Atrial Flutter and Myotonic Dystrophy in a Male Adolescent Treated With Radiofrequency Catheter Ablation

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A variety of cardiomyopathies are due to familial disease. Most are primarily associated with cardiac involvement and can lead to hypertrophic, dilated, or restrictive cardiomyopathy. Myotonic dystrophy (MD) is a multisystem disease with autosomal dominant inheritance and variable penetrance. Cardiac diseases are important causes of morbidity and mortality in MD patients. Patients with primary MD should be carefully investigated with an electrocardiogram, stress test, and an echocardiogram to identify preclinical cardiac involvement and to prevent life-threatening complications. Any new onset of atrial flutter or atrial fibrillation in a young patient without any underlying cardiac abnormality should be investigated for underlying myopathy. The authors report on a male adolescent with MD who presented with atrial flutter. The patient had been diagnosed with MD at birth. He had an impaired ejection fraction of 38% to 45%. The patient described sharp chest pain in the retrosternal area, with no radiation, that was induced by exercise.

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Key words: Atrial flutter • Myotonic dystrophy • Electrophysiological study

A 16-year-old male adolescent with past medical history of myotonic dystrophy (MD) was evaluated by his pediatrician for recurrent episodes of shortness of breath and chest pain while playing basketball. An electrocardiogram (ECG) showed a newly recognized atrial flutter with variable block. No cardiac valvular abnormalities were appreciated on 2-dimensional echocardiogram, but the patient had an impaired ejection fraction of 38% to 45%. He was referred for further evaluation of new onset of atrial flutter and

possible cardiomyopathy. The patient described sharp chest pain in the retrosternal area, with no radiation, that was induced by exercise. The pain was relieved by resting for a few minutes. The patient did not experience any accompanying nausea, vomiting, diaphoresis, or dizziness.

The patient had been diagnosed with MD at birth. He was noted to have MD physical features, and he had a strong family history of MD, with relatives in 4 generations on his maternal side. His mother was 42 years old and had adult-onset of MD, and his sister had been diagnosed with MD at age 21. There was no family history of sudden cardiac death. The patient never smoked tobacco, consumed alcohol, or used illegal intravenous drugs. His only medication was aspirin (81 mg/d).

On physical examination, he was a well-developed, well-nourished 16-year-old with no dysarthria. His vital signs revealed a blood pressure of 112/80 mm Hg, heart rate of 86 beats/min, and weight of 130 pounds. Cardiovascular examination showed irregular heart rate with variable S1, normal S2, and absence of cardiac murmurs. The rest of the examination was normal except for clear wasting of the thenar muscles. Typical type I (counterclockwise) atrial flutter with variable atrioventricular block was prominent on the ECG (Figure 1).

Anticoagulation with warfarin for stroke prophylaxis was started because the patient had mild left ventricular systolic dysfunction on echocardiogram. After 4 weeks of therapeutic anticoagulation, he underwent successful cardioversion to sinus rhythm with 50 Joules of biphasic current. His follow-up echocardiogram showed a normal EF of 50% to 55% and a normal left atrium size of 2.5 cm.

