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Brief Report

Retinal pigment epithelium changes in Kartagener syndrome

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ABSTRACT

Purpose: We present the first case in the literature of a patient with Kartagener syndrome and ocular findings of nonexudative age-related macular degeneration.

Observations: A 55-year-old woman with Kartagener syndrome and chronic angle closure glaucoma presented for evaluation of the retina. Optos ultra-widefield imaging of the fundus showed glaucomatous cupping, drusen, and retinal pigment epithelium changes within the macular region. Humphrey visual field testing confirmed glaucomatous changes. Drusenoid pigment epithelial detachments were observed bilaterally with optical coherence tomography.

Conclusions and importance: We hypothesize that in addition to the lungs, spermatozoa and the Fallopian tubes, the retinal pigment epithelium may also be affected by ciliary dysfunction in individuals with Kartagener syndrome. Given recent advances in our knowledge of retinal ciliopathies, further studies are needed to understand how ciliary dysfunction affects the retina in Kartagener syndrome.

1. Introduction

Kartagener syndrome is a rare primary ciliary dyskinesia characterized by the triad of sinusitis, bronchiectasis and situs inversus.[1,2] Patients with complete Kartagener syndrome exhibit the entire triad, while the incomplete form describes those without situs inversus.[2] It is usually inherited as an autosomal recessive disorder, but autosomal dominant and X-linked forms have been reported.[3,4] The incidence is estimated to be 50% of cases of primary ciliary dyskinesia, which has an incidence of 1 per 10,000 to 20,000 births.[1] Cilia in the lungs, spermatozoa and Fallopian tubes are characteristically affected.[3] Therefore, the literature focuses on sinopulmonary dysfunction, infertility and inversion of the visceral organs.[3] Although ocular abnormalities in Kartagener syndrome have not been reported, there have been cases of X-linked primary ciliary dyskinesia associated with retinitis pigmentosa.[4] To the best of our knowledge, our case is the first report of nonexudative age-related macular degeneration in association with Kartagener syndrome.

2. Case report

A 55-year-old woman, originally from Cuba, with a history of incomplete Kartagener syndrome and chronic angle closure glaucoma presented for evaluation of the retina. She had a history of hearing loss, as well as numerous hospitalizations for asthma and pneumonia since infancy. However, she was not diagnosed with Kartagener syndrome until the age of 35 when a nasal biopsy was performed in the United States. She had a family history of the disorder, with a younger brother having complete Kartagener syndrome. The patient was first diagnosed with angle closure glaucoma at the age of 28 and over the years underwent trabeculectomy in both eyes (OU), as well as peripheral iridectomy, goniosynechialysis and endocyclophotocoagulation of the left eye (OS).

On examination, her best corrected visual acuity was 20/50 in the right eye (OD) and 20/80 in OS. Pupils and extraocular movements were normal. Intraocular pressures (17 mmHg OD, 18 mmHg OS) were well-controlled on topical medications. Axial lengths were 21.58 mm OD and 21.36 mm OS. Dilated fundus examination showed a cup-to-disc ratio of 0.9 OU and evidence of retinal pigment epithelium changes with soft drusen in the macula (Fig. 1). Automated visual field testing revealed severe constrictions bilaterally (Fig. 2). On spectral domain (SD) OCT B-scans, all drusen and drusenoid pigmented epithelial detachments appeared convex with medium and homogenous internal reflectivity (Fig. 3). SD-OCT Angiography (OCT-A) did not show any abnormalities. These findings supported the diagnosis of dry age-related macular degeneration and chronic angle closure glaucoma.

3. Discussion

In this case report, we present a patient with Kartagener syndrome

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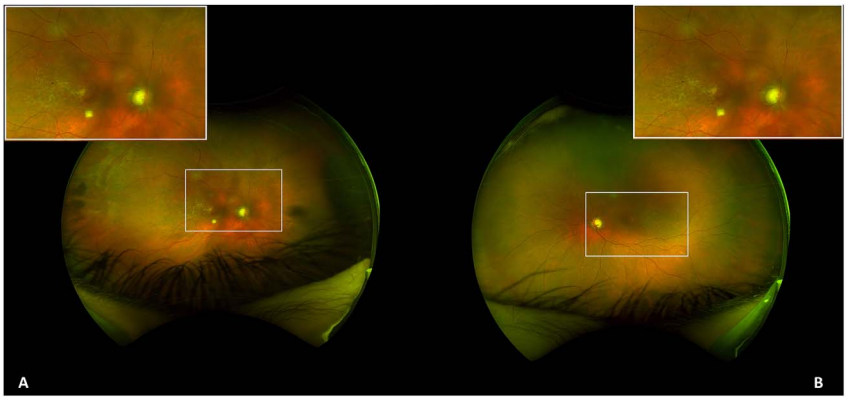

Fig. 1. Optos ultra-widefield imaging. Optos photos show drusenoid deposits (insets with magnified macular view) and retinal pigment epithelium atrophy of the macula in both the right (A) and left (B) eye.

