Interdepartmental Conference

Spontaneous Lens Dislocation

La-ongsri Atchaneeyasakul, M.D.*
Punnarueg Thongcharoen, M.D.**

Abstract : **We present a multidisciplinary approach to a patient with spontaneous lens dislocation. A 39-year-old female developed blurred vision in both eyes one year prior to presentation. This symptom could not be corrected with spectacles. Her previous history showed progressive bilateral myopia since the age of ten and a single episode of syncope without exertion 16 months before presentation. Ophthalmic examination demonstrated cataracts with upward and outward displacement of the crystalline lenses in both eyes. Manifest refraction and keratometry displayed moderate myopia with high lenticular astigmatism in both eyes. Marfan syndrome was suspected on the basis of general physical examination and a complete cardiovascular examination performed by the geneticist and the cardiologist respectively. In addition, the role of the cardiovascular-thoracic surgeon in the management of Marfan syndrome is presented.**

Key words: *lens dislocation/subluxation, Marfan syndrome, lenticular astigmatism*

เรื่องย่อ : **ภาวะเลนส์ตาเคลื่อนซึ่งเกิดขึ้นเอง**

ละอองศรี อัชชนียะสกุล พ.บ.*, ปุณณฤกษ์ ทองเจริญ พ.บ.**
**ภาควิชาจักษุวิทยา, **ภาควิชาศัลยศาสตร์, คณะแพทยศาสตร์ศิริราชพยาบาล, มหาวิทยาลัยมหิดล, กรุงเทพมหานคร 10700.*

สารศิริราช 2545; 54: 235-240.

รายงานผู้ป่วยหญิงอายุ 39 ปี มีประวัติตามัวทั้งสองข้างมา 1 ปี อาการตามัว แก้ไขไม่ได้ด้วยแว่นสายตา ผู้ป่วยเคยได้รับการตรวจว่ามีภาวะสายตาสั้นทั้งสองข้างตั้งแต่อายุ 10 ปี และภาวะสายตาสั้นเป็นมากขึ้นเรื่อย ๆ 16 เดือนก่อนมาโรงพยาบาล ผู้ป่วยมีอาการหมดสติโดยไม่ทราบสาเหตุ ผลการตรวจตาแรกรับพบต้อกระจกและเลนส์ตาเคลื่อนทั้งสองข้าง การวัดความโค้งกระจกตา และวัดสายตา พบว่าผู้ป่วยมีภาวะสายตาสั้นระดับปานกลางร่วมกับภาวะสายตาเอียงมากซึ่งเกิดจากเลนส์ตาเคลื่อน ผู้ป่วยได้รับการวินิจฉัยว่าเป็น Marfan syndrome ภายหลังการตรวจร่างกายอย่างละเอียด โดยอายุรแพทย์สาขาเวชพันธุศาสตร์ และตรวจระบบหัวใจและหลอดเลือดโดยอายุรแพทย์สาขาหทัยวิทยา ผู้รายงานได้นำเสนอแนวทางการดูแลรักษาปัญหาเรื่องเลนส์ตาเคลื่อน รวมทั้งนำเสนอบทบาทของแพทย์ศัลยศาสตร์หัวใจและทรวงอก ในการดูแลผู้ป่วย Marfan syndrome

**Department of Ophthalmology , **Department of Surgery, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok 10700, Thailand.*
From Interdepartmental Conference, August 10, 2001.

INTRODUCTION

Spontaneous lens dislocation is a condition in which the crystalline lens is displaced from its usual location without evidence of eye injury. The condition can be either unilateral or bilateral. The crystalline lens may displace anteriorly into the anterior chamber or posteriorly into the vitreous cavity. In the case of anterior dislocation, the lens diameter is usually smaller than normal (so called microspherophakia). Primary ocular disorders associated with spontaneous lens dislocation include congenital glaucoma/buphthalmos,[1] pseudoexfoliation syndrome,[2] syphilis/chronic uveitis,[3] retinitis pigmentosa,[4] aniridia,[5] and high myopia.[6] More importantly, spontaneous lens dislocation may be a presenting sign of serious systemic conditions such as Marfan syndrome, homocystinuria, Weill-Marchesani syndrome, sulfite oxidase deficiency, and hyperlysinemia.[7-10] There are several ocular complications related to lens dislocation including amblyopia, uveitis, glaucoma, and retinal detachment.

