Myotonia Dystrophica and Mitral Valve Prolapse*

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Two siblings are described, both afflicted with myotonia dystrophica and mitral valve prolapse. This family supports the recent association of these two familial diseases. One of the siblings had severe conduction disease and recurrent ventricular tachycardia, possibly reflecting potentiation of arrhythmia, because of the association of these two familial diseases.

In a previous report, Winters et al described the simultaneous occurrence of myotonia dystrophica and mitral valve prolapse in one family. In a subsequent report, Biddison and co-workers reported four families with X-linked muscular dystrophy (an unrelated myopathy) and mitral valve prolapse. It is not clear whether these reported cases reflected a true association (as opposed to chance association) between these myopathies and familial mitral valve prolapse.

We describe two siblings with both myotonia dystrophica (with cardiac involvement) and mitral valve prolapse. Report of our family supports the probability of a true association between these two apparently unrelated familial diseases. In addition, one of these afflicted family members had serious conduction defects, typically associated with myotonic dystrophy, as well as recurrent ventricular tachycardia (typically associated with mitral valve prolapse), suggesting that mitral valve prolapse might adversely influence the course of myotonia dystrophica.

CASE REPORTS

Case 1 (Fig 1)

A 35-year-old woman was referred to the University of Illinois Hospital for evaluation of syncope, A-V block, and ventricular dysrhythmia. Myotonia dystrophica was diagnosed at the age of 32 years and confirmed by appropriate studies. For the five years before admission, the patient complained of intermittent palpitation. Although atrial and ventricular premature beats were documented, she had not received antiarrhythmic therapy.

One month before the present admission, she had a syncopal episode for which she was hospitalized and released without definite diagnosis and without therapy. Three weeks later syncope recurred. She was then transferred to our institution for further evaluation.

Physical examination disclosed a myopathy with typical facies of myotonia dystrophica. Blood pressure was 100/70 mm Hg, and the peripheral pulse rate was 70 beats per minute and regular. Bilateral cataracts were present. The cardiac apical impulse was prominent and located in the

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Figure 1. Pedigree of the propositus' family.
CASE 1

The patient's father was also evaluated. At age 78 years, he was asymptomatic. Cardiac examination disclosed a midsystolic murmur. Neurologic examination revealed the typical facial appearance of myotonic dystrophy with frontal baldness. Percussion and action myotonia were evident. The ECG revealed left anterior hemiblock.

A M-mode echocardiogram showed a typical pattern of midsystolic mitral valve prolapse (Fig 2).

CASE 2

The patient's only brother was also evaluated. At age 33 years, he is asymptomatic. Cardiac examination disclosed a midsystolic click without murmurs. Neurologic examination revealed the typical facial appearance of myotonic dystrophy with frontal baldness. Percussion and action myotonia were evident. The ECG revealed left anterior hemiblock.

A M-mode echocardiogram showed a typical pattern of midsystolic mitral valve prolapse (Fig 2).

CASE 3

Records of our patient's deceased father were reviewed. At the age of 60 years he was hospitalized for work up of a six month history of muscle weakness. Physical findings and electromyography were reported as those of typical myotonia dystrophica. The cardiovascular examination result was reported as normal. An ECG showed nonspecific ST-T changes with an absent septal vector, and the patient was receiving procainamide for an unknown cause, possibly arrhythmia. No echocardiogram was performed.

CASE 4

The patient's mother was evaluated for presence of mitral valve prolapse. At the age of 65 years she was free of cardiovascular symptoms and had a long history of pulmonary emphysema. On physical examination no click or murmur was detected. Echocardiogram showed no evidence of mitral valve prolapse.

Figure 2. Burst of polymorphous ventricular tachycardia (torsade de pointes) observed in case 1.

Figure 3. Echocardiogram of case 1 showing pansystolic mitral valve prolapse (arrows).
DYSOTOINIC PROGRESSIVE MYOPATHY

Myotonia dystrophica is characterized by an autosomal dominant mode of inheritance and by slowly progressive myopathy. Cardiac involvement in this disorder has been well established by both clinical and pathologic findings. The ECG is the most sensitive indicator of cardiac involvement, conduction defects being the most common abnormalities encountered. Despite the frequency of conduction defects, the incidence of overt heart failure is low. Diagnostic catheterization has revealed abnormal hemodynamics and asynergy in several cases.

Progressive muscular dystrophy has also been associated with cardiovascular disease. As in myotonia dystrophica, the most frequent finding implicating cardiac involvement is derived from the ECG. In the X-linked pseudohypertrophic dystrophy of Duchenne, there is a distinctive ECG pattern characterized by tall right precordial R waves and deep Q waves in limb and lateral precordial leads. As in myotonia dystrophica, congestive heart failure is not usually a major or frequent clinical finding.

