maternal grandmother). At presentation, the patient was asymptomatic; routine examination revealed preserved visual acuity with BCVA of 20/20 OD and 20/20 OS; emmetropia. A biomicroscopy examination revealed a normal anterior segment. On examination of the ocular fundus, large bilateral macular drusen with temporal predominance were found, and a retinal imaging study was requested.

Color fundus photography and autofluorescence images are presented in Fig. 1. SD-OCT demonstrated large drusen in the temporal macula bilaterally, with no changes within the retina outside the foveal area beside showing the characteristic sawtooth pattern and donut aspect (Fig. 2).

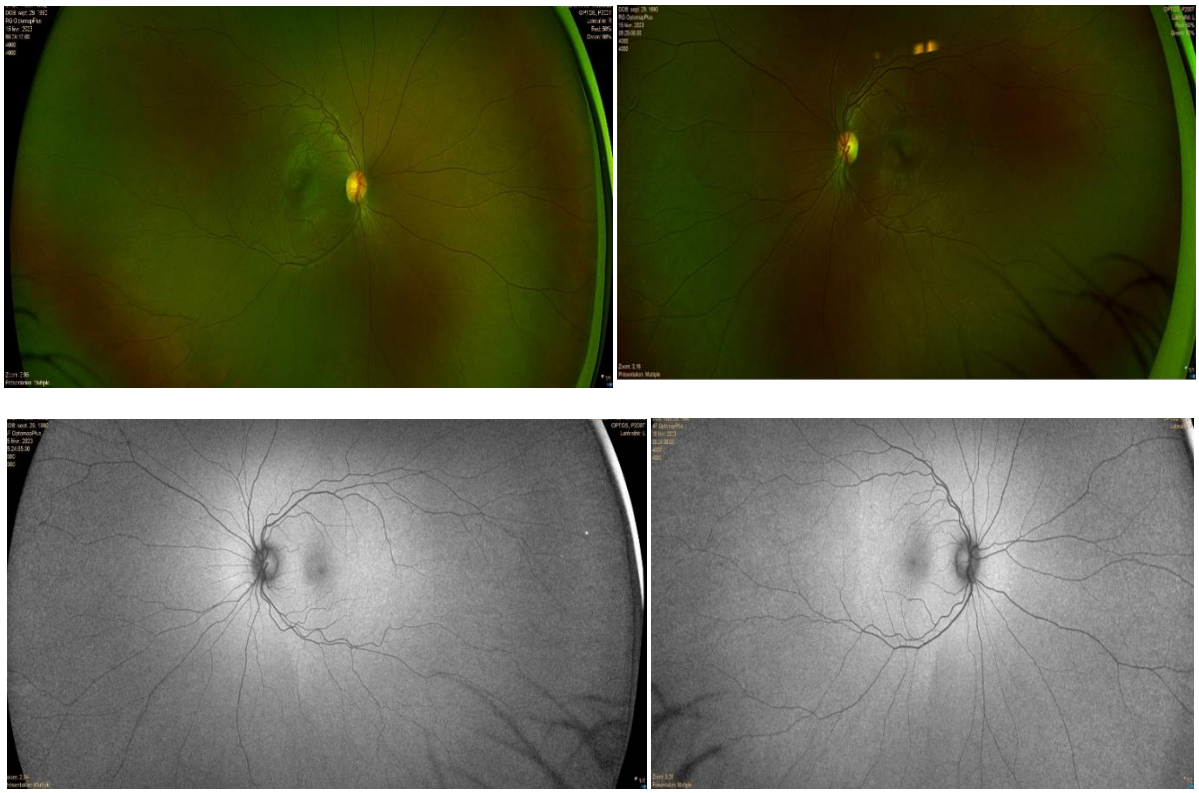

Fig. 1. Color fundus and autofluorescent photography images for left and right eyes at presentation