A patient with bilateral spontaneous lens dislocation is presented. The authors demonstrate a multidisciplinary approach to the definitive diagnosis and management of this condition.

CASE REPORT

A 39-year-old Thai female complained of decreased vision in both eyes for 1 year. The symptoms were gradually progressive and could not be corrected with her current spectacles. The patient has been wearing spectacles for nearsightedness since she was 10 years old. At age 22, bilateral subluxation of the crystalline lenses was first diagnosed by a local ophthalmologist. However, no further investigations were undertaken at that time. Sixteen months prior to hospitalization, the patient had a single episode of syncope without exertion. The records from the private hospital from where the patient was transferred showed an unremarkable general physical examination. The family history was negative for inherited eye diseases. The patient denied any underlying disease or previous eye injury.

The general physical examination and ophthalmic examination at presentation is summarized as follows:

General physical examination

Vital signs: HR 60/min, BP 144/65 mmHg
Stature: height 175 cm, span 174 cm, upper segment/ lower segment 89.5/85.5
Head and neck: high arched and narrow palate
Skeletal: pectus excavatum, arachnodactyly (positive wrist sign), hyperextensible joints, flat feet, lumbar lordosis
Cardiovascular: no heart murmur
Respiratory: normal
Abdomen: normal

Ophthalmic examination

VA with glasses: RE 5/60, LE 2/60
VA with glasses with pinhole: RE 6/36, LE 6/36
Intraocular pressure: RE 20, LE 20 mmHg
Extraocular muscles: normal movement
Slit lamp examination:
Cornea – clear
Anterior chamber – normal depth, clear
Pupil – 3 mm diameter, reactive to light
Lens – nuclear sclerosis 2+, posterior lens dislocation (upward and outward displacement) (Figure 1)
Vitreous – clear
Dilated fundus examination: Normal optic disc, macular, and retinal background
Keratometry: RE 41.25/40.50, LE 41.50/40.75
Axial length of the eyeball: RE 24.40, LE 24.36 mm
Refraction: RE –7.50 -4.00 x 15°, LE –6.75 -5.00 x 5°

The patient was scheduled for lens removal with intraocular lens implantation in both eyes.

DISCUSSION

There are several primary ocular disorders that are associated with spontaneous lens dislocation. These include congenital glaucoma, pseudoexfoliation syndrome, chronic uveitis, retinitis pigmentosa, aniridia, intraocular tumor, and high myopia. Systemic conditions commonly associated with spontaneous lens dislocation include Marfan syndrome, Weill-Marchesani syndrome, homocysti-

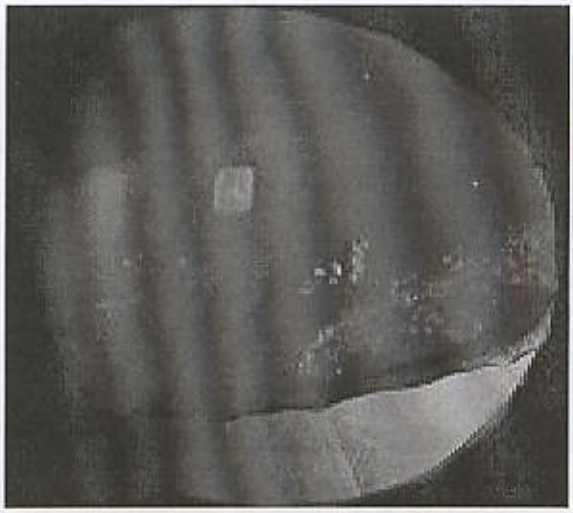

Figure 1. The picture shows localized cataract with upward displacement of the crystalline lens.

nuria, hyperlysinemia, and sulfite oxidase deficiency. Moreover, spontaneous lens dislocation can be isolated, in which the patient develops lens dislocation without other systemic and ocular problems. In syndromic cases, there are specific ocular and systemic features suggesting the diagnosis of each syndrome. For example, specific ocular findings in Marfan syndrome include elongated zonules, flat cornea, megalocornea, increased axial length of the globe, microspherophakia, cataracts, and high risk of retinal detachment after lens extraction. Myopia and microspherophakia are typical findings in both Weill-Marchesani syndrome and homocystinuria. However, usually ragged zonular remnants are seen in homocystinuria.