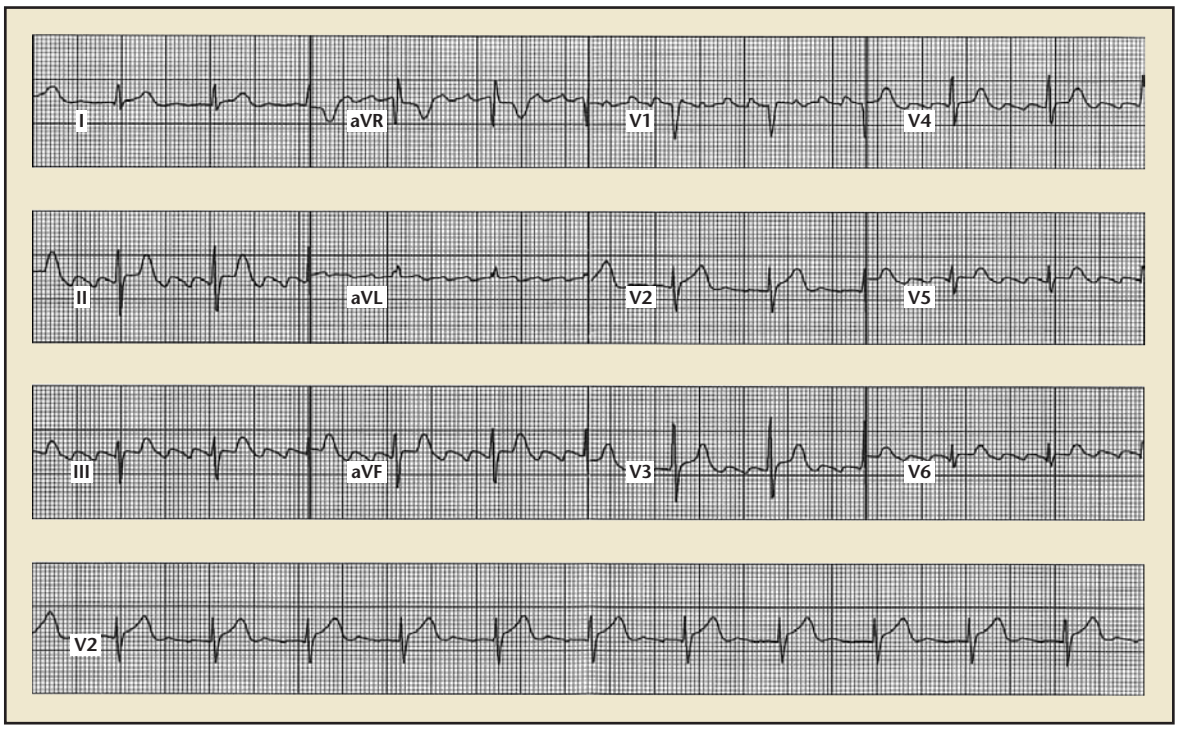
Six months after cardioversion, the patient was noted to have a recurrent atrial flutter. He was symptomatic and was referred for electrophysiological (EP) study and possible curative radiofrequency catheter ablation. Diagnostic EP study confirmed typical type 1 counterclockwise atrial flutter (Figure 2).

The patient underwent radiofrequency catheter ablation of the atrial flutter circuit with documentation of bi-direction conduction block. He did very well and was discharged home the same day. Warfarin was discontinued 3 months later because the patient did not have recurrent symptoms or atrial flutter on repeat ECG.

Discussion

Myotonia was initially described by Landau in 1952[1] as prolonged and excessive muscle contraction induced by repetitive firing of muscle

Figure 1. *This 12-lead electrocardiogram shows typical counterclockwise A flutter with variable atrioventricular block.* www.medreviews.com



