Keratoglobus Associated with Hypertrophic Cardiomyopathy: A Case Report of a Concomitant Disorder of Heart and Eye

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Abstract
A 30-year-old woman was referred to our department for a cardiology visit. She had a medical history of ophthalmologic disorders. Her chief complaints were dyspnea, lightheadedness, and fainting after the Valsalva maneuver. Physical examination showed systolic murmurs at the left upper sternal border as well as corneal thinning and bulging in the 2 eyes. Transthoracic echocardiography revealed local significant hypertrophy in the base of the interventricular septum with significant left ventricular outflow obstruction and severe mitral regurgitation.

Keywords: Keratoglobus, Hypertrophic Cardiomyopathy, LVOT Obstruction

1. Introduction
Keratoglobus is an ecstasy of both sides of the cornea with the pathology of corneal thinning, and this thinning is more evident at the peripheral area (1, 2). Various anomalies such as hearing loss, cardiac disorders, and dental anomalies have been reported in association with keratoglobus.

2. Case Presentation
A 30-year-old woman who worked in a ladies’ hairdressing salon presented with a history of dyspnea, lightheadedness, and fainting after the Valsalva maneuver. Her condition had started 9 months earlier but exacerbated in the preceding 3 weeks. Interestingly, an examination of the eyes revealed that the keratoglobus in both eyes had resulted in spherical and clearly enlarged eyes (Figure 1A, black arrow). The heart auscultatory finding was harsh systolic murmurs at the left upper sternal border, which was increased by the Valsalva maneuver. There was no history of premature sudden cardiac death and hypertrophic cardiomyopathy in her family members. Transthoracic echocardiography showed local significant hypertrophy (septal thickness = about 2.2 cm in the diastolic period) in the base of the interventricular septum (Figure 1B, yellow arrow) with a turbulent flow during systole in the left ventricular outflow tract (LVOT) (Figure 1C, black arrow) and normal thickness in the other segments. Further, there was localized interventricular septal hypertrophy concomitant with systolic anterior motion of the mitral valve leaflets, resulting in a dynamic outflow-tract obstruction. Doppler study quantified a high LVOT gradient, which clearly exaggerated with provocative maneuvers (peak pressure gradient = 110 mmHg). Also, left atrial enlargement and severe mitral valve regurgitation (Figure 1C, white arrow) were noted due to the systolic anterior motion of the mitral valve and also thickening of the leaflet tips. During 2 hospital admissions, electrocardiographic (ECG) monitoring did not record arrhythmias during episodes of dizziness and lightheadedness. There was no significant improvement in the symptoms or the LVOT gradient after full medical treatment, including administration of β-blockers; the patient was, therefore, considered for cardiac surgery. Cardiac magnetic resonance imaging confirmed localized hypertrophic cardiomyopathy. Septal myectomy and mitral valve repair were performed. The patient had an uneventful recovery and at 1-year follow-up, she was asymptomatic. Thus, we herein reported an association between keratoglobus and an atypical form of hypertrophic obstructive cardiomyopathy.

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3. Discussion

Keratoglobus is a noninflammatory and nonprogressive corneal ecstacy which is often bilateral and results in irregular astigmatism and high myopia. Acquired cases

are also seen accompanied by exophthalmia, hyperthyroidism, keratopathy, and trauma (1). The keratoglobus etiology still remains uncertain. Cardiac anomalies have been seen in association with keratoglobus. Due to connective tissue disorder with abnormal collagen synthesis in some congenital form of keratoglobus, the possibility of the concurrency of valve problems, leading to prosthetic heart valve replacement, has been reported. Case reports have presented an association between keratoglobus and coarctation of the aorta, multiple aneurysm formation in the ascending aorta, pulmonary artery, and brachiocephalic artery, patent ductus arteriosus and prolapse of the mitral valve leaflets (2). There are different syndromes in which the concurrency of heart and eye problems occurs. Brittle cornea syndrome is an autosomal recessive syndrome caused by mutation in ZNF469 and PRDM5 genes. In this syndrome, eye disorders such as progressive keratoconus, and keratoglobus and retinal detachment as well as hearing loss, musculoskeletal and skin problems are seen. In this group of patients, mitral valve dysfunction and cardiovascular symptoms have been seen, which shows the need for echocardiography in these patients (3). Another syndrome, which expresses concurrent mitral valve involvement with eye problems, is Stickler syndrome. This syndrome is a progressive arthro-ophthalmopathy, in which the concurrency of the ocular or facial and skeletal systems is clearly visible (4). This syndrome is autosomal dominant. The primary reports revealed that mitral valve prolapse was common between the patients; nonetheless, the subsequent reports demonstrated a reduction in the prevalence of mitral valve involvement (5). In the evaluation of these patients, echocardiography is recommended if the mitral valve involvement is suspected. Marfan syndrome is an autosomal dominant disease with a prevalence of 1 per 3,000 to 5,000 people (6). It is a connective tissue disorder first reported in a 5.5-year-old girl in 1896 (7). In this syndrome, ocular involvement is seen as ectopia lentis and the patients predispose to retinal detachment (8) and cardiovascular disorders such as mitral (9, 10) and aortic valve disease (11), coarctation of the aorta, atrial septal defect, pulmonary artery stenosis, patent ductus arteriosus, and persistent left superior vena cava present in infancy (7).

The current case report introduced a patient with the concurrency of ocular involvement in the form of keratoglobus and cardiac involvement in the form of hypertrophic obstructive cardiomyopathy, which is a rare associated anomaly.

References