Indications for lensectomy in spontaneous lens dislocation include lens in the anterior chamber, lens-induced uveitis, lens-induced glaucoma, lenticular opacity with poor visual function, refractive error not amenable to optical correction, and impending dislocation of the lens. In cases where the lens is dislodged into the anterior chamber, prolonged attachment between the lens capsule and the cornea might cause permanent damage to the corneal endothelium. Moreover, acute angle closure glaucoma may develop. Therefore, an attempt should be made to relocate the lens into the posterior chamber without delay; otherwise the dislocated lens should be removed surgically.

Our patient developed bilateral spontaneous lens dislocation, which was documented by an ophthalmologist since the age of 22. Unfortunately, a definite diagnosis of Marfan syndrome was not confirmed until 17 years later. At presentation, her vision was significantly decreased as a result of cataracts and lens induced moderate myopia with high astigmatism. We could not improve her vision by spectacle correction. Therefore, surgical removal of the dislocated lenses is the appropriate management. Theoretically, we could leave the lens in the eye if it dislocates into the vitreous without any evidence of intraocular inflammation or retinal injury and prescribe the glasses to correct any refractive error that occurs after the lens dislocation.

Role of cardiovascular-thoracic surgeon in Marfan syndrome, incidence and outcomes at Siriraj Hospital

There are mainly three sites of involvement: the aorta, the aortic valve and the mitral valve. The most common cardiovascular manifestation is a pear-shaped aneurysm of the ascending aorta with or without aortic regurgitation (Figures 2-4). The dilatation of the aorta almost always starts at the ascending aorta because of its greatest wall tension. This change may lead to aortic valve annulus dilatation and aortic regurgitation especially when the diameter of the ascending aorta is greater than 6 cm. Another important condition is aortic dissection (Figure 5), which occurs in around 1/3 of patients with Marfan syndrome. Significant dilatation of the aorta and a family history of dissection are risk factors or the development of dissection. The prognosis of the patient who develops aortic dissection is much poorer. Long term b - blocker administration can retard aortic dissection. Close follow up and more aggressive surgical treatment of asymptomatic aortic dilatation (diameter > 5 cm) reduce the incidence of aortic dissection. Mitral valve prolapse and mitral valve regurgitation are also common.

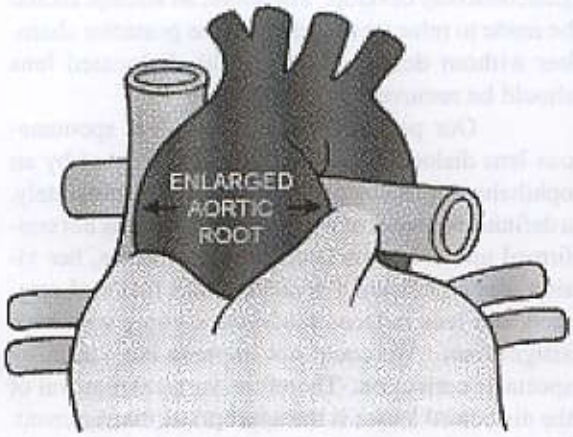

Figure 2. Diagram demonstrates a pear-shaped aneurysm of the ascending aorta.

Figure 3. Photograph of the aneurysm of the ascending aorta.

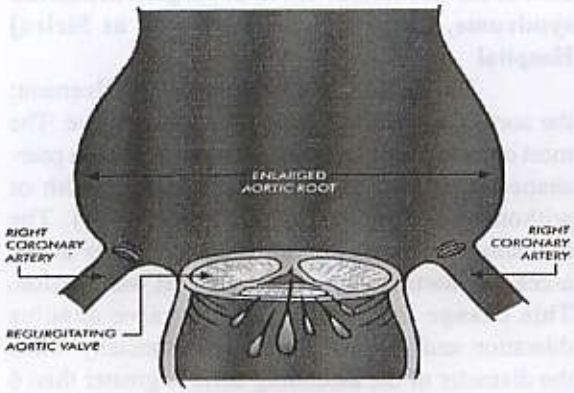

Figure 4. Diagram demonstrates the aneurysm of the ascending aorta with aortic regurgitation.