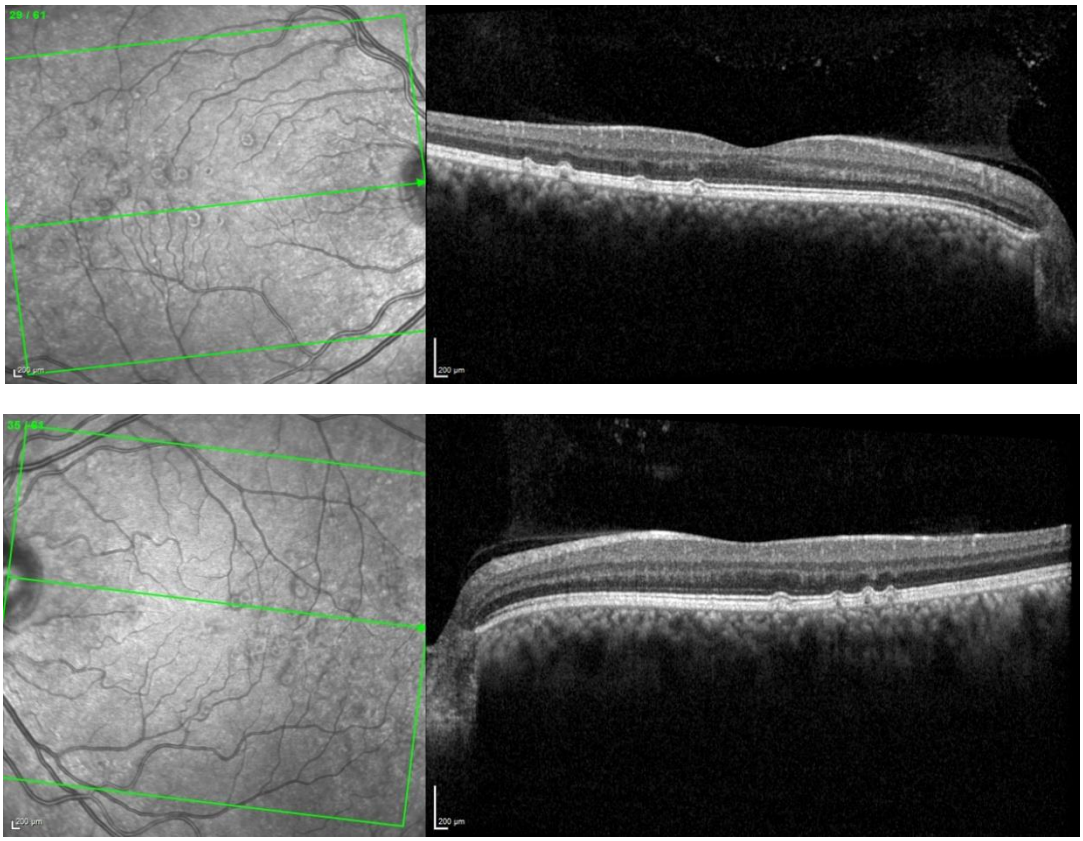

Fig. 2. SD-OCT demonstrated large drusen in the temporal macula bilaterally, with no changes within the retina outside the foveal area

An ophthalmological workup was performed. There was no intervention required and the patient will be monitored for complications.

3. DISCUSSION

Early onset drusen is defined as the presence of drusen in patients younger than the age of 50. The age of 50 is chose because the presence of drusen or other features suggestive of age related macula degeneration in patients younger than 50 is suggestive of inherited macula dystrophy. Many of these patients may have a family history of early onset drusen [1].

Small hard macular drusen can be observed in the retina of young adults; in a British study , we sought to describe the in vivo topography and geometry of these drusen. High lateral resolution imaging of small lobular hard retinal drusen suggested formation through the confluence of two or more smaller round lesions. The outline and size of these smaller lesions corresponded to 1-4 retinal pigment epithelium cells. The conclusion was that prospective longitudinal studies are needed to determine the ultimate fate of small hard drusen and their potential relation to age-related macular degeneration [2].

Drusen in the young subject mainly include: cuticular drusen, dominant drusen and colloid drusen.

A prospective descriptive study including 39 patients (27 women and 12 men) was carried out at the Créteil Hospital in France. The aim of the study was to analyze the angiographic and optical coherence tomography (OCT) features of drusen in subjects under 50 years of age. All patients underwent a complete ophthalmological work-up with fluorescein (AgF) and indocyanine green (ICG) angiography and OCT [3]. The mean age of the patients was 44 years. 27 patients had cuticular drusen, 5 patients had dominant drusen and 7 patients had juvenile colloid drusen.

Colloid drusen are large and diffuse, with no known specific genetic mutation like shown in our patient. They are preferentially temporo-macular, with a hyporeflective center that is discreetly hyperautofluorescent (AF). They are hypo- then hyper-fluorescent on fluorescein angiography,

and their "donut" appearance on ICG is original. They are generally benign, our patient was asymtomatic, but rare cases of atrophy, choroidal neovessels and chronic polypoidal vasculopathy have been described therefore a follow up is necessary. There is one reported association between loss-of-function complement factor H variants and large colloid drusen. Otherwise, little is know about the heritability and the genetic basis of large colloid drusen [4].

