CASE REPORT

Proliferative diabetic retinopathy in typical retinitis pigmentosa

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SUMMARY

A 39-year-old woman with typical retinitis pigmentosa (RP) for 9 years and a positive family history of night blindness was diagnosed with diabetes mellitus (DM). She developed proliferative diabetic retinopathy (PDR) during the course of disease. She was promptly managed with pan retinal photocoagulation (PRP). PDR developing in a case of typical RP is extremely rare and has not been reported in the literature to date. Recognition of this rare, vision threatening complication, points out a definite need to further look deep into the pathogenesis of diabetic retinopathy.

BACKGROUND

Diabetic retinopathy is an important cause of preventable blindness and retinitis pigmentosa is an ocular condition known to have a protective effect against development of diabetic retinopathy.¹ We describe a case of a 39-year-old woman with typical retinitis pigmentosa who developed proliferative diabetic retinopathy. To the best of our knowledge, this is the first case to be reported of proliferative diabetic retinopathy in a case of typical retinitis pigmentosa.

CASE PRESENTATION

A 39-year-old woman presented with painless progressive loss of vision of 1 year duration, more in the left than the right eye. She had night blindness for 9 years and a significant positive family history of night blindness, with her father and two siblings having similar symptoms. She was a diabetic on treatment with oral hypoglycaemic agents and insulin for 2 years. There was no history of other systemic illness and family history of systemic diseases was unremarkable.

A detailed ocular examination was performed. Visual acuity was 20/30 in both eyes and intraocular pressure 20 mm Hg by non-contact tonometer. Anterior segment evaluation of both eyes showed early posterior sub capsular cataract (P1-P2 by LOCS III classification). The patient did not have evidence of anterior segment inflammation on slit lamp examination. A detailed fundus examination of both eyes revealed typical bony spicule pigmentation in the mid-peripheral region, arteriolar attenuation and pallor of the disc. In addition, micro aneurysms were seen in both eyes and neovascularisation of the disc was noted in the left eye (figure 1). There was no evidence of vitreous floaters/opacities, retinal vascular sheathing, retinal holes, tears, retinal telangiectasia or subretinal exudation. A clinical diagnosis of bilateral typical

retinitis pigmentosa with mild non-proliferative diabetic retinopathy of the right eye and proliferative diabetic retinopathy of the left eye was made.

INVESTIGATIONS

Automated perimetry with a Humphrey field analyser revealed bilateral constriction of visual field, which was correlating with the finding of RP (figure 2). Fluorescein angiography was carried out, which revealed the presence of multiple point hyper-fluorescent areas in the perifoveal region of both eyes with leak in the late phase and blocked fluorescence caused by the bony spicule pigmentation in the mid-periphery. In addition, there was also, in the left eye, hyper-fluorescence at the disc, which increased in the mid to late phase suggestive of definite neovascularisation at the disc. Fluorescein angiography confirmed the clinical diagnosis of proliferative diabetic retinopathy (PDR) in the left eye with neovascularisation of the disc along with presence of microaneurysms and thickening of the macula not involving the centre, and mild non-proliferative diabetic retinopathy in the right eye (figure 3).

The systemic investigations revealed an uncontrolled hyperglycaemic status with fasting blood glucose of 240 mg% and glycated hemoglobin 8.4%. The patient's blood urea was 23 mg% and serum creatinine was 0.9 mg%. Her biochemical and haematological investigations were unremarkable except for uncontrolled hyperglycaemia.

TREATMENT

The patient was promptly managed with pan retinal photocoagulation (PRP) of the left eye in three sittings, and strict glycaemic control. At the end of 1 year follow-up, the patient had a stable disease in the left eye post PRP. The right eye showed progression of the diabetic retinopathy to proliferative stage, with appearance of neovascularisation, which was confirmed by fluorescein angiography (figure 4). The patient was managed with PRP of the right eye in three sittings.

OUTCOME AND FOLLOW-UP

After 2 years follow-up, the patient has stable disease in both eyes post PRP with absence of leak in the fluorescein angiography (figure 5). Visual acuity was maintained at 20/40 in both eyes.

