

Case report

Benign yellow dot maculopathy

Elad Moisseiev^{a,b,*}^a Department of Ophthalmology, Tel Aviv Medical Center, Tel Aviv, Israel^b Affiliated to the Sackler School of Medicine, Tel-Aviv University, Tel-Aviv, Israel

ARTICLE INFO

Keywords:

Yellow dot
Maculopathy
Phenotype
Familial

ABSTRACT

Purpose: To describe a new family with benign yellow dot maculopathy.

Observations: A young male patient was found to have bilateral multiple small yellow dots in both maculae. Visual acuity and color vision were normal, and no pathological findings were demonstrated on automated visual field, optical coherence tomography (OCT) and electrophysiological testing. Examination of his parents revealed similar findings in his mother, suggesting autosomal dominant inheritance. No deterioration of vision occurred over long term follow up. These findings are consistent with the newly described phenotype of benign yellow dot maculopathy.

Conclusions and importance: This is the first report of patients with benign yellow dot maculopathy since its original description, and the first to document it in a family of North African descent. This report will serve to raise awareness to this phenotype, which may be more common than currently known.

1. Introduction

The finding of yellow dots in the macula of young patients has a wide differential diagnosis, which includes inherited macular dystrophies, drug-induced retinopathies, metabolic disorders such as cystinosis and oxalosis, and conditions such as Bietti crystalline retinopathy, autosomal dominant drusen, Kjellin syndrome, and idiopathic juxtafoveal telangiectasis type 2.^{1,2} These are often associated with reduced central visual acuity (VA), are progressive and have typical additional systemic or ocular findings.

Recently, a new phenotype has been described, and termed “benign yellow dot maculopathy”.³ This phenotype consists of bilateral macular yellow dots, which is non-progressive and without visual impairment. The reported cases were familial, and their distribution was compatible with autosomal dominant inheritance. The purpose of this report is to describe a new family with this phenotype, and to raise awareness to this condition which may be more common than previously perceived.

2. Case report

A 24 year-old male was referred to our clinic due for evaluation of reduced VA. The patient had no previous systemic or ocular medical history, and did not use any systemic or topical medications. His complaint of decreased VA was described as transient and with no significant impact on his life for a few months prior to his presentation.

On initial examination, his best corrected VA was 20/25 in the right

eye and 20/20 in the left eye. Intraocular pressure (IOP) was 13 mmHg in both eyes, the range of ocular movements was full bilaterally, color vision was 10/10 in both eyes, and there was no relative afferent pupillary defect (RAPD). Both anterior segments were normal, and the lenses were clear. The fundus examination revealed multiple, small yellow dots in both maculae (Fig. 1A and B). The retinal periphery was normal in both eyes, with no yellow dots.

The patient had no history of travel, exposure to animals, insect bites, use of any illicit drugs or medications, and no other systemic or ocular complaints. There was also no history of any familial conditions and no members of his family had decreased VA.

A 30-2 Humphrey visual field and fluorescein angiography were normal (Fig. 1C). Optical coherence tomography (OCT) imaging was normal, with no irregularities of the retinal layers (Fig. 1D). In addition, complete blood count, blood chemistry, erythrocyte sedimentation rate, lipid profile, coagulation tests, and a complete collagenogram were normal. Electrophysiological testing was also normal bilaterally.

As an inherited condition was suspected, the patient's parents were also examined. Both were of Libyan descent, but were not related (no consanguinity). Both had no history of any systemic or ocular conditions, and had no visual complaints. The father had a VA of 20/20 OU, and his ocular exam was normal with no macular yellow dots. The mother also had a VA of 20/20 OU, with similar macular yellow dots noted bilaterally (Fig. 2A). OCT demonstrated mild asymptomatic vitreomacular traction in the right eye, but no abnormalities in the outer retinal layers in both eyes (Fig. 2B).

* Department of Ophthalmology, Tel Aviv Medical Center, Weitzman 6 St., Tel Aviv, 64239, Israel.

