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SHORT COMMUNICATION

Bilateral disc edema and unilateral macular hole in a patient with retinitis pigmentosa

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PURPOSE. *A unique case of retinitis pigmentosa (RP) associated with bilateral disc edema and unilateral macular hole is presented.*
METHODS. *A 49-year-old woman, a known RP patient, was found to have bilateral disc edema and a macular hole in the left eye during routine clinical examination. Fluorescein angiography revealed hyperfluorescent leakage of the optic nerve head significantly OD and minimally OS. There was staining in the macular regions which was consistent with retinal pigment epithelium atrophy OD and cystoid macular edema (CME) OS. Cerebrospinal fluid pressure and examination by lumbar puncture was normal. Disc edema spontaneously decreased bilaterally during follow-up.*
DISCUSSION. *Bilateral disc edema was thought to be secondary to inflammation caused by rapid degeneration of photoreceptors and retinal pigment epithelium and macular hole was secondary to CME.*
CONCLUSIONS. *Inflammatory response in the course of retinitis pigmentosa may result in disc edema and cystoid macular edema, which may further progresses to macular hole. (Eur J Ophthalmol 2006; 16: 487-90)*

KEY WORDS. *Macular hole, Optic nerve head edema, Retinitis pigmentosa.*

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INTRODUCTION

Retinitis pigmentosa (RP), one of the most common hereditary retinal degenerations, is characterized by night blindness and constricted visual fields with retinal bone spicule pigmentary changes. Other ocular findings associated with RP include arteriolar narrowing, optic atrophy, posterior subcapsular cataracts, posterior vitreous detachment, cystoid macular edema, hyaline bodies or drusen of the optic nerve head (ONH), and pallor of the ONH.

Although pseudo-papilledema is common in RP, true disc edema and RP is a rare association (1-3). Atrophic maculopathy, macular pucker formation, and cystoid macular edema are common macular changes seen in patients with RP. Macular hole has also been reported rarely in association with RP (4). We report a unique case with bilateral ONH edema and unilateral macular hole in a patient with RP.

Case report

A 49-year-old woman with a previous history of RP presented to our clinic for routine examination. General medical history was unremarkable with a good general health. She had no history of recent head trauma. Her family history revealed no ocular or systemic disease. Ophthalmologic examination revealed best-corrected visual acuity of 0.9 in the right eye and 0.7 in the left. Ocular motility, pupillary reactions, and intraocular pressure were within normal limits. Slit-lamp examination findings were unremarkable with clear lenses. Pupillary dilatation was limited in spite of both 1% cyclopentolate and 2.5% phenylephrine instillation. Biomicroscopic examination of the vit-

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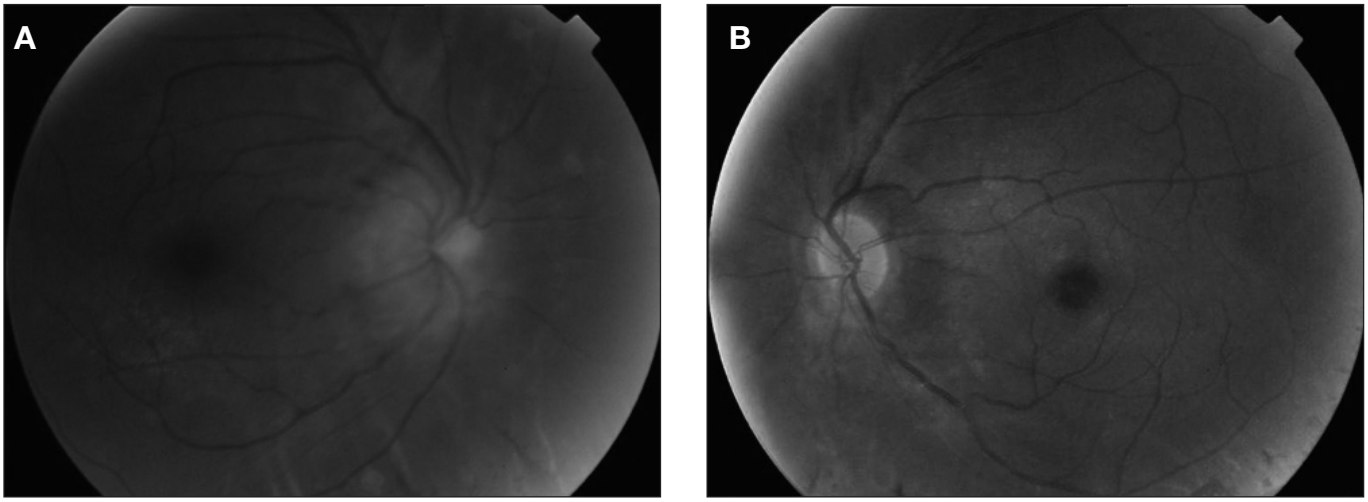

