What if drusenwere not exclusive to the elderly? A case report and literraturereview

Comment [SC1]: RemoveWhat

Comment [SC2]: Lierature

Abstract

The differentappearances of drusen in youngsubjectscouldbeexplained by a different structure and origin. These conditions are closelyrelated to AMD, but theirrelationship has not been clearlyestablished.

Comment [SC3]: Demonstrated

Keywords : epithelium, levothyroxine, maculardrusen, anterior segment

Comment [SC4]: First Letter in Caps

Introduction

Drusen are small, yellowish-white deposits visible at the back of the eye. Predominantlylipid-based, they are extracellulardepositsaccumulatedbetween the pigment epithelium and the collagen layer of the bruchial membrane. Theyusuallyappearafter the age of 50 and represent the early stages of age-relatedmaculardegeneration (AMD) or age-relatedmaculopathy. In some cases, drusenmaybepresentbefore the age of 50.We report the case of a 32-year-old woman, radiologytechnician, thyroidectomized on levothyroxine, anemic on ironsupplementation. There was a familyhistory of AMD (maternalgrandmother). Routine examinationrevealedpreservedvisualacuity of 10/10 ODG, emmetropia, no particularities on examination of the anterior segment, and an incidental finding of diffuse maculardrusen on the fundus. Colloiddrusen are large and diffuse, with no knownspecificgenetic mutation. They are preferentially temporo-macular, with a hyporeflective center thatisdiscreetlyhyperautofluorescent (AF).

Material and methods

We report the case of a 32-year-old woman, radiologytechnician, thyroidectomized on levothyroxine, anemic on ironsupplementation. There was a familyhistory of AMD (maternalgrandmother). Routine examinationrevealedpreservedvisualacuity of 10/10 ODG, emmetropia, no particularities on examination of the anterior segment, and an incidental finding of diffuse maculardrusen on the fundus.