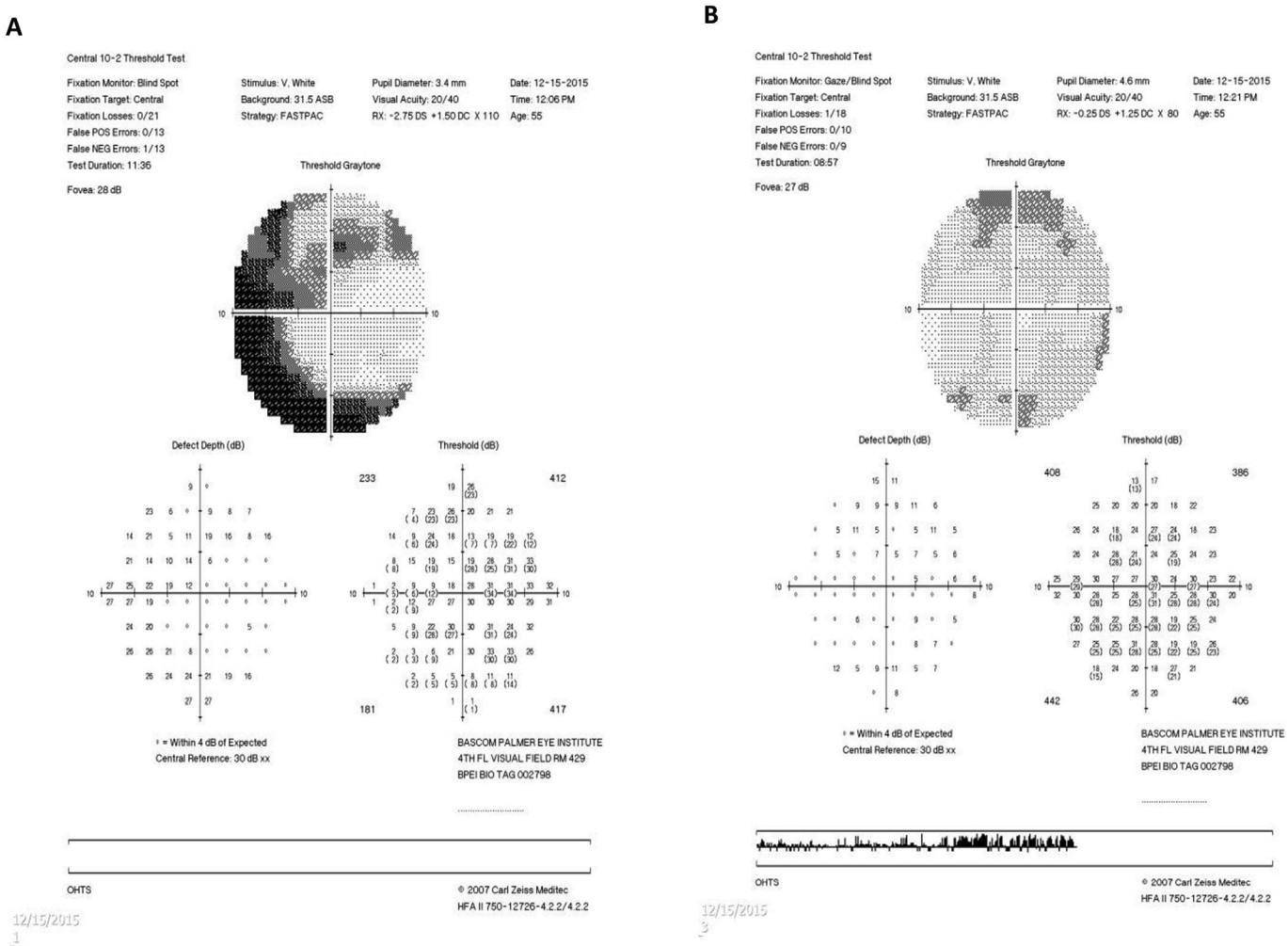

Fig. 2. Humphrey 10-2 visual field pattern deviation map. An inferior arcuate extending superiorly was noted temporal to fixation in the right eye (A). An arcuate defect of both superior hemifields was present in the left eye (B).

with ocular findings of nonexudative age-related macular degeneration. Based on these observations, we hypothesize that in addition to the lungs, spermatozoa and the Fallopian tubes, the retinal pigment epithelium (RPE) may also be affected by ciliary dysfunction in individuals with Kartagener syndrome.