Figure 5. Photograph of aortic dissection.

Indications for surgery

Annulo-aortic ectasia, aneurysm

Asymptomatic annulo-aortic ectasia with ascending aorta diameter > 5 cm or enlarging asymptomatic annulo-aortic ectasia > 5mm/year (Appendix A) (This recommendation should be influenced by age, expected normal aortic size (Appendix B), family history of dissection, etc.)

Aortic aneurysm at another site – diameter > 5 cm or symptomatic (Surgeons tend to be more aggressive in operating earlier in Marfan patient)

Aortic dissection

Acute dissection involving ascending aorta (type A)– immediate repair

Chronic dissection involving ascending aorta – elective repair

Acute dissection not involving ascending aorta (type B) – medical treatment. Surgery is indicated if complications arise such as leakage, vascular compromise, or persisting pain.

Chronic dissection not involving ascending aorta (type B) – medical treatment. Surgery is indicated if complications arise such as aneurysmal dilatation > 5 cm.

Valve regurgitation

Significant, progressive valvular regurgitation – Symptoms, severity of regurgitation on echocardiogram/ cardiac cath. and LV dimension/ function are important factors to determine when to operate.

Surgical procedures

Surgical procedure for annulo-aortic ectasia

Bentall-de Bono operation (Appendix C)

Replace the aortic root and aortic valve with composite graft (vascular graft with attached prosthetic valve) and re-implant the coronary arteries Valve sparing – procedure for repair of an ascending aortic aneurysm – aortic valve may be repaired,

Graft replacement and aortic valve repair

Risk factors for hospital mortality in Marfan patient undergoes surgical repair for cardiovascular complication are poor functional class and urgent surgery.

Siriraj experience

There have been 20 patients with Marfan syndrome who have undergone cardiac operations at Siriraj hospital between 1979 and 2000. The age of the patients at first operation ranged between 4 – 47 years old (mean 30). There were 12 male and 8 female patients, three of these were siblings. Six of them also had aortic dissection which were chronic type A in 3, chronic type B in 2 and acute type B in 1. During this period there were around 100 patients who underwent thoracic aortic operations, 16 of them were Marfan patients who underwent a Bentall operation. The result of the Bentall procedure in Marfan patients was fairly good. There were 2 deaths (1 hospital death, and 1 late death) and the other 2 patients were lost from the follow up clinic. Twelve patients attended the clinic last years, all were in functional class I (NYHA). One patient also had a thoracoabdominal aneurysm repaired on two occasions apart from the Bentall operation. The longest survival was 8 years and the mean length of follow up was 4 years. There were 3 Marfan patients who underwent mitral valve replacement with one late death and the other two were lost from the follow up clinic. The only Marfan patient who underwent aortic valve replacement is doing well.

CONCLUSION

We present a case of Marfan syndrome with bilateral spontaneous lens dislocation. Proper management of this condition and indication for surgery are discussed. In addition, the role of the cardiovascular-thoracic surgeon is also presented.

Appendix A

The rate of growth of the aortic diameter averages 2 mm per year.

Appendix B

The equation for aortic root diameter for height in tall men and women (>188cm and 175 cm, respectively) is

Aortic diameter = -45.9 + (0.493 height (cm) – 0.001) x 2

In children and growing teenagers, the equation is

Aortic root diameter = 24 $(BSA\ m^2)^{1/3}$ + 0.1 (age) – 4.3

Appendix C

The Bentall operation is a procedure, which uses a composite graft (a portion of tube graft with attached prosthetic valve) to replace the aortic root (Figure 6). It is widely used in case of aortic root dilatation with severe aortic valve insufficiency. The aortic root is removed (sinus and tubular portion), leaving the cuffs of the coronary artery orifices. The aortic valve leaflets may or may not be excised. The composite graft is placed, then the origins of the coronary arteries are re-implanted into the graft. (Figure 7)