E-mail address: elad_moi@netvision.net.il.

<https://doi.org/10.1016/j.ajoc.2018.01.040>

Received 7 August 2017; Received in revised form 19 January 2018; Accepted 22 January 2018

Available online 28 January 2018

2451-9936/ © 2018 The Author. Published by Elsevier Inc. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

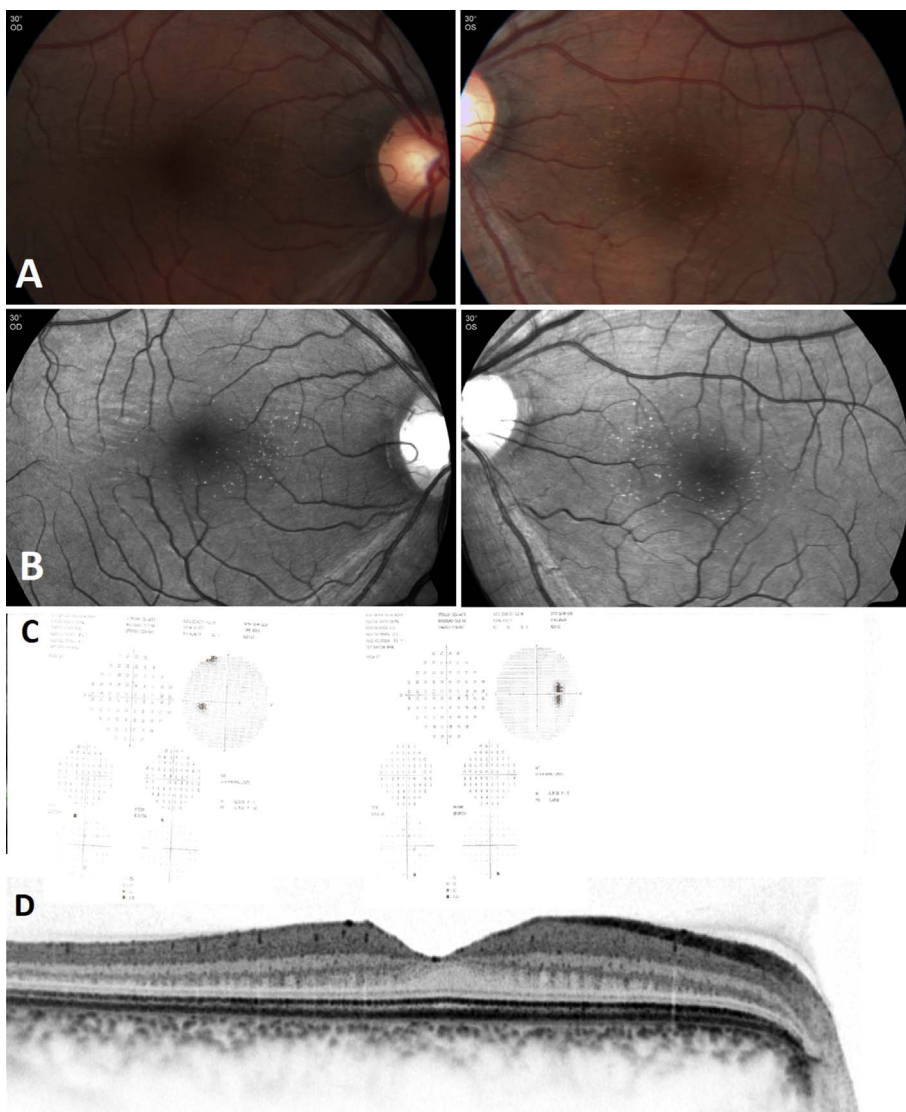


Fig. 1. Imaging of the patient. A. Color fundus photographs of both eyes of the patient. Note the macular distribution of multiple small yellow dots in both eyes. B. Red free photograph of both eyes of the patients, highlighting the macular dots. C. Visual fields of both eyes, which demonstrated no visual field defects. D. Optical coherence tomography (OCT) of the patient's right eye, which was normal. OCT of the left eye was also normal. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