DISCUSSION

Diabetic retinopathy and retinitis pigmentosa are two common conditions not shown to coexist in the same individual. In a large population based

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Figure 1 Fundus picture of both eyes. (A) Right eye shows retinal pigment epithelial mottling, arteriolar narrowing with waxy pallor of the disc. (B) Left eye shows presence of neovascularisation of the disc (black arrow), arteriolar narrowing and pigment mottling. (C) Typical bony spicule pigmentation of the mid-periphery in the right eye is seen. A similar condition was seen in the left eye as well.

study, Chen *et al*² clearly showed that retinitis pigmentosa reduces the risk of proliferative diabetic retinopathy. Tarr *et al*³ have given a detailed description of the various interconnecting pathways and key contributors to the development of diabetic retinopathy. Sternberg *et al* have shown a negative coincidence of DR and RP. They suggested that attenuation of the blood vessels and the presence of early posterior vitreous detachment prevented the progression to proliferative stage.¹ Arden *et al*, in their detailed survey of patients with DM and RP, showed that there was no evidence of proliferative changes in the retina in patients with RP. They explained that the possible loss of rods decreased the severity of hypoxia and production of vascular endothelial growth factor, thereby preventing the changes of diabetic retinopathy from developing in patients with RP.⁴ Spalton *et al*⁵ explained the role of inflammation due to degeneration of photoreceptors and retinal pigment epithelium in the development of retinal oedema.

There are anecdotal reports of retinal vascular abnormalities previously described in RP patients and include a Coats-type RP, sub retinal exudation, retinal detachment, and neovascularisation of disc and periphery. In all these reported cases, loss of receptor cells, RPE dysfunction, altered metabolic environment of the retinal vasculature and capillary non perfusion were identified as possible causes. These cases were not associated with

diabetes mellitus.^{6 7} Hotta and Hotta⁸ described an isolated case of diabetic macular oedema in an RP patient that was managed with trans-Tenon's retrobulbar triamcinolone infusion.

Our patient was a case of typical retinitis pigmentosa with characteristic fundus appearance. The form of inheritance in this patient is unlikely to be of an X linked recessive nature considering that she was a female. The possibility of autosomal dominant or recessive inheritance remains, although we could not perform genetic tests because of logistic and financial constraints. The case under discussion was a known case of RP with uncontrolled hyperglycaemia. She did not have evidence of anterior segment inflammation on slit-lamp examination. On detailed fundus examination, there was no evidence of vitreous floaters/opacities, retinal vascular sheathing, retinal holes, tears, retinal telangiectasia or subretinal exudation. Her biochemical and haematological investigations were unremarkable except for the uncontrolled hyperglycaemia. The presence of microaneurysms and new disc vessels in addition to presence of uncontrolled DM led us to consider PDR in this case. Arden⁴ suggested that the degree of functional retina at the time of onset of DM could influence the development of DR in cases of RP, which can be considered in our case as well, considering the age of our patient, the duration of RP and the duration of DM.

Figure 2 Automated perimetry (HFA) demonstrates characteristic visual field constriction of both eyes.

Figure 3 Fundus fluorescein angiography of both eyes. (A, B) Right eye shows point hyper-fluorescence, suggestive of microaneurysms (black arrow) and blocked fluorescence caused by the typical bony spicule pigments (white arrow). (C, D) Left eye shows leakage at the disc (star) and point hyper-fluorescence with late leak in the perifoveal areas.

Figure 4 Fundus fluorescein angiography of both eyes at 1 year post PRP in the left eye. (A,B) Early phase of both eyes showing point hyperfluorescence suggestive of microaneurysms (black arrow); blocked fluorescence of the pan retinal photocoagulation scars in the left eye (white arrow). (C–E) Left eye in the mid to late phase shows blocked fluorescence by PRP scars (quad arrow), regression of new vessels and absence of leakage near disc (red star). (F–H) Right eye shows late leakage of the disc confirming neovascularisation (black star). Also shown are the microaneurysms (black arrows) and bony spicule pigments causing blocked fluorescence (white arrow).

Figure 5 Fundus fluorescein angiography post proliferative diabetic retinopathy in both eyes shows regression of new disc vessels with absence of leakage 2 years later.

Learning points

- ▶ This is the first case of proliferative diabetic retinopathy (PDR) to be reported in a typical case of retinitis pigmentosa (RP).
- ▶ This leads to consideration of how and why PDR developed in a patient with RP and warrants further research into the pathophysiology of PDR developing in RP.
- ▶ Beyond hypoxia and free radicals, the role of inflammation might be an important underlying pathophysiology in this patient.
- ▶ Similar presentations, if encountered by ophthalmologists, may provide more insights into the pathophysiology of the disease and its management.

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All authors contributed to literature review, drafting of the manuscript and approved the final version of the manuscript.

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