Sometimes, when the aortic valve annulus is not severely affected, the aortic valve regurgitation may be repaired, avoiding the risk of a prosthetic

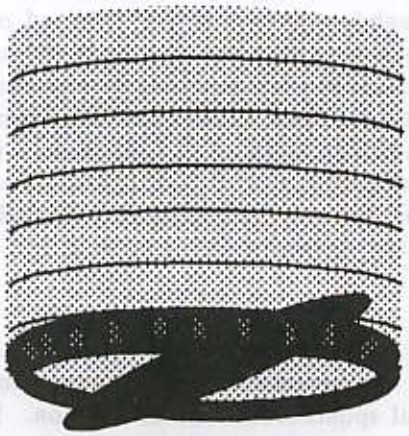

Figure 6. Diagram of a composite graft.

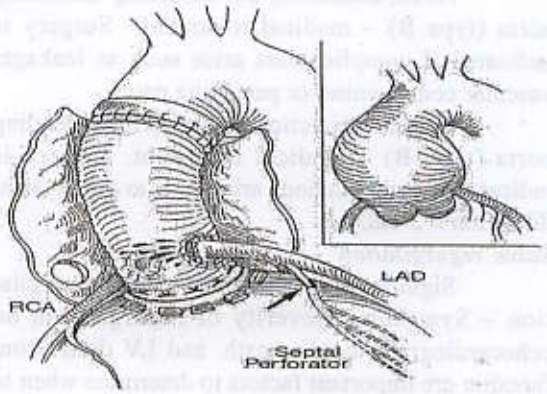

Figure 7. The Bentall operation.

valve and anticoagulant-related complication. However the long-term result of valve repair in connective tissue disease may not be promising. The coronary arteries may not need to be re-implanted as the sinus portion of the aortic root can usually be preserved.

ACKNOWLEDGEMENTS

The authors thank Drs. Pradit Punjavenin and Chanin Limvongsre from the Department of Internal Medicine for participation in the interdepartmental conference.

REFERENCES

1. Bjerrum K, Kessing SV. Congenital ectopia lentis and secondary buphthalmos likely occurring as an autosomal recessive trait. Acta Ophthalmol (Copenh) 1991; **69:** 630-34.
2. Tyagi AK, McDonnell PJ. Pseudoexfoliation syndrome and spontaneous lens dislocation. Eye 1998; **12:** 1033-34.
3. Rapkin JS, Bogorad DD. Bilateral dislocation of the crystalline lens in a patient with presumed syphilitic uveitis. Henry Ford Hosp Med J 1986; **34:** 207-10.
4. Halpern BL, Sugar A. Retinitis pigmentosa associated with bilateral ectopia lentis. Ann Ophthalmol 1981; **13:** 823-24.
5. David R, MacBeath L, Jenkins T. Aniridia associated with microcornea and subluxated lenses. Br J Ophthalmol 1978; **62:** 118-21.
6. Gillum WN, Anderson RL. Dominantly inherited blepharoptosis, high myopia, and ectopia lentis. Arch Ophthalmol 1982; **100:** 282-84.
7. Traboulsi EI, Whittum-Hudson JA, Mir SH, Maumenee IH. Microfibril abnormalities of the lens capsule in patients with Marfan syndrome and ectopia lentis. Ophthalmic Genet 2000; **21:** 9-15.
8. Taylor RH, Burke J, O'Keefe M, Beighi B, Naughton E. Ophthalmic abnormalities in homocystinuria: the value of screening. Eye 1998; **12:** 427-30.
9. Nagata M, Takagi S, Yamasaki A, Tsunematsu S, Kumagami T, Itamochi C, et al. Histopathological study of microspherophakia in the Weill-Marchesani syndrome. Jpn J Ophthalmol 1995; **39:** 89-95.
10. Beemer FA, Duran M, Wadman SK, Cats BP. Absence of hepatic molybdenum cofactor. An inborn error of metabolism associated with lens dislocation. Ophthalmic Paediatr Genet 1985; **5:** 191-95.