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Unilateral benign yellow dot maculopathy

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ABSTRACT

Purpose: To describe a unique case of unilateral benign yellow dot maculopathy.
Observations: A 25-year-man was evaluated after incidental finding of yellow dots in the right macula. The findings of examination and multimodal imaging were in keeping with a diagnosis of benign yellow dot maculopathy.
Conclusions and importance: Benign yellow dot maculopathy is a recently described entity with either a sporadic or dominant inheritance pattern. This is the first known report of the characteristic findings of this phenotype presenting unilaterally.

1. Introduction

Yellow macular dots have an extensive differential diagnosis including drusen, inherited maculopathies, and fleck retinopathy. Inherited macular dystrophies are a group of heterogenous disorders that lead to macular changes and vision loss.[1] Fleck retinopathy encompasses a large number of diseases with characteristic white-yellow retinal flecks. These two groups of disorders are typically associated with vision changes and possible systemic abnormalities.

Recently, a novel macular phenotype termed 'benign yellow dot maculopathy' has been described by Dev Borman et al.[2] To date, reports of this phenotype describe non-progressive bilateral macular yellow dots with normal visual acuity and color vision. Herein, we present the first known case of unilateral benign yellow dot maculopathy.

2. Case presentation

A 25-year-old Caucasian male of Dutch and mixed European ancestry was referred for assessment of white-yellow dots in the macula of his right eye (OD). He was asymptomatic. His past ocular history was significant for trauma to the left eye (OS) from a paintball approximately 10 years prior. This injury led to angle recession in the left eye, but no glaucoma. The patient was otherwise healthy and not taking any medications. There was no travel history or exposure to animals. The patient denied smoking and recreational drug use. There was no history of macular degeneration in the patient's family.

On presentation, best corrected visual acuity was 20/20 bilaterally and intraocular pressures were normal. Pupils were equal and reactive to light, and no afferent pupillary defect was present. Farnsworth D15 color vision testing was normal. Examination of the anterior segment was unremarkable OD and revealed a small traumatic cataract OS. In both eyes, there was no evidence of cell or flare in the anterior chamber and the vitreous was clear with no cell or haze. Optic nerves were normal and symmetric.

Retinal examination revealed intraretinal diffuse fine yellow dots in the posterior pole OD, predominantly located circumferentially around the fovea and also extending into the temporal macula (Fig. 1A). The mid and far periphery of the retina were normal with no evidence of the yellow dots. The retinal vessels appeared normal. Examination of the left retina was normal, with no yellow dots observed (Fig. 1B).

The scattered yellow dots that were appreciated clinically were hyperautofluorescent on fundus autofluorescence imaging (Fig. 2). Subtle abnormalities were noted on spectral-domain optical coherence tomography (OCT) of the macula OD at the level of the retinal pigment epithelium (RPE) (Heidelberg Engineering, Dossenheim, Germany) (Fig. 3A and B). The yellow dots appeared hyper-reflective on near-infrared imaging (Fig. 3C). There were no abnormalities appreciated on OCT OS. Macular OCT thickness maps appeared normal in both eyes,

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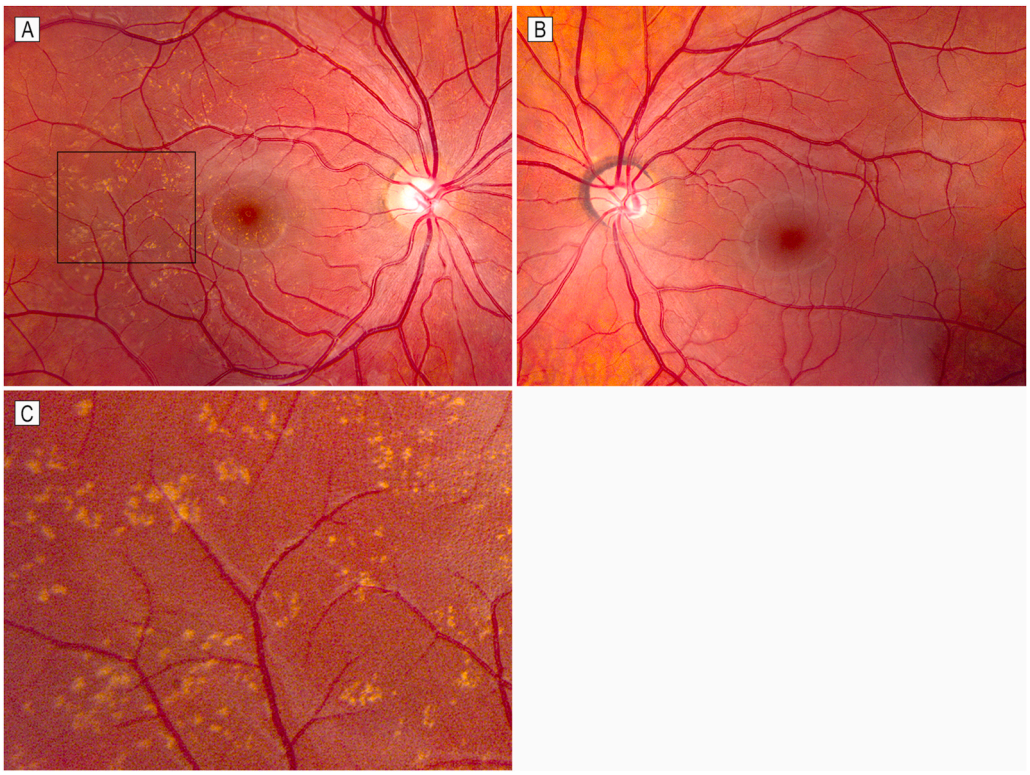

