Case Report
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Syndromic myopia in marfan's disease: about a case with review of the literature

Abstract

Objective: To describe a case of myopic syndrome in Marfan disease.

Results: The patient was 45 years old, with high myopia and wearing corrective lenses, with no previous history of any particular problem. She was seen for the management of a progressive bilateral decrease in visual acuity in a white, painless eye, with no history of trauma and no other associated signs. On the clinical examination, the corrected visual acuity was estimated at 02/10 in both eyes. The refraction revealed strong myopia at -11D. The examination of the lens after pupillary dilatation reveals a cortical cataract and upward crystalline ectopia in both eyes. At the posterior pole, there was a myopic cone, chorioretinal atrophy and poor macular reflex, with vessels of normal caliber. The retina was flat with no peripheral retinal tears

Keywords: syndromic,myopia, marfan, lens, ectopia

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Introduction

Marfan syndrome is an autosomal dominant inherited disorder characterized by mesenchymal dysplasia leading to degeneration of elastic fibers.[1]

The objective is to describe a rare multi-organ disease (skeleton, eye, heart) whose ocular manifestation may be part of syndromic myopia. We report the case of a patient seen in consultation.

Case report

The patient was 45 years old, with high myopia and wearing corrective lenses, with no previous history of any particular problem. She was seen for the management of a progressive bilateral decrease in visual acuity in a white, painless eye, with no history of trauma and no other associated signs.

On the clinical examination, the corrected visual acuity was estimated at 02/10 in both eyes.

The refraction revealed strong myopia at -11D; intraocular pressure was 14 mm hg in the right eye and 15 mm hg in the left.

In the anterior segment, the cornea is clear and transparent, with preserved integrity.

The anterior chamber is calm, deep and optically empty in both eyes.

The iris has good coloration and normal trophicity in both eyes.

The examination of the lens after pupillary dilatation reveals a cortical cataract and upward crystalline ectopia in both eyes (Figure 1&2).

In the posterior segment, the vitreous is transparent, posterior vitreous detachement.

At the posterior pole, there was a myopic cone, chorioretinal atrophy and poor macular reflex, with vessels of normal caliber. The retina was flat with no peripheral retinal tears. A retinophotography and a fluorcein angiography were repeated to explore the posterior pole. (Figure 3&4)

The rest of the examination was completed by a cardiovascular workup to check for complications.

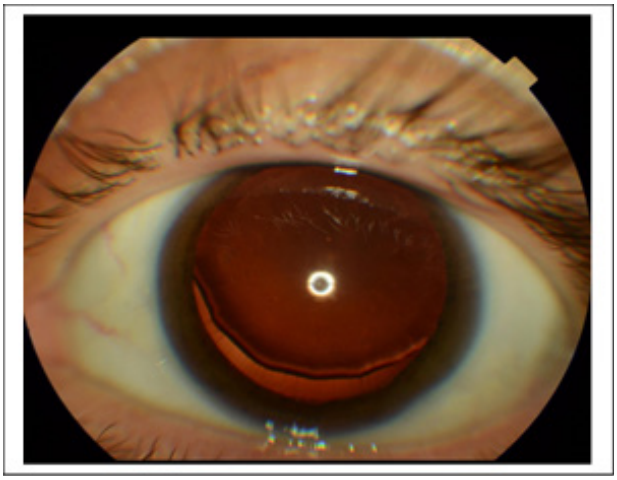

Figure 1 Right eye.

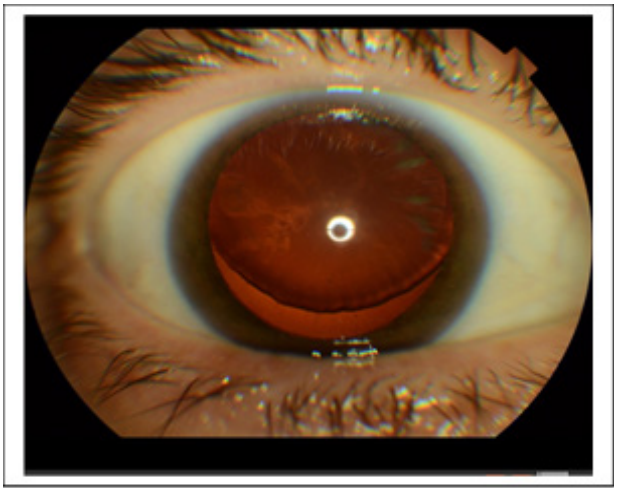

Figure 2 Left eye.

