CASE REPORT

Ocular complications of marfan syndrome. Report of two cases

Mema V, Qafa N

University Hospital Centre "Mother Theresa", Ophthalmologic Service, Tirana, Albania

Abstract

Background: Marfan Syndrome occurs in 8 to 10 per 100.000 of population / year[1]. Ocular features of this syndrome have been extensively reported. We report two patients in the same family with miscellaneous complications of this syndrome.

Case report: Two sisters with Marfan Syndrome were examined in our clinic. They revealed severe ocular complications as bilateral spontaneous complete posterior lens dislocation, total rhegmatogenous retinal detachment, and secondary glaucoma. Combined surgical and conservative treatment was applied to both cases with relative successful results. They were also referred to cardiologist for further evaluation and management.

Conclusions: Ocular complications commonly occur in Marfan's Syndrome. Total rhegmatogenous retinal detachment, secondary glaucoma, etc. These findings require prompt and aggressive treatment to minimize visual loss in these patients. Hippokratia 2010; 14 (1): 45-47

Keywords: Marfan Syndrome, Rhegmatogenous retinal detachment, secondary glaucoma, posterior crystalline lens dislocation

Corresponding author: Vilma Mema, University Hospital Centre "Mother Theresa", Ophthalmologic Service, P.C. 1012, Tirana, Albania, Tel. +355 67 20 29 613, e-mail: vilmaoculus@yahoo.com

Marfan's Syndrome has been reported to occur in 8 to 10 per 100.000 of population / year[1]. Ocular features of this syndrome have been repeatedly reported[2]. Ectopia lentis, the most common ocular feature, occurs in 70 to 80 % of cases[3-6]. Isolated reports of spontaneous complete posterior crystalline lens luxation, or developing secondary complications like glaucoma or uveitis have also been described[7]. However, bilateral posterior dislocation is a rare feature[5].

Two cases-sisters of established Marfan's Syndrome with bilateral spontaneous posterior lens dislocation, total rhegmatogenous retinal detachment, secondary glaucoma are reported.

Case 1

A 32-year-old female presented in our clinic with acute painless visual loss in the right eye of two days duration and chronic pain in the left eye. Clinical examination revealed skeletal abnormalities (long, thin extremities), arachnodactyly and high-arched palate;. These findings were consistent with Marfan Syndrome (Figure 1 a-b). Ocular examination demonstrated in the right eye a visual acuity of hand movement (no correction) and significant decrease (3/20) (sph.+4.5 D; cyl.+1.75 X 170°) in the left eye. Intraocular pressure was in the right eye 13 mm Hg and in the left eye 36 mm Hg. She underwent surgery for glaucoma (Trabeculectomy and basal iridectomy) in the left eye when she was 18 years old. Moreover she referred that her little sister had visual disturbances as well.

Dilated fundus examination showed **bilateral** spontaneous posterior lens dislocation associated with total rhegmatogenous retinal detachment in the **right eye** (Figure 2 a,b).

Ultrasonography of the right eye revealed spikes consistent with a total rhegmatogenous retinal detachment and a posterior dislocated lens (Figure 2).

The patient underwent successful retinal detachment surgery (Pars Plana Vitrectomy + Silicone Oil Injection) in the right eye. The visual acuity at 6 months was 4/20, (+6.5 D/+1.00X10°) (Figure 3 a,b). She also received antiglaucomatous therapy for the left eye, but with poor results (presence of vitreous in the anterior chamber). A surgical intervention was also planned in the left eye, but till now we got no patient consent to do that.

She was then referred to a cardiologist were she underwent a complete investigation.

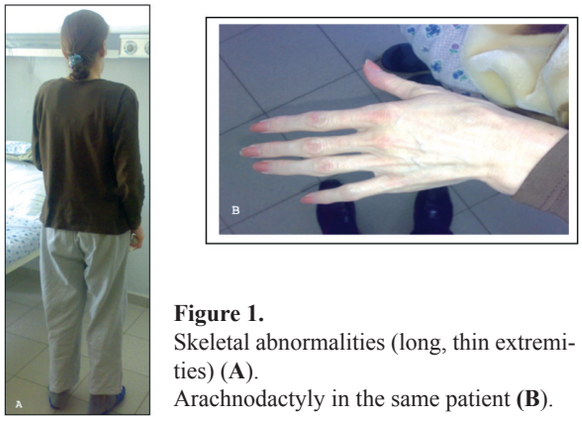

Figure 1.
Skeletal abnormalities (long, thin extremities) (**A**).
Arachnodactyly in the same patient **(B)**.