Fig. 1. Color fundus photo of the right eye (A) demonstrating fine discrete parafoveal yellow dots, which extend into the temporal macula. The left fundus is normal (B). 3.5× magnified image (C) of the yellow dots shows retinal vessels passing over the yellow dots. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

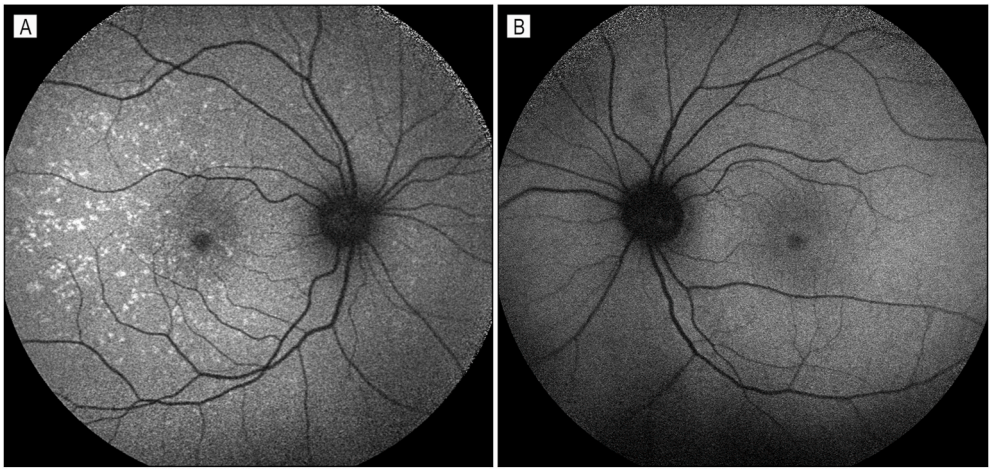

Fig. 2. Blue fundus autofluorescence (Heidelberg Spectralis, Dossenheim, Germany) shows hyperautofluorescent dots in the posterior pole of the right eye (A) and a normal left fundus (B).

with central subfield foveal average thickness measuring 285 μm OD and 273 μm OS. Goldmann kinetic perimetry showed no abnormalities in either eye. Electrophysiologic tests were also performed. The pattern visual evoked potentials were in the normal range with no significant interocular difference. Multifocal electroretinogram (ERG) revealed a slight decrease in peak signal intensity OD, although foveal activity with implicit times were in the normal range in both eyes (Fig. 4).

3. Discussion

Benign yellow dot maculopathy was first described in 2017 by Dev Borman et al.[2] In the 36 individuals (26 female) with this characteristic non-progressive macular phenotype, the majority presented in childhood with no associated visual disturbances or systemic associations. The inheritance pattern of benign yellow dot maculopathy was observed to be either sporadic or autosomal dominant, and 13 patients had a positive family history. Two of 19 tested patients were found to have ERG abnormalities, consisting of mild to moderate decrease in the ERG P50 component. Since the original description in 2017, two other reports have described similar findings.[3,4]

To date, all of the patients with benign yellow dot maculopathy have had bilateral macular findings. In contrast to these reports, our patient's presentation is compatible with unilateral benign yellow dot maculopathy OD. His visual acuity, color vision, and visual fields were normal.

