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Bilateral Nasal Ectopia Lentis with no Skeletal Abnormality: Is it Marfan Syndrome?

İskelet Anomalisi Olmaksızın Bilateral Nazal Lens Ektopisi: Marfan mı?

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Summary

To report three male Marfan patients from one family, with no skeletal anomalies, but with nasally subluxated lenses in two and bilateral total crystalline lens dislocation into the vitreous cavity in one of them. Ophthalmological, cardiological, orthopaedic evaluation. Although none of the patients had any skeletal abnormalities, all three had ophthalmological involvement. Cardiological examination revealed mitral valve prolapse and aortic root dilatation in all patients. Associated grade 1tricuspid valve insufficiency was present in one patient.

Conclusion: Whenever bilateral ectopia lentis is observed, Marfan syndrome should be suspected, and careful systemic evaluation should be carried out in order to avoid overlooking the other systemic life-threatening failures. *(TOD Dergisi 2010; 40: 245-7)*

Key Words: Ectopia lentis, Marfan Syndrome

Özet

Aynı aileden iskelet anomalisi olmayan üç erkek-iki nazal lens subluksasyonu, bir vitreusa total kristallin lens dislokasyonu-Marfan olgusunu sunmak. Oftalmolojik, kardiyolojik, ortopedik inceleme. Olguların hiçbirinde iskelet anomalisi olmamasına rağmen, üçünde de oftalmolojik tutulum mevcuttu. Kardiyolojik bakıda tüm olgularda mitral kapak prolapsusu ve aort kökü dilatasyonu saptandı. Olgulardan birinde eşlik eden evre 1 triküspit kapak yetmezliği olduğu belirlendi. Lens ektopisi saptandığında - ektopi ne tarafa olursa olsun-dikkatli oftalmolojik muayene yapılmalıdır. İskelet bulguları mevcut olmasa da, hayatı tehdit eden sorunları atlamamak için detaylı kardiyolojik muayene de yapılmalıdır. *(TOD Journal 2010; 40: 245-7)*

Anahtar Kelimeler: Lens ektopisi, Marfan Sendromu

Introduction

Marfan syndrome is an autosomal dominant connective tissue disorder characterized by skeletal, cardiovascular and ocular anomalies (1). The fibrillin-1 gene (FBN1) located on choromosome 15 is reported to be defective in this syndrome (1,2).

Ectopia lentis, which is almost always bilateral, symmetric and in superior/superotemporal direction, is the most common ocular manifestation in Marfan syndrome (1,3,4). Other less reported ocular manifestations include enophthalmos, strabismus, axial myopia, increased corneal diameter, anterior chamber angle abnormalities, glaucoma, and retinal pathologies (1,5).

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We report on three family members with Marfan syndrome with no skeletal anomalies-two with bilateral nasal ectopia lentis and one with bilateral total crystalline lens dislocation into the vitreous cavity.

Cases

We present three male patients: a father (Patient 1), his son (Patient 2), and his elder brother (Patient 3) (Table 1) (Figure 1). Bilaterally, the crystalline lenses of Patient 3 were totally dislocated in the inferior quadrant of the vitreous, causing no inflammatory reaction. Remaining ophthalmological examinations were normal.

The patients underwent a detailed systemic evaluation to detect the presence of any skeletal or cardiovascular abnormalities. Cardiological examination revealed mitral valve prolapse and aortic root dilatation in all patients. Additionally, Patient 2 had grade 1 tricuspid valve insufficiency. None of the patients had any skeletal anomalies.

Discussion

Ectopia lentis may occur as an isolated disorder, accompany other ocular abnormalities, or be associated with systemic syndromes (6). Marfan syndrome is the most frequent cause of heritable lens subluxation.

To make the diagnosis of Marfan syndrome more consistent and of more prognostic value, the clinical features were codified as the Ghent nosology in 1996 (7). In the Ghent nosology, clinical features are assessed within seven body 'systems' (skeletal, ocular, cardiovascular, pulmonary, skin/integument, dura, genetic findings), to determine whether that system provides a major criterion, or only system involvement. A diagnosis of Marfan syndrome requires a major criterion in two systems and involvement of a third. The cardiovascular, ocular and skeletal systems can provide major criteria, or system involvement, the pulmonary system and skin/integument can provide only system involvement, the dura and family/genetic history provide only major criteria (7). Other features associated with Marfan syndrome include dural ectasia, spontaneous pneumothorax, recurrent hernia, and striae atrophicae, and all of them are considered as minor findings. In the absence of family history, a person should display major criteria in at least two organ systems and involvement of a third organ system for Marfan syndrome diagnosis (7). In the presence of family history, a person should display one major criterion in an organ system and involvement of a second organ system (7).

