Unusual association of diseases/symptoms

Spontaneous resolution of full-thickness macular hole in retinitis pigmentosa

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Summary

A 57-year-old woman, diagnosed with retinitis pigmentosa and positive family history, was referred to the retinal clinic. Full-thickness macular hole was noted in the right eye with no significant vitreomacular interface abnormalities confirmed at baseline visit with spectral-domain optical coherence tomography scan. Routine follow-up showed spontaneous closure of the full-thickness macular hole with four-letter improvement from baseline visit and remains unchanged with 3 years of follow-up.

BACKGROUND

Loss of central vision in patients with long-standing retinitis pigmentosa can be devastating as the peripheral visual fields are usually compromised much earlier. Macular pathology is not uncommon in retinitis pigmentosa as cystoid maculopathy, epiretinal membrane or less commonly full-thickness macular holes.

Recent publications^{1 2} in peer-reviewed literature suggest possible role of vitreoretinal surgery to improve outcomes for macular holes in retinitis pigmentosa patients, with OCT evidence of anatomical success, albeit, with limited functional gains.

Our case demonstrates that unless dynamic vitreomacular interface abnormalities can be accounted for by SD-OCT imaging where surgical intervention is logical, spontaneous closure can be a natural outcome and anatomical closure not necessarily a successful surgical outcome unless accompanied by functional and/or visual improvement.

CASE PRESENTATION

A middle-aged Caucasian woman was referred to the retinal clinic, having been diagnosed with retinitis pigmentosa with annual follow-up for 18 years in a general ophthalmic clinic. She had symptoms of night blindness as long as she could remember. Her father and brother were diagnosed with retinitis pigmentosa as well. She was fit and well generally and only took medication for well-controlled systemic hypertension, with no syndromic features known to be associated with her ophthalmic condition as deafness, myopathy, peripheral neuropathy or cardiac problems. She was noted to be non-smoker and a social drinker.

Baseline visual acuity was noted to be LOGMAR 0.9 (Snellens 6/48) right eye and 0.3 (Snellens 6/12) left eye with no significant distance refractive error. Pupillary reactions were normal with no relative afferent pupillary defect and full ocular motility on testing. Anterior segment examination was significant only for early nuclear sclerotic cataracts. Goldmann applanation tonometry revealed intraocular pressures of 16 and 17 mm Hg, respectively for the eyes.

Dilated fundus examination showed classic midperipheral bone-spicule pigmentation, attenuated retinal vasculature and pale optic discs as shown in figure 1. Full-thickness macular hole was noted in the right eye, confirmed by spectral-domain optical coherence tomography (SD-OCT) scan (3D 1000 Topcon Mark I Topcon Corporation, Tokyo, Japan). No evidence of significant epiretinal membrane or vitreomacular interface abnormalities were noted to account for the macular hole.

The patient had not noted any recent decrease in vision and believed that her right eye vision had been poor for some years, possibly related to retinitis pigmentosa. She denied any trauma to account for the macular hole.

INVESTIGATIONS

Detailed spectral domain OCT examination at baseline showed full-thickness macular hole in the right eye with no significant epiretinal membrane or vitreomacular traction noted as for idiopathic macular holes. (figure 1A,B) The left eye at baseline showed few inner retinal cysts typical of retinitis pigmentosa cystoid maculopathy with no vitreomacular interface changes.

Spontaneous closure was noted at 12 months and 36 months visits after baseline (figure 1C,D), confirmed by SD-OCT scan.

Clear hyper-reflective tissue was noted in the right fovea confirmed by raster scans and 3D imaging at 36 months visit (figure 2C,D) compared to baseline visit (figure 2A,B).

DIFFERENTIAL DIAGNOSIS

Clinical and SD-OCT examination ruled out any element of significant vitreomacular interface abnormalities, choroidal neovascularisation or evidence of previous retinal trauma to account for the full-thickness macular hole.

TREATMENT

Guarded prognosis was discussed with the patient for possible surgery; however, the patient elected to be observed as functionally she had not noticed any recent decrease in vision.

Figure 1 spectral-domain optical coherence tomography B scans and colour fundus photos. Top row: baseline visit (A) shows full-thickness macular hole (arrow) and (B) colour fundus photograph (arrow). Bottom row: At 3 years follow-up visit (C) shows spontaneously resolved hole with hyper-reflective lamellar tissue (arrow) and corresponding colour fundus photograph (D).

Figure 2 Top row baseline visit: (A) shows 3D rendered optical coherence tomography scan with segmentation showing full-thickness macular hole at baseline (left) and (B) shows series of six scans at the presumed foveal centre show no neurosensory reflective tissue in the four frames (arrows). Bottom row at 3-year follow-up, (C) shows 3D segmented image for comparison with no evidence of macular hole and (D) shows series of six scans clearly showing hyper-reflective neurosensory retinal tissue in all the frames centred around the presumed foveal centre (arrows).

OUTCOME AND FOLLOW-UP

At 12 months follow-up from baseline, clinical and SD-OCT examination showed spontaneous closure of the right full-thickness macular hole although the visual acuity was still noted to be LOGMAR 0.8 (Snellens 6/38) and the patient herself had not noted any significant subjective improvement. The left eye status remained essentially unchanged.

At 3-year follow-up after baseline visit, her visual acuity remains LOGMAR 0.8 right eye and 0.3 left eye, with a residual lamellar macular hole in the right eye.

DISCUSSION

Macular abnormalities are well known features of retinitis pigmentosa, ranging from mild epiretinal membrane, cystoid maculopathy to full-thickness macular holes. The pathophysiology of macular hole formation in retinitis pigmentosa remains speculative.^{3 4} Jin *et al* in their small case series possibly alluded to the presence of vitreomacular traction, myopia and epiretinal membrane as contributory. Our case is unique that no such clinical changes were evident to account for the macular hole although we can not confirm the status of her eyes prior to her presentation to the retina clinic as no prior OCT imaging was available for comparison. Atrophic full-thickness macular hole hypothesis with de-roofing of coalescent macular cysts seems likely but the spontaneous improvement would be an unlikely feature.

One possible confounding factor can be the eye movement during OCT scanning that can produce artifacts; however, the patient's eyes were imaged with SD-OCT 1000 Topcon raster scan protocol and good fixation was noted on all the visits.

Recent interest in surgical options to for full-thickness macular holes in retinitis pigmentosa has been reported, although the evidence base is weak with few patient outcomes reported. Unless significant epiretinal membrane or dynamic vitreomacular traction is confirmed clinically and

with SD-OCT imaging, current choice of pars plana vitrectomy with internal limiting membrane peel for atrophic holes seems illogical. Our case clearly demonstrates that high resolution and reproducible imaging with SD-OCT can confirm the spontaneous improvement of atrophic macular holes, albeit with little functional gain. The patients need to be fully informed prior to offering vitreoretinal surgical intervention for such cases.

Learning points

- ▶ The pathogenesis of full-thickness macular holes in retinitis pigmentosa, unless clearly showing dynamic vitreomacular interface changes, remains speculative
- ▶ Atrophic full-thickness macular holes can occur in retinitis pigmentosa patients with possible spontaneous closure
- ▶ Surrogate markers for anatomical and functional success in retinitis pigmentosa patients for macular hole surgery need to be interpreted with caution.

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Competing interests None.

Patient consent Obtained.

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