In recent years, there has been an increased understanding of retinal ciliopathies.[3] Hunter et al., were the first to report the concept of retinal ciliopathies, as they observed abnormalities in the sperm of patients with Usher Syndrome and X-linked Retinitis Pigmentosa.[5,6] Interestingly, it was the similarity in structure between the cilium in photoreceptors and flagellum in sperm that led to their initial hypothesis that anomalies in ciliary function are linked to both retinal and sperm dysfunction in these disorders.[5,6] More recent advances in molecular genetics have shown that several genes associated with retinal cilia, such as RP1, RPGR and RPGR-IP, play a role in retinal degeneration.[3] Specifically, X-linked mutations in the retinitis pigmentosa GTPase

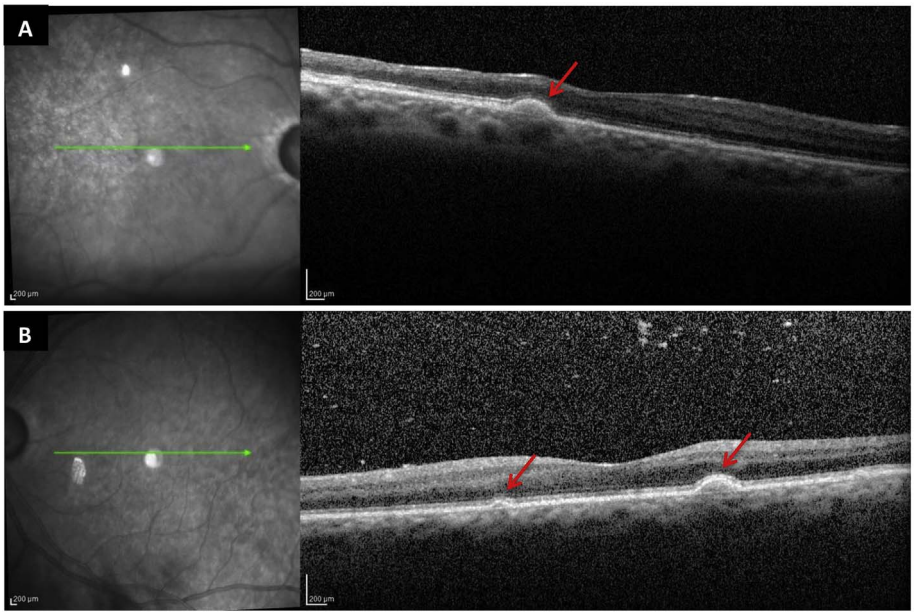

Fig. 3. Optical coherence tomography. Drusenoid pigmented epithelial detachments (red arrows) are seen in the right (A) and left (B) macula. There is medium internal reflectivity under the RPE and no fluid accumulation. (For interpretation of the references to colour in this figure legend, the reader is referred to the Web version of this article.)

Regulator (RPGR) gene have been associated with retinal ciliopathies seen in primary ciliary dyskinesia and macular degeneration.[3,4,7,8] Furthermore, OCRL-1, a gene product associated with Lowe oculocerebrorenal syndrome, has been shown to be present in primary cilia of the RPE.[9]

4. Conclusions

The retinal findings identified in the current case suggest that a similar mechanism of ciliary dyskinesia could be present in patients with Kartagener syndrome. However, given the patient in our case report was 55-years-old, the findings of macular degeneration may be unrelated to Kartagener syndrome. Information regarding the significance and purpose of the cilia in the RPE cilia is limited.[10] Further studies are needed to better understand the effect ciliary dysfunction has on the retina. We recommend an ophthalmologic examination for all patients with Kartagener syndrome to rule out macular degeneration.

Patient consent

Consent was obtained from the patient before the study and publication of the case report.

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Conflicts of interest

The following authors have no financial disclosures: MDG, CVV, JRD, TPC, AMB.

Authorship

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

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None.

Appendix A. Supplementary data

Supplementary data related to this article can be found at http://dx.doi.org/10.1016/j.ajoc.2018.02.006.

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