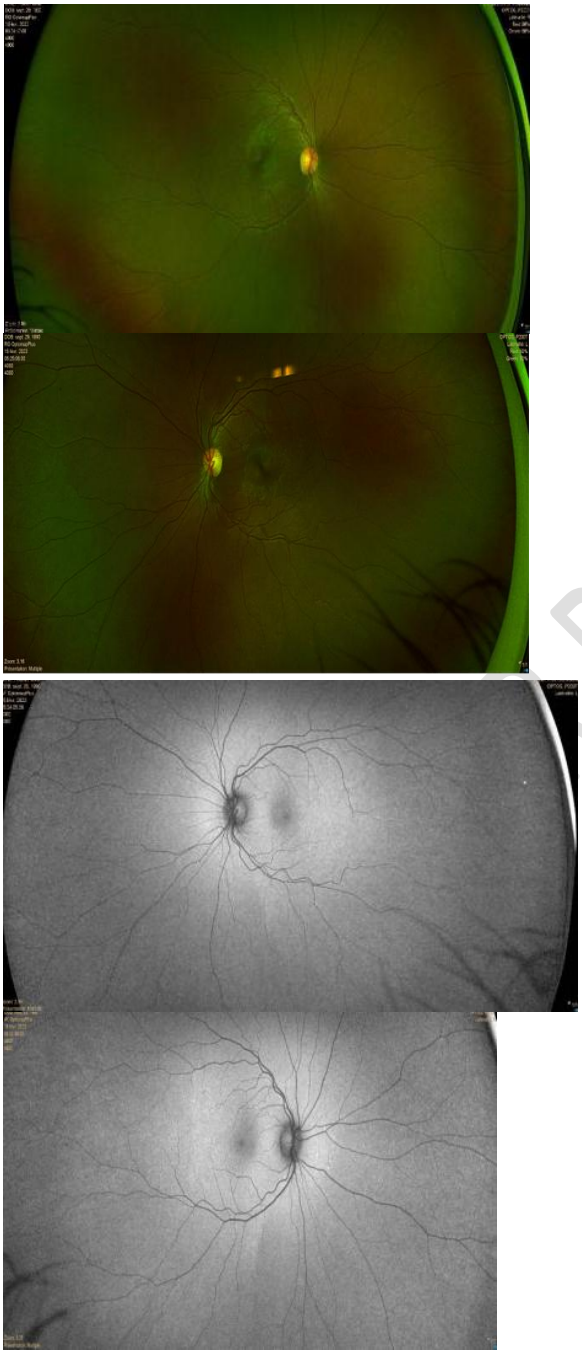

Figure 1 : funduscopical and autoflurescence aspect of colloiddrusen

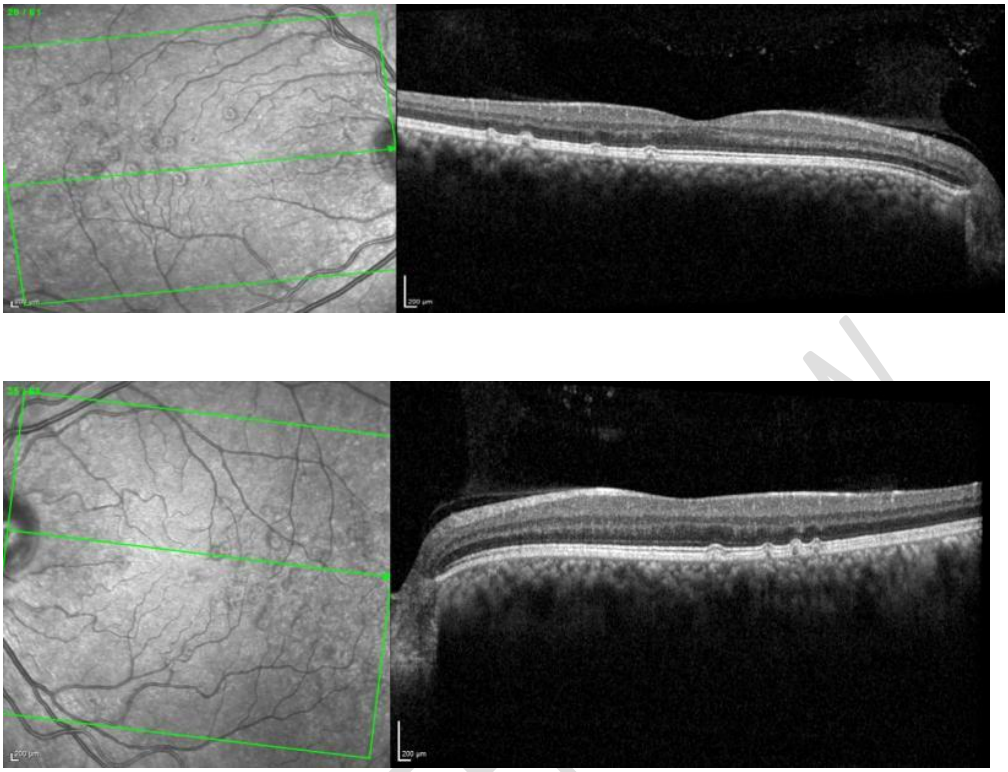

Figure 2 : oct aspect and « donut aspect » of colloiddrusen

Results

An ophthalmologicalworkupwasperformed. Our attitude consisted in a therapeutic abstention given the normality of the examination and the absence of complications with monitoring.

Discussion

Drusen in the youngsubjectmainlyinclude:cuticulardrusen, dominant drusen and colloiddrusen.

A prospective descriptive studyincluding 39 patients (27 women and 12 men) wascarried out at the Créteil Hospital in France . The aim of the studywas to analyze the angiographic and opticalcoherencetomography (OCT) features of drusen in subjectsunder 50 years of age. All patients underwent a completeophthalmologicalwork-up withfluorescein (AgF) and indocyanine green (ICG) angiography and OCT (1). The meanage of the patients was 44 years. 27 patients hadcuticulardrusen, 5 patients had dominant drusen and 7 patients hadjuvenilecolloiddrusen.

Colloiddrusen are large and diffuse, with no knownspecificgenetic mutation. They are preferentially temporo-macular, with a hyporeflective center thatisdiscreetlyhyperautofluorescent (AF). They are hypo- then hyper-fluorescent on fluoresceinangiography, and their "donut" appearance on ICG is original. They are generallybenign, but rare cases of atrophy, choroidalneovessels and chronicpolypoidalvasculopathy have been described.

Cuticulardrusen are fine, punctate and diffuse, oftenassociatedwith an accumulation of vitelline material. The AgFappearanceistypical, showingearlystarry-sky hyperfluorescence. On oct (opticalcoherencetomography), a sawtooth-like appearanceisfound. It is a borderline form of AMD, occurring at a youngerage, with a strongergeneticcomponent(2).