Cuticular drusen are fine, punctate and diffuse, often associated with an accumulation of vitelline material. The AgF appearance is typical, showing early starry-sky hyperfluorescence. On oct (optical coherence tomography), a sawtooth-like appearance is found. It is a borderline form of AMD, occurring at a younger age, with a stronger genetic component [5].

Doyne Honeycomb retinal dystrophy or Malattia Leventinese is a monogenic dystrophy with autosomal dominant inheritance (EFEMP1 mutation), resulting in a pathological fibulin 3 protein that induces local thickening of the bruchial membrane [6]. These drusen are organized in honeycomb lines radiating from the center, with a macular and peripapillary arrangement. They are hyperautofluorescent and hyperfluorescent at late times on AgF and ICG. They may become confluent and evolve into a central fibrous plaque [7]. Another study reports that patients with DSJ show a decrease in macular sensitivity compared with healthy controls, assessed by microperimetry, with preservation of visual acuity. Focal loss of sensitivity correlated with structural abnormalities demonstrated on SD-OCT. Currently, there are no genetic or targeted therapies to correct the underlying EFEMP1 genetic mutation in DHRD. As a rule, patients are managed conservatively with observation, unless choroidal neovascularization develops. In this case, anti-VEGF injections such as bevacizumab have been shown to improve vision and resolve subretinal fluid. Another treatment option is the use of lasers to eliminate the sub-retinal fluid. One study showed that low-energy argon laser treatment improved visual acuity and retinal sensitivity, and reduced drusen volume [8].

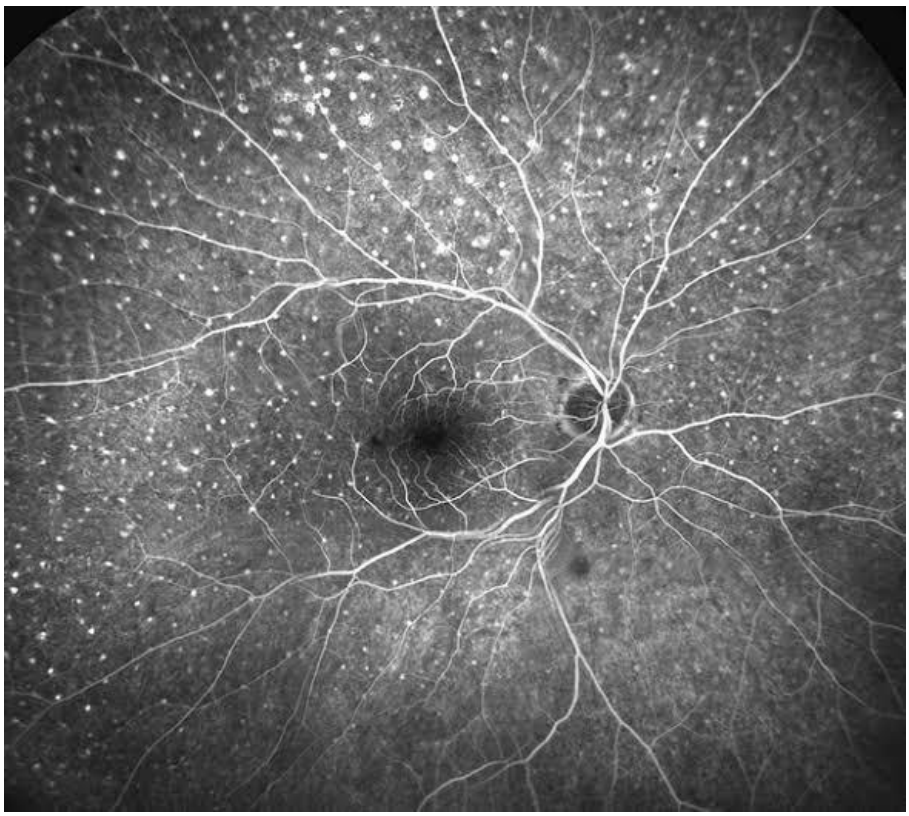

Fig. 3. On fluorescein angiography, cuticular drusen appear as a large group of small hyperfluorescent lesions scattered throughout the fundus, resulting in the “stars-in-the-sky” pattern that is characteristic of these drusen

4. CONCLUSION

The different appearances of drusen in young subjects could be explained by a different structure and origin [9]. These conditions are closely related to AMD, but their relationship has not been clearly established. Further work is needed to investigate the relationship between these different conditions. In all cases, the fundus of these patients should be regularly monitored for complications, in particular the appearance of choroidal neovessels.

CONSENT

As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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