Fig. 1 - *Fundus photography of the (**A**) right and (**B**) left eye. Note significantly elevated disc margins in the right eye and minimal blurring of disc margins in the left with bilateral peripheral bone-spicule pigmentation.*

reous showed no abnormality except partial posterior vitreous detachment in the left eye. Fundus examination revealed bone spicule pigmentation and arteriolar narrowing consistent with RP bilaterally. There was marked elevation of ONH with indistinct margins in the right eye (Fig. 1A) and blurred margins of the ONH with a subtle elevation together with cystoid macular changes associated with a partial thickness macular hole in the left eye (Fig. 1B). No optic disc hemorrhage was detected in either eye. There was no abnormal autofluorescence consistent with optic disc drusen, which was confirmed with B-scan ultrasonography and computerized tomography (CT) of the eyes, both of which were free of calcification on ONH. On fluorescein angiography (FA), there was significant hyperfluorescence and leakage of the optic disc starting in early phases together with some hyperfluorescence due to retinal pigment epithelium (RPE) atrophy in the macular area of the right eye (Fig. 2A). There was only minimal staining of the ONH, in addition to typical perifoveal flower-petal pattern of the cystoid macular edema in the FA of the left eye (Fig. 2B). Optical coherence tomographic examination revealed normal foveal structure with a central foveal thickness of 178 µm in the right eye, and cystoid macular edema associated with partial thickness macular hole in the left (Fig. 3). There was severe constriction of peripheral visual fields in both eyes. The results of the cranial CT were unremarkable. The patient was referred to the neurology department for evaluation of cerebrospinal fluid (CSF) with lumbar puncture, which revealed an opening pressure of 195 mmH_20 and a normal CSF composition excluding a possible diagnosis of pseudotumor cerebri. There were no signs of CSF inflammation or infectious disease.

The 6-month follow-up examination revealed unchanged visual acuities and visual fields with spontaneously decreased ONH edema in the right eye and resolved ONH edema in the left without treatment.

DISCUSSION

This patient had bilateral ONH edema and unilateral macular hole. The differential diagnosis of bilateral edematous ONH for this patient included ONH drusen and papilledema secondary to increased intracranial pressure. ONH drusen are commonly associated with RP causing pseudo-papilledema (1). Drusen can appear on the surface of the ONH, which can be seen with ophthalmoscopy, or they can be buried beneath the surface. On B-scan ultrasonography, drusen will appear as highly reflective calcified nodules at or within the ONH. These calcified nodules can also be seen easily with CT. Another way to differentiate ONH drusen from true ONH edema is to perform a FA. Visible drusen on the surface of the ONH will autofluoresce in the prefluorescein injection photographs. During the angiogram they will hyperfluoresce, but they will not cause any fluorescein dye leakage. FA of the presented case was consistent with a true ONH edema with dye leakage. Both hypertensive retinopathy and diabetic papillopathy can present with bilateral ONH ede-