Mitral valve prolapse is seen in a multiplicity of clinical settings. Mitral valve prolapse is often idiopathic and sometimes transmitted as a familial trait. The pathologic substrate for the idiopathic variety appears to be myxomatous degeneration of the mitral valve leaflets and chordae tendineae. The complications of mitral valve prolapse include severe ventricular arrhythmias as observed in our patient and a small but definite risk of sudden death.

Possible Mechanism of the Association of Myopathy and Mitral Valve Prolapse

The question arises whether the association of these various myopathies and mitral valve prolapse is real or a result of chance. Familial myopathies are

Reports of Familial Mitral Valve Prolapse and Myopathy

Winters et al presented a study of 25 relatives of a patient with associated mitral valve prolapse and myotonia dystrophica. Of the 25 relatives, eight had both myotonia dystrophica and mitral valve prolapse, 14 had neither, two had myotonia dystrophica alone, and one had mitral valve prolapse alone. The authors also reported 19 additional patients with myotonia dystrophica and found five unrelated patients from separate kindreds with evidence of mitral valve prolapse.

Biddison et al noted an association of mitral valve prolapse and X-linked muscular dystrophy. A total of four families were scrutinized (three with the Duchenne type and one with the Becker type of muscular dystrophy). In all of the families, the first-generation female carrier had evidence of mitral valve prolapse. Of the 13 second- and third-generation males, five were afflicted with both muscular dystrophy and mitral prolapse, five with muscular dystrophy alone, one with mitral valve prolapse alone, and two with neither. Of five second-generation females, two were possible carriers and two were definite carriers of muscular dystrophy, three of four had evidence of mitral valve prolapse. One second-generation female was normal.

One additional study of interest is the recent report by Isner et al demonstrating a 26 percent incidence of mitral valve prolapse in 88 patients with peroneal muscle atrophy (Charcot-Marie-Tooth disease). This study emphasized the general lack of cardiovascular disease in patients with peroneal muscle atrophy.

FIGURE 4. Echocardiogram of case 2 showing a midsystolic mitral valve prolapse (arrows).
uncommon, though mitral valve prolapse is relatively frequent. One would certainly expect to see an occasional patient with myopathy and unrelated mitral valve prolapse. However, one can gather some support for a true association of myopathy and prolapse. This support would be as follows: 1) the description of two families with familial myotonia dystrophica and familial mitral valve prolapse (one family reported by Winters et al\(^1\) and the family reported in this article); 2) the description of four families with familial progressive muscular dystrophy and familial mitral valve prolapse;\(^2\) 3) the high incidence of sporadic mitral valve prolapse (not proven to be familial) reported with myotonia dystrophica (26 percent),\(^1\) and peroneal muscle atrophy (26 percent).\(^1\)

Accepting a true association between mitral valve prolapse and some myopathies, one must still explain this association. If we were trying to explain only a single family with both diseases, we could postulate a chance association of two familial genes. However, in view of the multiple families and affected individuals from different families, this does not seem to be applicable.

The possibility exists that the reported association of familial myopathy and prolapse reflect pleiotropy, with one gene having several expressions (these expressions being myopathy and myxomatous degeneration of the mitral valve). In support of this hypothesis, one can note the demonstration of myxomatous degeneration of the valve in the one autopsied case of progressive muscular dystrophy and mitral valve prolapse.\(^3\) There is another possibility, that mitral valve prolapse reflects cardiomyopathy (related to myopathy) involving the papillary muscles, which then results in functional prolapse (without myxomatous degeneration). This is an attractive hypothesis, but is at least partially inconsistent with the occurrence of mitral valve prolapse without myopathy in some members of afflicted families. However, in these specific individuals, it is still possible that papillary muscle dysfunction (cardiomyopathic) is an expression of underlying myopathy that is not clinically manifest.

**Clinical Significance**

The presence of mitral valve prolapse should be looked for in all patients with myotonia dystrophica, muscular dystrophy, and peroneal muscle atrophy. The association of these two entities in the same patient could represent a high risk group for syncope and sudden death, because both entities independently are known to be associated with severe arrhythmias. How myopathy and prolapse could modify each other is unknown. Further pathologic studies are needed in patients afflicted with various myopathies and mitral valve prolapse to further delineate the pathologic basis for prolapse. Clinical studies are also warranted to determine the significance of mitral valve prolapse in regard to the presence of ventricular dysrhythmia in the myopathic patient.

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