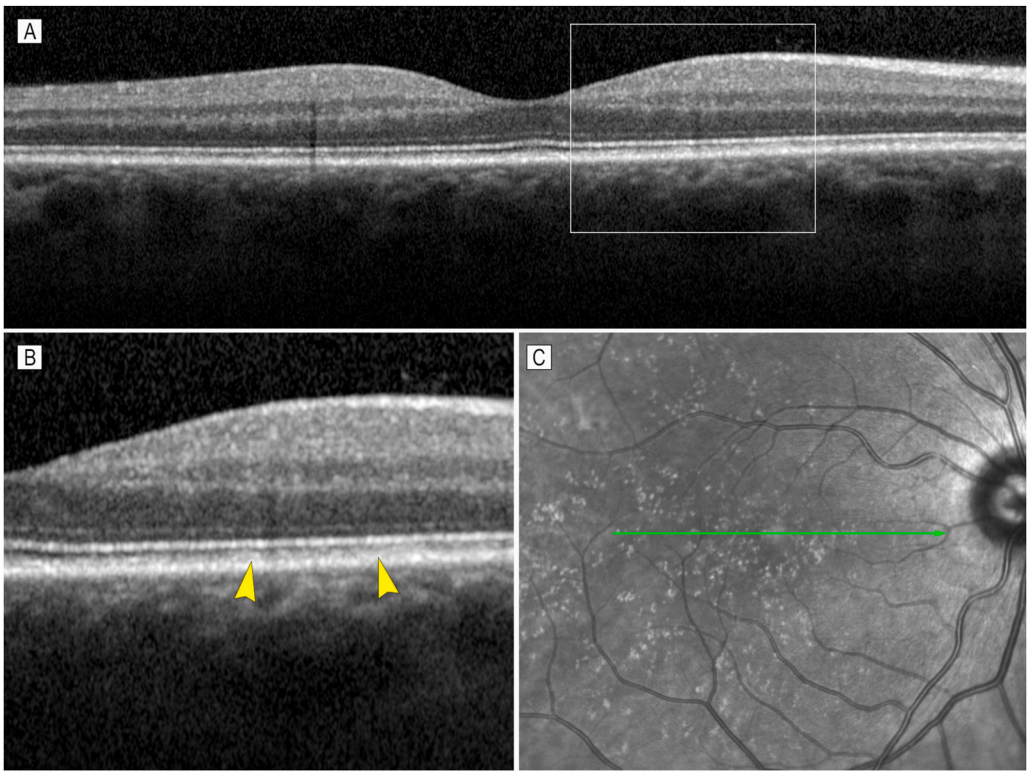

Fig. 3. Spectal-domain optical coherence tomography (Heidelberg Engineering, Dossenheim, Germany) section through yellow dots (A). 3.5× magnified image (B) demonstrates diffuse hyperreflective spots and slight retinal pigment epithelium irregularity (arrowheads). Near-infrared reflectance imaging (C) shows hyperreflective appearing dots in the macula. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