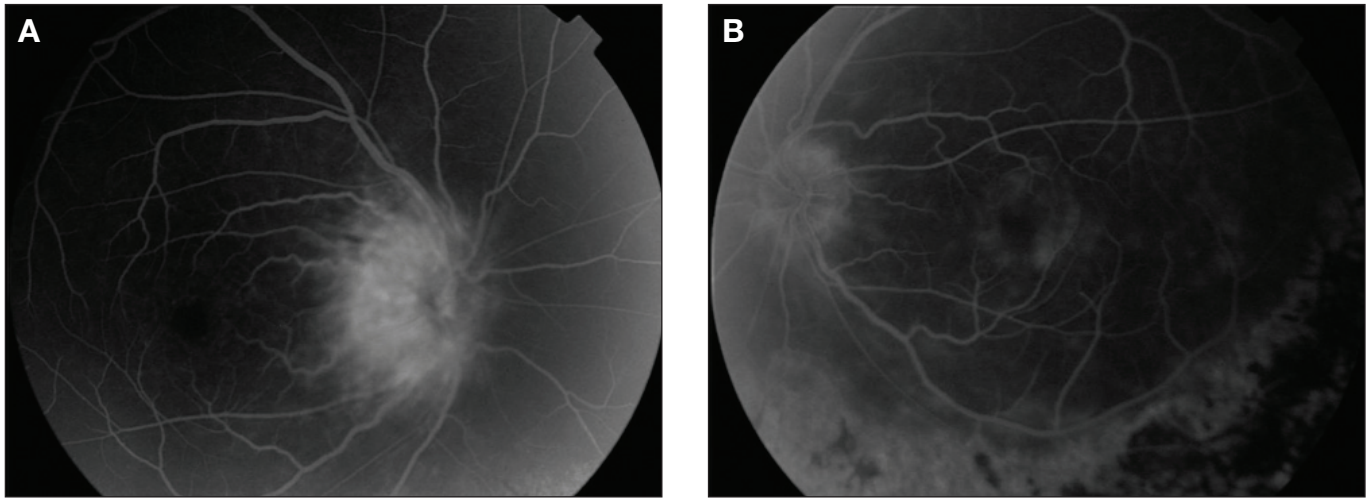

Fig. 2 - *(**A**) Fluorescein angiography of the right eye; note significant fluorescein leakage of the optic disc and with some macular hyperfluorescence due to retinal pigment epithelium atrophy. (**B**) Fluorescein angiography of the left eye; note minimal staining of the optic nerve head with perifoveal hyperfluorescence due to cystoid macular edema.*

ma; however, the presented case had no hypertension or diabetes mellitus.

Bilateral ONH edema without any significant visual loss realized by the patient was most consistent with the diagnosis of papilledema, whereby disc swelling from increased intracranial pressure is caused by blockage of axoplasmic transport in the prelaminar portion of the ganglion cell axons (5). The patient had none of the symptoms associated with increased intracranial pressure including headache, nausea, vomiting, or transient visual loss. Work-up included CT scan of the brain to rule out an intracranial mass and CSF pressure measurement to rule out pseudotumor cerebri as a cause of the increased intracranial pressure.

In the absence of increased intracranial pressure, one possible theory to explain the ONH edema is that this patient had an inflammatory disc edema associated with RP. It was theorized that an inflammatory reaction begins as the photoreceptors and RPE undergo a bout of rapid degeneration in RP, which causes retinal edema and disc vessel leakage, leading to the ONH edema (1). Additionally, the altered state of retinal vascular permeability and breakdown of the outer blood retinal barrier in RP may cause cystoid macular edema (6). Heckenlively et al reported that there is a significant association between CME and the presence of circulating antiretinal antibodies in cases who presented with RP and CME (7). Literature review revealed two cases of bilateral ONH edema associated with RP (1, 3). Here we report a unique case with bilateral ONH edema and unilateral macular hole in a patient with RP.

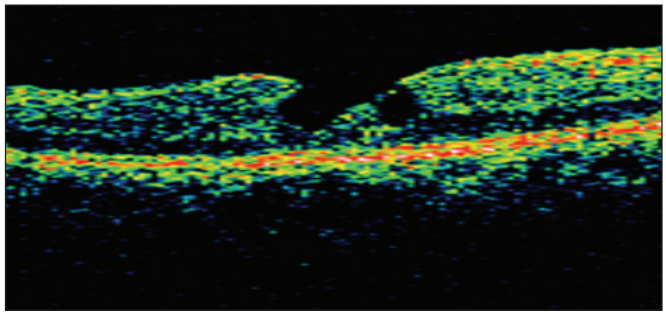

Fig. 3 - *Optic coherence tomography of the left eye showing a partial thickness macular hole.*

The presented case had relatively advanced RP which presented with bilateral ONH edema and cystoid macular changes together with a partial thickness macular hole in the left eye verified by FA and OCT. Macular hole seems to be a result of cystoid macular changes in this patient. ONH edema in the left eye might have been more evident before her presentation to us which started to resolve spontaneously before her application. We propose that the cause of the optic disc and macular edema was breakdown of blood retinal barrier together with inflammation caused by severe RPE degeneration.

The authors have no, proprietary interest.

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REFERENCES

1. Villa AM, Anderson SF, Abundo RE. Bilateral disc edema in retinitis pigmentosa. Optom Vis Sci 1997; 74: 132-7.
2. Hayasaka S, Noda S, Setogawa T. Disc edema in juvenile retinitis pigmentosa. Ann Ophthalmol 1991; 23: 304-7.
3. Sachdev MS, Verma L, Gary SP, Moonis M, Shelear CH, Gupta NK. Bilateral disc oedema in retinitis pigmentosa: an unusual sign. Jpn J Ophthalmol 1987; 31: 621-6.
4. Rao K, Shah G, Blinder K. Bilateral macular hole formation in a patient with retinitis pigmentosa. Ophthalmic Surg Lasers 2002; 33: 152-4.
5. Hayreh SS. Optic disc edema in raised intracranial pressure. VI. Associated visual disturbances and their pathogenesis. Arch Ophthalmol 1977; 95: 1566-79.
6. Fogle JA, Welch RB, Green WR. Retinitis pigmentosa and exudative vasculopathy. Arch Ophthalmol 1978; 96: 696-702.
7. Heckenlively JR, Jordan BL, Aptsiauri N. Association of antiretinal antibodies and cystoid macular edema in patients with retinitis pigmentosa. Am J Ophthalmol 1999; 127: 565-73.