CASE REPORT

Unilateral giant peripapillary drusen and retinal drusenoid deposits in a case of X-linked retinoschisis

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Accepted 6 February 2016

SUMMARY

A 25-year-old man with X-linked retinoschisis presented with multiple giant optic nerve head drusen and multiple retinal drusenoid deposits in one eye. There was an area of coarse pigmentation in the lower half of the retina, with convex upper borders extending on either side of the optic disc, suggestive of a spontaneously resolved retinal detachment. The possible aetiopathogenesis of such occurrence is discussed.

BACKGROUND

We report the formation of giant peripapillary drusen and retinal drusenoid deposits in a case of juvenile retinoschisis with unilateral spontaneously-resolved retinal detachment (SRRD). Giant peripapillary drusen have been described previously in cases of retinitis pigmentosa (RP).¹ Large drusen have also been reported following long-standing retinal detachment (RD).² To the best of our knowledge, such large peripapillary drusen have not been reported in X-linked retinoschisis (XLRS).

CASE PRESENTATION

A 25-year-old man presented to us with dimness of vision in the left eye noted since childhood. Best corrected visual acuity was 6/18 ($-0.50@100$) in the right eye and 1/60 ($-0.75@100$) in the left eye. Intraocular pressure of both eyes was normal. The fundus of the right eye showed a spoke-wheel pattern at the fovea, vitreous veils and a normal peripheral retina (figure 1A). The left fundus revealed mild disc pallor, blotchy coarse pigmentation of the inferior fundus involving the macula,

with distinct convex superior margins separating this pigmented zone from normal appearing superior retina (figure 1B).

INVESTIGATIONS

Multiple glistening white, confluent spherical lesions, resembling a bunch of grapes, were seen at the peripapillary area and at the superotemporal fundus. These were autofluorescent and hyperechoic on ultrasound, with acoustic shadowing. Fundus fluorescein angiogram (FFA) did not show any leak at the macula in either eye. Optical coherence tomography (OCT) confirmed the foveoschisis in the right eye (figure 2A) and foveal thinning with no cysts and no schisis in the left eye. On OCT, the lesions in the peripapillary region and superotemporal part of fundus had distinct hyper-reflective outer shells with inner hyporefectivity. The hyper-reflective shells cast a shadow posterior to the lesion (figure 2B, C). Axial length was 23.2 mm in the right eye and 23.5 mm in the left eye. Electroretinogram showed reduced 'b' wave amplitude with preservation of the 'a' wave. The patient had neither café au lait spots, neurofibroma, mental retardation nor adenoma sebaceum, nor did he have a history of trauma or epilepsy. There was no family history of ocular tumours, especially of retinoblastoma. A diagnosis of XLRS with SRRD in the left eye with giant optic disc drusen (ODD) was made.

TREATMENT

The patient was started on topical dorzolamide 2% three times a day in the right eye.

Figure 1 (A) The fundus of the right eye revealed a spoke-wheel (stellate) pattern at the fovea suggestive of foveoschisis. (B) The montage fundus photograph of the left eye revealed mild pallor of the optic disc, whitish glistening round nodules at the optic disc and superotemporal retina. Blotchy coarse pigmentation in the inferior fundus is noted with convex superior margins and a normal appearing superior retina.

To cite: Tripathy K, Chawla R, Meena S, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-214558

Figure 2 (A) Optical coherence tomography (OCT) of the right macula showed multiple areas of hyporeflectivity at outer nuclear and nerve fibre-ganglion cell layer level with multiple bridging retinal elements in between, suggestive of foveoschisis. (B) On OCT, the nodules over the disc margins showed a hyporeflective round inner core with hyper-reflective margins in the inner retina casting a shadow posterior to the lesions. (C) OCT of the retinal drusenoid deposits in the superotemporal retina also demonstrated a hyper-reflective outer shell with hyporeflective inner core.

OUTCOME AND FOLLOW-UP

At follow-up after 1 year, the vision and central macular thickness were stable in both eyes.

DISCUSSION

We would like to highlight a few observations from this case. First, there was a loss of schitic cavities in the eye with SRRD. Second, unilateral giant optic disc and retinal drusenoid deposits have not been reported in XLRS. Bilateral disc drusen have been reported earlier in association with posterior microphthalmos, foveoschisis and RP.³ The typical distinct superior convex margin with blotchy coarse pigmentation of the rest of the

inferior fundus in our case was highly suggestive of an SRRD. The presence of retinoschisis in the other eye was also a risk factor for RD. Schisis cavities at the macula are known to disappear with ageing and treatment with carbonic anhydrase inhibitors. In our case, the loss of such cavities at the macula may indicate generalised retinal thinning in SRRD. Third, presence of such large peripapillary and retinal drusenoid deposits has not been reported following spontaneous retinal re-attachment. Giant drusen have been noted in 50% of cases with phthisis bulbi, which may develop after 4 weeks of RD and become more prominent in the following 2–3 months.⁴ Peripapillary lesions such as those seen in our case have been reported as presumed astrocytic hamartoma (AH) in RP.⁵ Both AH and ODD may be associated with calcification and autofluorescence. The available histopathological evidence indicates that, even in RP, these peripapillary lesions are more likely to be large drusen and not hamartomas.¹ The drusen in chronic RD appear to be formed due to metaplasia of retinal pigment epithelial cells into fibroblasts.² Perhaps in RP and chronic RD's or SRRD, drusen formation results from the metaplasia of clumps of anteriorly migrating retinal pigment epithelial cells. These may further undergo ossification with time and might be the source of intraocular calcification seen in long-standing blind eyes with chronic retinal detachments.

Learning points

- ▶ The foveoschisis in X-linked retinoschisis (XLRS) may not be evident after spontaneous retinal reattachment.
- ▶ Spontaneously settled retinal detachment in XLRS may be associated with giant peripapillary drusen such as those typically seen in retinitis pigmentosa.
- ▶ Giant drusen around the optic disc may denote metaplastic response of the retinal pigment epithelium.
- ▶ Optical coherence tomography features of giant peripapillary drusen are described.

Contributors RC and KT had full access to all data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. KT, SM and PA were responsible for acquisition of data. RC provided administrative, technical and material support, and supervised the study. All the authors were involved in study concept and design, analysis and interpretation of data, drafting of the manuscript and critical revision of the manuscript for important intellectual content.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Puck A, Tso MO, Fishman GA. Drusen of the optic nerve associated with retinitis pigmentosa. *Arch Ophthalmol* 1985;103:231–4.
- 2 Shenoy SK, Babu K, Prabhakaran VC. Large drusen following long-standing retinal detachment. *Indian J Pathol Microbiol* 2010;53:329–30.
- 3 Ayala-Ramirez R, Graue-Wiechers F, Robredo V, et al. A new autosomal recessive syndrome consisting of posterior microphthalmos, retinitis pigmentosa, foveoschisis, and optic disc drusen is caused by a MFRP gene mutation. *Mol Vis* 2006;12:1483–9.
- 4 Stefani FH. Phthisis bulbi—an intraocular fluoride proliferative reaction. *Dev Ophthalmol* 1985;10:78–160.
- 5 Kinori M, Moroz I, Rotenstreich Y, et al. Bilateral presumed astrocytic hamartomas in a patient with retinitis pigmentosa. *Clin Ophthalmol* 2011;5:1663–5.

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