Short communication

Late onset lens particle glaucoma in Marfan syndrome

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ABSTRACT

Case report: A case is presented of an acute onset lens particle glaucoma originating from a crystalline lens spontaneously dislocated into the vitreous for more than 20 years in a patient diagnosed with Marfan syndrome.

Discussion: Marfan syndrome is a connective tissue disorder with autosomal dominant inheritance caused by fibrillin gene mutation. Ectopia lentis is the predominant ocular abnormality and a major diagnostic criterion. An association between Marfan syndrome and glaucoma has also been demonstrated. The reported case is unusual in that a complete spontaneous lens dislocation to vitreous was present and progressed to secondary lens particle open angle glaucoma.

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Glaucoma tardío por partículas de cristalino en síndrome de Marfan

RESUMEN

Caso clínico: Glaucoma agudo por partículas de cristalino, en una paciente afecta de síndrome de Marfan que presentaba un cristalino luxado en vitreo de más de 20 años de evolución.

Discusión: El síndrome de Marfan es un trastorno hereditario, autosómico dominante, del tejido conectivo causado por mutaciones del gen de la fibrilina. La ectopia lentis es la alteración ocular predominante y el criterio mayor de diagnóstico, siendo frecuente el desarrollo de glaucoma en los pacientes afectos de síndrome de Marfan. El caso que se expone es particular, dado que presenta una luxación completa, espontánea y bilateral del

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Clinical case

A case is presented of a 42-year-old patient diagnosed with Marfan syndrome. Her ophthalmology history showed the presence of a bilateral spontaneous lens dislocation (for more than 20 years), which had been subject to surgery in the left eye by means of pars plana vitrectomy and crystalline phacoemulsification, with no intraocular lens implant for four years. In addition, the patient was receiving bilateral ocular hypotensive treatment with timolol 0.5% due to primary open-angle glaucoma (POAG).

The patient has a family history of two sisters and a niece who had been diagnosed with Marfan syndrome, all of whom showed a bilateral lens dislocation and, in one case, POAG.

The patient went for the first time to our Emergency Room due to two-day course severe pain and decreased right eye visual acuity.

Right eye exploration revealed a best-corrected visual acuity of hand movement at 1 m, an intense ciliary injection, epithelial corneal edema and crystalline cortical residues in a very deep anterior chamber; intraocular pressure (IOP) was 39 mmHg and the funduscopy and gonioscopy tests were difficult to evaluate due to the corneal edema. By means of ocular ultrasound, besides the crystalline lens dislocated into the vitreous, a properly attached retina and no other complications were observed (Fig. 1).

Given the diagnosis of suspected lens particle acute glaucoma, treatment with topical ocular hypotensive agents (fixed combination of timolol 0.5% and brimonidine tartrate 0.2% every 12 h) and oral ocular hypotensive agents (acetazolamide 250 mg every 12 h), as well as topical corticosteroids (dexamethasone 0.1% every 3 h) was started.

Two days after having started treatment, the corneal edema had disappeared and the IOP was of 24 mmHg, which enabled a better observation of the crystalline lens residues and the cellular Tyndall effect in the anterior chamber (Fig. 2), as well as a Shaffer’s grade IV open angle and the presence of an intumescent lens in the vitreous chamber.

At cornea level, endothelial guttas and cellular pleomorphism were observed. A test with confocal microscopy (HRT-II/RCM) was performed, showing hyperreflective particles among the endothelial cells (Fig. 3).

The patient was subject to surgery to remove the dislocated crystalline lens by means of 3G pars plana vitrectomy with phacoemulsification, with no intraocular lens implant.

At present, the patient has a best-corrected Snellen visual acuity in the right eye of 0.4 (refraction +4.00, −1.00 at 80°), her eye still shows no signs of inflammation, the IOP is controlled around 12–13 mmHg with topical medication (fixed combination of brinzolamide/timolol every 12 h) and a cupping/blind spot ratio of 0.7.

Discussion

The Marfan syndrome falls within the group of type 1 fibrillinopathies, whose main manifestations take place in the cardiovascular, musculoskeletal and ocular systems. It consists of a set of clinical pictures with overlapping phenotypic variations derived from gene FBN1 mutation. This gene
encodes a protein called fibrillin 1, which provides strength and elasticity to connective tissue.\(^1\)

Between 50 and 80% of patients diagnosed show lens dislocation. The evidence of ectopia lentis is, together with the aortic root aneurysm, a major diagnostic criterion\(^2\) (Table 1). The alteration in the structure of fibrillin 1, a main component of zonular microfibrils, seems to be responsible for zonular instability, which would lead to lens dislocation. However, cases of complete lens dislocation are rare in Marfan syndrome. This patient, as well as the rest of her family members, shows juvenile onset bilateral lens dislocation. Recently, a case was published where a family showed a novel mutation in FBN1 gene, which was related to juvenile onset complete lens dislocation;\(^3\) just like in this series, the case presented showed a bilateral cornea guttata which, although it could be an isolated corneal dystrophy, might also be due to a mutation in the fibrillin gene. In fact, some series have been published where the relationship between Marfan syndrome and changes in corneal endothelium\(^4\) is described, with a highly reflective stromal matrix and bright and reflective particles among the corneal endothelial cells, which were also observed by confocal microscopy\(^5\) in the patient presented.