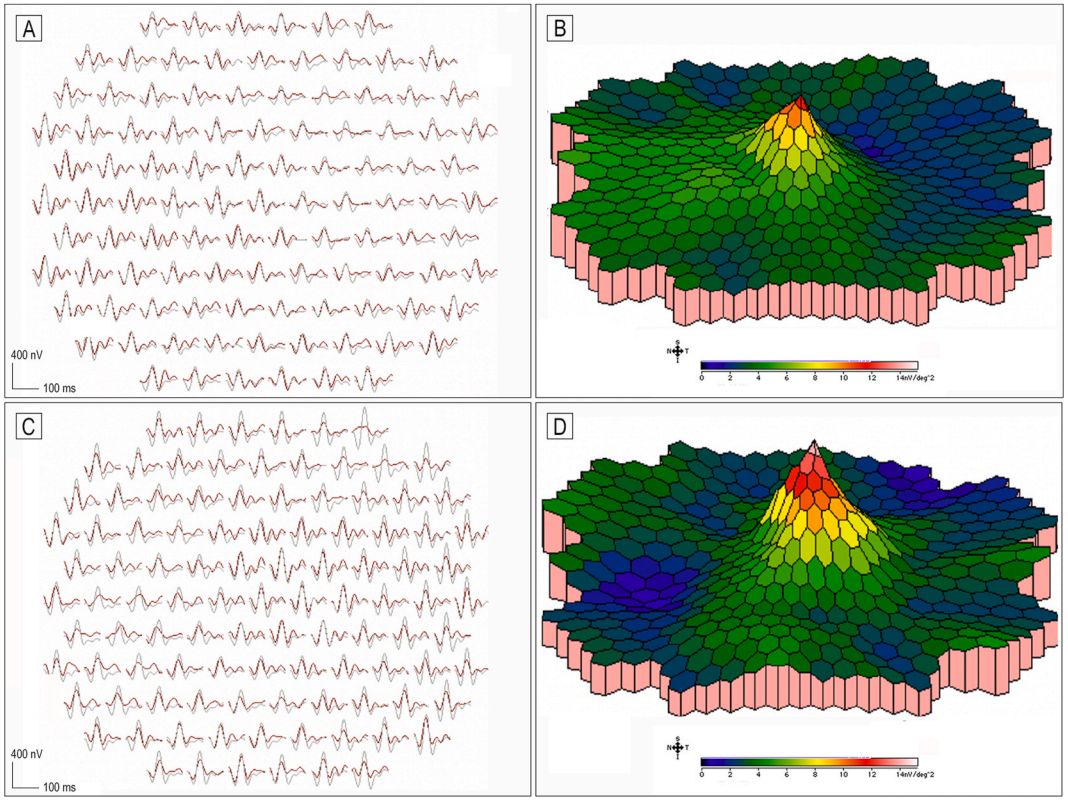

Fig. 4. Multifocal electroretinography results. Right affected eye trace array recordings (A) and 3-dimensional amplitude density plot (B) show slightly diminished peak signal intensity relative to the fellow eye but are within normal limits. Left trace array recordings (C) and 3-dimensional amplitude density plot (D) are within normal limits.

The ERG findings were within normal limits, although there was a slight decrease in the peak signal OD compared to OS. As in our patient, multimodal imaging in previously described patients has shown hyperautofluorescence of the yellow dots with fundus autofluorescence imaging.[3] Although normal in the majority of patients with benign yellow dot maculopathy, OCT imaging may demonstrate mild irregularities at the level of the RPE and ellipsoid zone corresponding to the yellow dots.[2,3] These OCT findings were noted OD in our patient.

Careful consideration was given to other etiologies of yellow dots including crystalline retinopathies, Gunn's dots, North Carolina macular dystrophy (NCMD), and familial drusen. Both the unilateral findings and lack of known exposure to causative agents, suggest against crystalline retinopathy. Gunn's dots are benign, small glistening dots on the surface of the retina. They are located near the optic disc at the level of the internal limiting membrane, which was not consistent with our case.[5,6] The diagnosis of NCMD dystrophy was also considered. NCMD is an inherited disease causing bilateral infantile macular degeneration.[7] NCMD can have a diverse clinical presentation ranging from drusen-like lesions in the central macula to disciform scars. Although the patient's immediate family was not available to be examined, the absence of a family history of macular disease further suggests against a diagnosis of NCMD, which is inherited in an autosomal dominant fashion with complete penetrance. Early onset drusen were also considered in the differential diagnosis; however, the lack of drusenoid deposits on OCT is not consistent with this diagnosis.

Limitations to this unique report include the absence of family members available for examination. As a male, the sex of our patient categorizes him in the minority of patients previously described with benign yellow dot maculopathy. Further, among other cases reported, the predominant distribution of yellow dots has been noted evenly around the fovea or concentrated in the nasal parafoveal region. While our patient had diffuse yellow dots around the fovea, additional dots were prominent in the temporal and superior macula. Until an etiology (e.g., genetic, biochemical, or infectious) is determined for benign yellow dot maculopathy, there remains the possibility that this is a heterogenous group of diseases with a similar phenotype.

To the best of our knowledge, this is the first report of benign yellow dot maculopathy presenting unilaterally. Increased awareness of this emerging entity can help clarify the diagnosis, prevent misattribution of these findings to other etiologies,[8] and provide patients and families with a reassuring prognosis.

Patient consent

Written consent to publish this case has not been obtained. This report does not contain any personal identifying information.

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Authorship

All listed authors meet the ICMJE criteria. We attest that all authors contributed significantly to the creation of this manuscript, each having fulfilled criteria as established by the ICMJE.

Intellectual property

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Research ethics

We further confirm that any aspect of the work covered in this manuscript that has involved human patients has been conducted with the ethical approval of all relevant bodies and that such approvals are acknowledged within the manuscript.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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