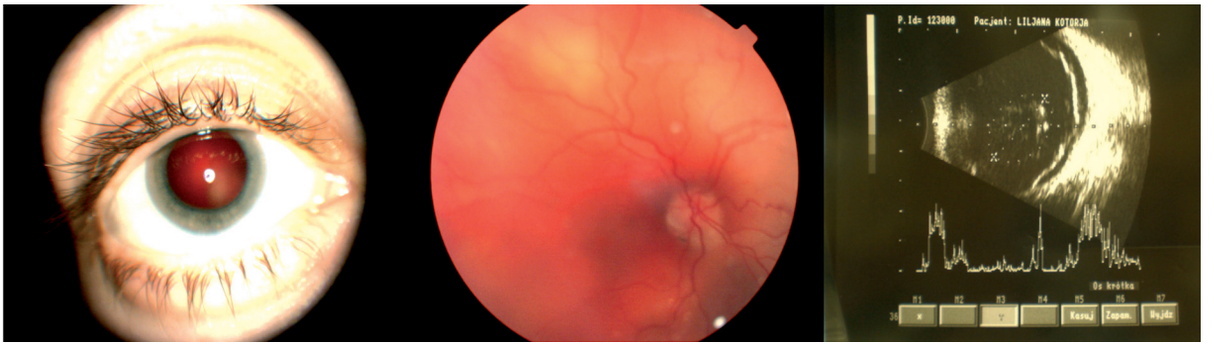

Figure 2: The lack of the lens (left). Fundoscopic view of total rhegmatogenous retinal detachment (center). Ultrasound examination of the right eye – Retinal detachment and presumed lens material in the vitreous cavity (right).

Case 2

Her little sister (24 years old) presented to the department of Ophthalmology after contacting her. The patient never been examined by an ophthalmologist in the past. She complained for blurred vision in both eyes. Physical examination revealed again skeletal abnormalities (long, thin extremities), arachnodactyly and high-arched palate. These findings were once again compatible with the diagnosis of Marfan Syndrome.

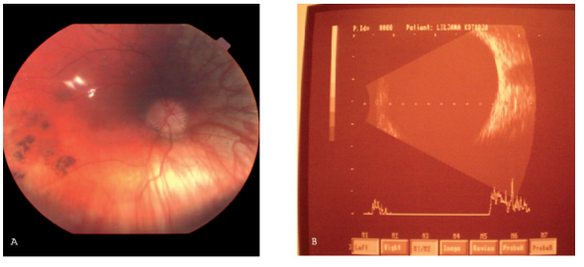

Figure 3: Right eye fundoscopic view 6 months after surgery (A). Ultrasound examination of the same eye (B)

Ocular examination showed diminished visual acuity in both eyes (right eye: 6/20,sph.+6.5; left eye: 6/20,sph.+8.5 D). Intraocular pressure was 17.3 mm Hg both eyes.

Fundus evaluation showed **bilateral** spontaneous posterior lens dislocation associated with retinal thinning and degeneration. Interestingly no tears **were noted** (Figures 4,5).

The patient was treated with spectacles in both eyes. She was advised to maintain a close follow-up due to the potential of the development of future ocular complications (glaucoma, retinal detachment, etc). She was also referred to a cardiologist for further evaluation.

Discussion

Ocular complications are commonly found in Marfan Syndrome. Of them, complete bilateral spontaneous posterior lens dislocation is a rare manifestation [5]. Posterior dislocation may be silent and rarely causes glaucoma or uveitis [7].

Vitreous changes are well described in Marfan's Syndrome. Commonly observed changes include liquefaction of the gel and posterior vitreous detachment (PVD). Presence of fluid vitreous in the anterior chamber and PVD in our cases are in agreement with this description. Posterior dislocation of lens could have been augmented by the disruption of anterior hyaloids face resulting in loss of support. Usually these cases are prone to have more severe vitreous and retinal pathologies. Such complications as total rhegmatogenous retinal detachment, secondary glaucoma, need prompt and aggressive treatment to minimize the degree of visual loss.

To finalize the need (and duty) to refer the patients with Marfan Syndrome to cardiologic evaluation should be emphasize in order to decrease morbidity and mortality related to cardiovascular complications (aorta aneurysm, mitral insufficiency, etc.).

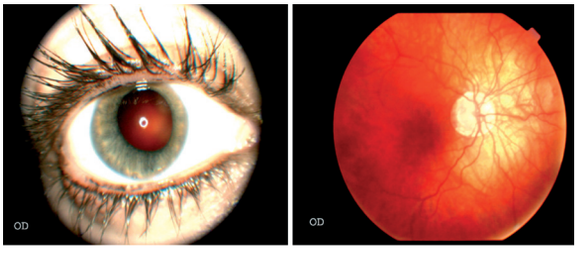

Figure 4: Right eye. the lack of the lens (left). Fundoscopic view of the same eye (right).

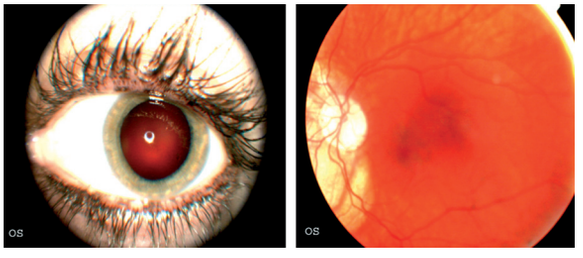

Figure 5: Left eye.The lack of the lens (left). Fundoscopic view of the same eye (right).

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