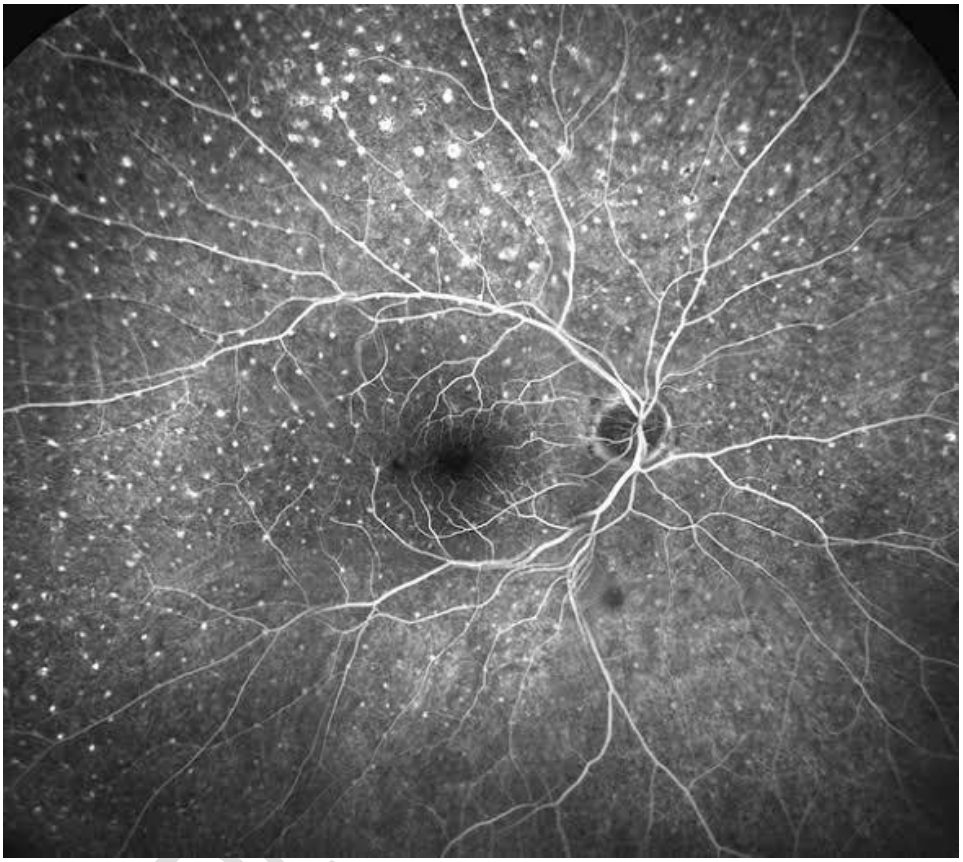

Figure 3 : On fluoresceinangiography, cuticulardrusenappear as a large group of smallhyperfluorescentlesionsscatteredthroughout the fundus, resulting in the “stars-in-the-sky” pattern thatischaracteristic of thesedrusen

DoyneHoneycombretinaldystrophy or MalattiaLeventineseis a monogenicdystrophywith autosomal dominant inheritance (EFEMP1 mutation), resulting in a pathologicalfibulin 3 proteinthatinduces local thickening of the bruchialmembrane(3). Thesedrusen are organized in honeycomblinesradiatingfrom the center, with a macular and peripapillary arrangement. They are hyperautofluorescent and hyperfluorescent at late times on AgF and ICG. Theymaybecome confluent and evolveinto a central fibrousplaque(4). Anotherstudy reports that patients with DSJ show a decrease in macularsensitivitycomparedwithhealthycontrols, assessed by microperimetry, withpreservation of visualacuity. Focal loss of sensitivitycorrelatedwith structural abnormalitiesdemonstrated on SD-OCT. Currently, there are no genetic or targetedtherapies to correct the underlying EFEMP1 genetic mutation in DHRD. As a rule, patients are managedconservativelywith observation, unlesschoroidalneovascularizationdevelops. In this case, anti-VEGF injections such as bevacizumab have been shown to improve vision and resolvesubretinalfluid. Anothertreatmentoption is the use of

lasers to eliminate the sub-retinalfluid.One studyshowedthatlow-energy argon laser treatmentimprovedvisualacuity and retinalsensitivity, and reduceddrusenvolume(5).

Conclusion

The differentappearances of drusen in youngsubjectscouldbeexplained by a different structure and origin. These conditions are closelyrelated to AMD, but theirrelationship has not been clearlyestablished. Furtherworkisneeded to investigate the relationshipbetweenthesedifferent conditions. In all cases, the fundus of these patients shouldberegularlymonitored for complications, in particular the appearance of choroidalneovessels.

Références

1 Benjamin Guigui, Nicolas Leveziel, Virginie Martinet, Nathalie Massamba, Margaret Sterkers, et al..

Angiographyfeatures of earlyonsetdrusen. British Journal of Ophthalmology, 2010, 95 (2), pp.238.

ff10.1136/bjo.2009.178400ff.

2 Camiel J.F. Boon , Johannes P.H. van de Ven , Carel B. Hoyng , Anneke I. den Hollander a, B. Jeroen Klevering .Cuticulardrusen: Stars in the sky. Progress in Retinal and Eye Research ,Volume 37, November 2013, Pages 90-113

3 Stone, Edwin M., et al. "A single EFEMP1 mutation associatedwithbothMalattiaLeventinese and Doynehoneycombretinaldystrophy." Nature genetics 22.2 (1999): 199-202.

4 Sohn, Elliott H., et al. "Responsiveness of ChoroidalNeovascular Membranes in Patients With R345W Mutation in Fibulin 3 (DoyneHoneycombRetinalDystrophy) to Anti–VascularEndothelialGrowth Factor Therapy." Archives of ophthalmology 129.12 (2011): 1626-1628.

5 Cusumano A, Falsini B, Giardina E, Cascella R, Sebastiani J, Marshall J. Doynehoneycombretinaldystrophy - functionalimprovementfollowingsubthresholdnanopulse laser treatment:a case report. J Med Case Rep. 2019 Jan 10;13(1):5.

Comment [SC5]: Add few more references