Lens subluxation, the most common ocular abnormality, may occur in 50% to 80% of Marfan patients (3,4). It tends to be bilateral and symmetric, and is reported to be the most specific finding (78%) in Marfan syndrome (8). The lens is almost always displaced towards superior or superotemporal direction, and total dislocation into the vitreous cavity is uncommon (1,3,4). Our Patients 1 and 2 had bilateral nasal ectopia lentis, and Patient 3 had bilateral total lens dislocation into the vitreous cavity with no inflammatory reaction.

Skeletal anomalies in Marfan syndrome include excessive height relative to family members, arachnodactyly, joint laxity, scoliosis and anterior chest deformities (7,9). None of our patients demonstrated any skeletal abnormalities.

Cardiovascular manifestations in Marfan syndrome include mitral valve prolapse, aortic root dilatation and dissecting aortic aneurysm (10). Although having no skeletal abnormalities supporting Marfan syndrome, all three patients had mitral valve prolapse and aortic root dilatation. Patient 2 had associated grade 1 tricuspid valve insufficiency. The most life-threatening complication of Marfan syndrome is thoracic aortic aneurysms leading to

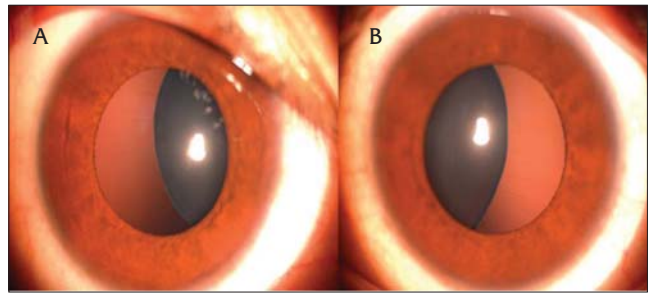

Figure 1. A, B. Bilateral nasal ectopia lentis in Patient 1. Zonular fibres are in sight

Table 1. Clinical characteristics of the patients

	Age	Eye	Refractive Error	Visual Acuity	Lens OD/OS	Fundoscopic Appearance	Axial Length (mm)
Patient 1	41	OD	+10,00-1,50x10	20/200	Nasally subluxated	Normal	24,0
		OS	+10,00-1,00x20	20/200	Nasally subluxated	Normal	24,2
Patient 2	18	OD	+9,00+1,00x90	20/200	Nasally subluxated	Normal	24,5
		OS	+9,50+0,75x85	20/200	Nasally subluxated	Normal	24,3
Patient 3	48	OD	+10,50+0,75x80	20/200	Dislocated in the vitreous	Normal	24,0
		OS	+8,00+0,50x100	20/400	Dislocated in the vitreous	Normal	24,8

aortic dissection, rupture, or both (11). The most common cardiovascular complication in Marfan syndrome is progressive aortic root enlargement (11). As the success of current medical and surgical treatment of aortic disease in Marfan syndrome has substantially improved the average life expectancy, extending it up to 70 years (11), it is so important to diagnose the cardiological disorders as early as possible.

When nasal ectopia lentis is detected in members from one family with no metabolic disorders and no skeletal anomalies, Marfan syndrome is hardly considered. Familial ectopia lentis, an autosomal dominant disorder, in which no systemic disorder takes place, would be the first diagnostic choice. Homocystinuria, which affects the metabolism of the amino acid methionine, has several features in common with Marfan syndrome. It has autosomal recessive inheritance pattern and ectopia lentis is usually inferonasal (3). As our patients had family history and major criteria in the cardiovascular and ocular systems, they were diagnosed as having Marfan syndrome (7).

To the best of our knowledge, bilateral nasal ectopia lentis in Marfan syndrome has not been reported; therefore, the ophthalmologist must be aware even if the direction of the ectopia is atypical. In patients with ectopia lentis, Marfanoid habitus is not a sine qua non, as skeletal changes are not seen in one third of the subjects with Marfan syndrome (12). Considering that 93% of patients die of cardiovascular diseases and the life expectancy without treatment is 32±16 years, early diagnosis of the disease and its life-threatening sequela is mandatory (12).

Whenever ectopia lentis is observed, regardless of its direction and even if skeletal findings are absent, careful ophthalmologic, orthopaedic and cardiological examination should be performed in order to establish an early diagnosis.

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