A 26-year-old woman was referred to our hospital for further evaluation of a grade three systolic murmur at the apex revealed during routine examination. She was free of symptoms on arrival. Findings at physical examination were as follows: joint laxity, positive wrist and thumb sign, and facial appearance for Marfan syndrome (MFS). However, she did not have striae atrophicae or recurrent or incisional hernias.

Transthoracic two-dimensional echocardiogram revealed mitral leaflet thickening and a prolapse of the anterior mitral leaflet (A3 segment) (Figure 1). The ejection fraction was 68.6% with a left ventricular diameter of 64 mm in diastole and 36 mm in systole. The color Doppler image displayed moderate mitral regurgitation directed toward the posterior portion of the left atrium (Figure 1). However, of the major diagnostic criteria of MFS, we did not observe dilatation of ascending aorta, enlargement of aortic root, or aortic regurgitation [1]. The clinical diagnosis of MFS depends on a combination of major and minor signs defined in the revised 1996 Ghent Nosology [1]. We proceeded to check other major signs. Ophthalmologic evaluation revealed that she did not have any abnormal findings, such as ectopia lentis. No abnormality was observed on chest x-ray films. Furthermore, none of the patient’s first-degree family members has Marfan syndrome. We could find only one major sign (skeletal features) and one minor sign (mitral valve prolapse), but not enlargement of aortic root. In such patients, it would be useful to detect dural ectasia by MRI in the diagnosis of MFS.

**Discussion**

Mitral valve prolapse (MVP) is caused by not only degeneration of valve, but also ischemia [3] and infection [4]. A previous report showed that MVP was
noted in 35% to 100% of patients with MFS [5]. Our patient had already shown left ventricular dilatation. However, echocardiographic findings did not reach indication of operation with an ejection fraction of 68.6% (>60%) with a left ventricular diameter of 36 mm in systole (<45mm). Furthermore, we did not detect dilatation of ascending aorta, enlargement of aortic root, or aortic regurgitation. It has been reported that mitral regurgitation in women with MFS is more progressive than those without MFS, and patients with MFS often develop aortic root enlargement [6]. Thus, we have to check left ventricular chamber size, ejection fraction, grade of mitral valve regurgitation, and diameter of aortic root carefully by echocardiogram.

The diagnosis of MFS is difficult because of the wide variability of clinical signs and the low specificity of many of the clinical signs [2]. Indeed, our patient showed only one major sign and one minor sign in the doctor’s office. Dural ectasia is also detected in patients with other diseases such as Ehlers-Danlos syndrome, neurofibromatosis type 1, ankylosing spondylitis, trauma and scoliosis. However, a previous report showed that dural ectasia was identified in 92% of patients with MFS[2]. Therefore, dural ectasia is considered to be a highly characteristic sign of Marfan’s syndrome [2]. Our patient had typical skeletal findings of MFS and displayed mitral valve prolapse, but not enlargement of aortic root. In such patients, it would be useful to detect dural ectasia by MRI in the diagnosis of MFS.

References

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Fig. 1. Parasternal long-axis echocardiogram recorded (A) in diastole (left) and end-systole (right). Note the mitral leaflet thickening (white arrow). The size of aortic root was normal. The color Doppler image displayed moderate mitral regurgitation directed toward the posterior portion of the left atrium (B). The B-mode echocardiogram at the A3 segment (C) revealed a prolapse of anterior mitral leaflet (white arrow) more clearly compared to Figure A (A2 segment). Parasternal short-axis view recorded (D) in diastole (left) and end-systole (right). Note the prolapse of the anterior leaflet (A3 segment, yellow arrow). A color Doppler image also recorded in systole (E). Note the mitral regurgitation jet directed posteriorly (black arrow).
Fig. 2. A sagittal T2-weight spin-echo MR image (left) showed bulging of dural sac and scalloping of vertebral bodies in lumbar segment. A transverse T2-weight spin-echo MR image (right) revealed extensive dural ectasia (arrows) and showed that it extended into the soft tissues of the lower back and into the presacral space.