Glaucoma prevalence in patients with Marfan syndrome is high. It has been suggested that defects in the connective tissue, present in the aqueous humor drainage canal, would be responsible for the frequency with which these patients show POAG; moreover, a dislocated crystalline lens would also lead to angle-closure glaucoma due to pupillary block (phacopic), which is usually present in these patients; iatrogenic glaucoma (postoperative) or phacolytic\(^6\) glaucoma have also been described in a lower percentage of cases. In the case presented herein, although the patient had been previously diagnosed with POAG, the acute clinical picture that led her to visit the Emergency Room, the absence of goniosynechia and the presence of crystalline lens residues in the anterior chamber make us think that the increased IOP was secondary to the lens particles released from the vitreous chamber, where the lens had been dislocated for many years. Although there are cases published of phacolytic glaucoma after traumatic lens dislocation into the vitreous, or in patients with pseudoexfoliation, cases of phacolytic glaucoma after spontaneous dislocation are very rare in hereditary diseases.\(^7\) In the case presented herein, the defect of the crystalline lens capsule dislocated into the vitreous was such that it enabled the entry of fragments thereof to the chamber and the iridocorneal angle, thus causing a secondary increase in IOP. Despite the fact that there are several publications reporting this type of glaucoma after cataract surgery, we believe this is the first case published of lens particle glaucoma after lens spontaneous dislocation into the vitreous, within the context of Marfan syndrome.

### Table 1 – Revised Ghent criteria for the diagnosis of Marfan syndrome.\(^2\)

<table>
<thead>
<tr>
<th>In the absence of a family history</th>
<th>In the presence of a family history</th>
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<tr>
<td>(1) Ao (Z ≥ 2) + EL = MFS</td>
<td>(5) EL + FH of MFS + MFS</td>
</tr>
<tr>
<td>(2) Ao (Z ≥ 2) + FBN1 = MFS</td>
<td>(6) Syst (≥ 7 points) + FH of MFS = MFSa</td>
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<tr>
<td>(3) Ao (Z ≥ 2) + Syst (≥7 points) = MFSa</td>
<td>(7) Ao (Z ≥ 2 if above 20 years old, ≥3 if below 20 years old) + FH of MFS = MFSa</td>
</tr>
<tr>
<td>(4) EL + FBN1 with known Ao = MFS</td>
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**Systemic score**

- Wrist + thumb sign – 3 (wrist OR thumb sign – 1)
- Pectus carinatum – 2 (pectus excavatum or chest asymmetry – 1)
- Hindfoot deformity – 2 (plain pes planus – 1)
- Pneumothorax – 2
- Dural ectasia – 2
- Protrusio acetabuli – 2
- Reduced US/LS + increased arm/height + no severe scoliosis – 1
- Scoliosis or thoracolumbar kyphosis – 1
- Reduced elbow extension – 1
- Facial features (3/5) – 1 (dolichocephaly, enophthalmos, downslanting palpebral fissures, malar hypoplasia, retrognathia)
- Skin striae – 1
- Myopia > 3 diopters – 1
- Mitral valve prolapse (all types) – 1

**Maximum total: 20 points; score ≥ 7 indicates systemic involvement**

\(Ao,\) aortic diameter at the sinuses of Valsalva above indicated Z-score or aortic root dissection; \(EL,\) ectopia lentis; \(FBN1,\) fibrillin-1 mutation; \(FBN1\) with known \(Ao,\) \(FBN1\) mutation identified in a patient with aortic root aneurysm; \(FH,\) family history; \(Syst,\) systemic score; \(US/LS,\) upper segment/lower segment ratio; \(Z,\) Z-score.

**MFSa (warning):** without discriminating features of Shprintzen–Goldberg syndrome, Loeys–Dietz syndrome or Ehlers–Danlos syndrome:

- \(TGFBR1/2,\) collagen biochemistry, COL5A1 testing indicated.
- Other conditions or genes will emerge with time.

### Conflicts of interest

The authors declare that they do not have any conflicts of interest.
REFERENCES