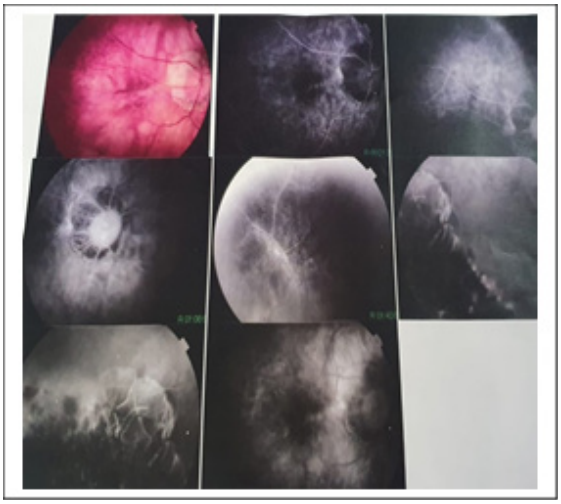

Figure 3 Right eye.

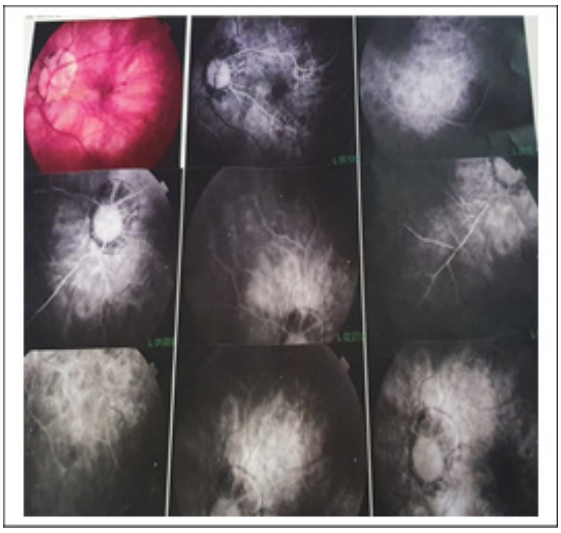

Figure 4 Left eye.

Discussion

The marfan syndrome is a rare disorder with an estimated prevalence of 1 in 5000 or 10000 individuals, depending on the region. The diagnosis is based on a number of major and minor criteria.[1]

In the ocular involvement,[2,3] crystalline ectopia is the main criterion, found in 95% of cases, followed by degenerative lesions of the retinal periphery in 22.5% of cases. The minor criteria include flat cornea, elongated eyeball, hypoplastic iris or ciliary muscle hypoplasia. In our case, the patient presented with crystalline ectopia, with no visualization of degenerative lesions to date. The ectopy is frequently directed downwards and rarely upwards, as in our case.

The progression may be towards the anterior chamber dislocation with an acute ocular hypertonia, or into the vitreous, followed shortly afterwards by retinal detachment.[4,5]

When the dislocation occurs at an early age and asymmetrically, a profound amblyopia may set in, reducing the chances of visual recovery without an urgent treatment. The development of cataracts in ectopic lenses is also frequent.

The management of ectopia is complex, and studies are not always unanimous. Some authors recommend the monitoring in the presence of good visual acuity, and intervention in the presence of profound visual acuity deterioration or signs of complication. Other authors prefer early intervention to avoid amblyopia.[5]

Retinal detachment occurs in 5-30% of Marfan syndrome patients, with an average age of 20-25 years.[5,6]

It is important to always evaluate for life-threatening cardiac involvement.[7,8]

Conclusion

Syndromic myopia secondary to Marfan syndrome is a rare pathology requiring early management and close monitoring because of the visual and often vital prognosis at stake.

Acknowledgments

None.

Conflicts of interest

The authors report no conflicts of interest.

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