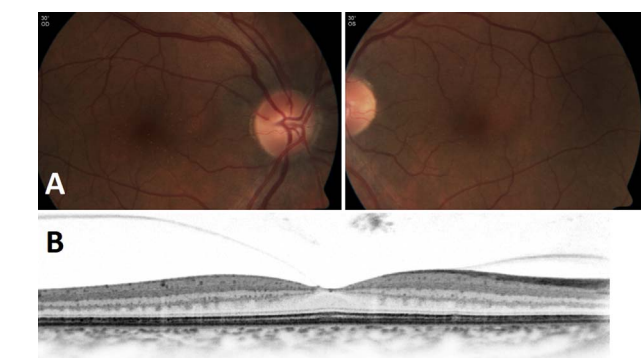


Fig. 2. Imaging of the patient's mother. A. Color fundus photographs of both eyes of the patient's mother. Note the similar macular distribution of multiple small yellow dots in both eyes. B. Optical coherence tomography (OCT) of the patient's mother's right eye, which demonstrated mild vitreomacular traction, but no abnormalities in the outer retinal layers. No abnormalities were noted in the OCT of the left eye. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

The patient and his mother were followed for 6 years, with no progression in the phenotypic appearance and no change in VA.

3. Discussion

Benign yellow dot maculopathy is a newly described phenotype. The recent study in which it was presented included 36 patients from 23 unrelated families.³ The macular findings of bilateral multiple yellow dots in most of these patients were discovered in routine eye examinations or because a family member has been examined. Only 3 (8.3%) of the patients were symptomatic, but had additional findings explaining their reduced VA, such as strabismus and amblyopia. In the majority of patients, VA was intact, remained stable over time. Color vision was also normal. In the majority of cases (30 patients, 83.3%) OCT imaging was normal, while in the others (6 patients, 16.7%) very mild irregularities in the inner segment ellipsoid zone were noted. Electrophysiologic testing was normal.

The distribution of the cases in familial clusters, affecting both males and females, suggested an autosomal dominant inheritance pattern. Whole exome analysis across several families excluded any mutations in known macular dystrophy genes, and haplotype sharing analysis excluded linkage with North Carolina macular dystrophy.

The patient we present, along with the findings discovered in his mother, are compatible with all of the characteristics of this new

phenotype. He was a young patient, with bilateral macular yellow dots compatible with those described in the original study.³ His VA and color vision were unaffected, there was no progression over 6 years of follow up, and he had no abnormalities in OCT and electrophysiologic testing. Similar findings were discovered in his mother's eyes, but not his father, consistent with autosomal dominant inheritance.

This is the first report of yellow dot maculopathy following the original study in which it was described.³ The families included in that study were mostly white, and none was reported to be of North African descent, as the family we report here. This may indicate that the prevalence of this phenotype is more common, and its relatively benign nature may be responsible for its previous lack of recognition and under-reporting. Further reporting of patients with this phenotype may shed more light on its prevalence, and perhaps future genetic studies may elucidate its cause.

Patient consent

Consent to publish the case report was obtained in writing. This report does not contain any personal information that could lead to the identification of the patients.

Acknowledgments and disclosures

Funding: No funding or grant support was received for this work.

Conflict of Interest: The author has no financial disclosures.

Authorship: All authors attest that they meet the current ICMJE criteria for Authorship.

Acknowledgements: None.

Appendix A. Supplementary data

Supplementary data related to this article can be found at <http://dx.doi.org/10.1016/j.ajoc.2018.01.040>.

References

1. Moore AT. Childhood macular dystrophies. *Curr Opin Ophthalmol*. 2009;20:363–368.
2. Drenser K, Sarraf D, Jain A, Small KW. Crystalline retinopathies. *Surv Ophthalmol*. 2006;51:535–549.
3. Dev Borman A, Rachitskaya A, Suzani M, et al. Benign yellow dot maculopathy: a new macular phenotype. *Ophthalmology*. 2017;124:1004–1013.