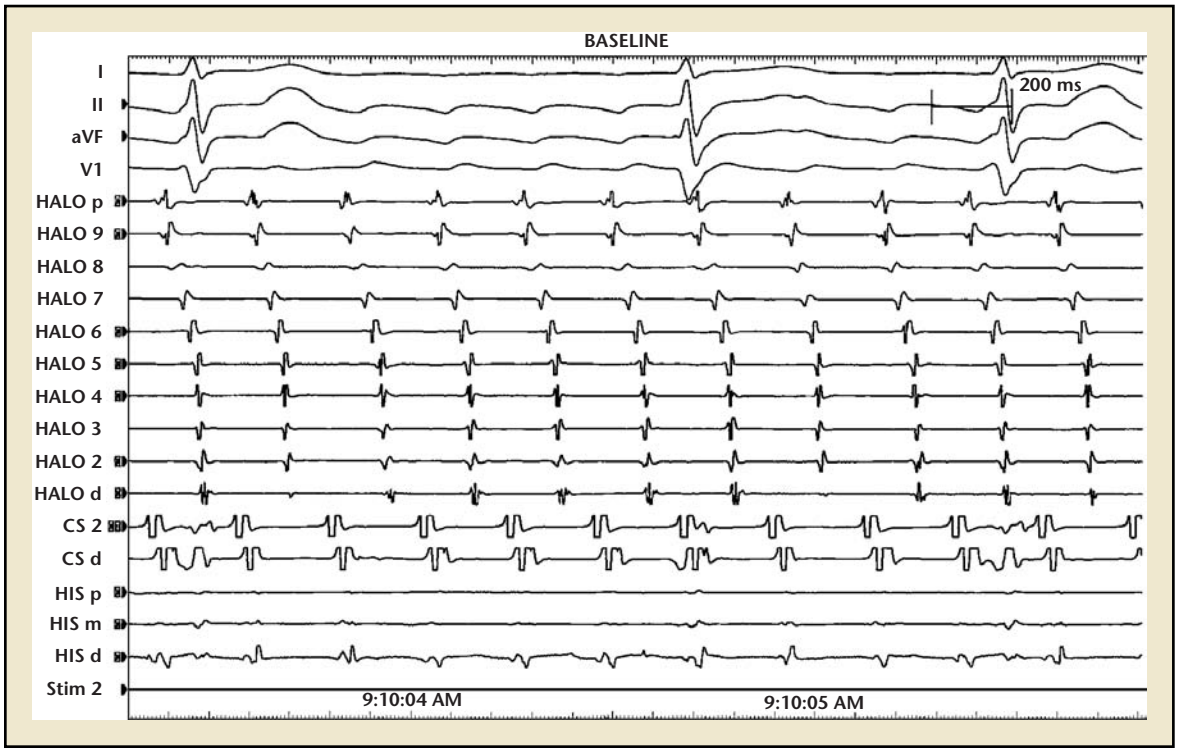


Figure 2. *Intracardiac electrogram during electrophysiological study showing typical counterclockwise A flutter.* www.medreviews.com

action potentials after mechanical stimulations have been stopped. MD is the most common form of myopathy in adults, and it affects both skeletal and myocardial muscles.[1] It was classified into 2 types according to clinical and genetic differences. Both types are inherited by autosomal dominant genes. Type 1 is related to a repeat in the CTG 3′ untranslated region of the MD protein kinases (DMPK) gene on chromosome 19q 13.3. Type 2 is related to the expansion of the CCTG tetraplet repeat in the intron 1 or the zinc finger 9 gene on chromosome 3q 21.3.[1] MD does not present with muscle weakness, but muscle atrophy is the prominent sign. Type 1 usually affects the distal muscles, and type 2 affects the proximal muscles.[1]

Other clinical presentations include frontal baldness, cataracts, and testicular atrophy. Sometimes, systemic involvement can be the primary presentation, with hypothyroidism, insulin-resistant diabetes mellitus, adrenal gland dysfunction, and hypogonadism.[1]

Cardiac disease is an important cause of morbidity and mortality; it is responsible for approximately 30% of deaths in patients with MD. MD type 1 patients are more vulnerable to cardiac complications (occurring in 7% to 23% of type 1 MD cases) compared with type 2.[2,3] The clinical presentations vary between cardiomyopathies, valvular diseases, arrhythmias, and sudden cardiac death.

In a longitudinal study of 367 MD patients followed for 10 years, 75 patients died (20%); the mean age at death was 53 years.[4] The main causes of death were respiratory problems (43%), probably as a consequence of respiratory muscle involvement; cardiovascular disease or sudden death (31%); and neoplasia (11%). Sudden cardiac death may result from conduction system disease or ventricular tachycardia.[5]

Although cardiac injury is very common in MD type 1 patients, extensive involvement of the myocardium is uncommon on histological examination. Few, if any, changes are usually seen, but they can include fatty infiltration, fibrosis, and atrophy of the cardiac conduction system.[6] Even in MD type 2, dilated cardiomyopathy and endocardial fibrosis have been reported, and myocardium ribonuclear inclusions have been noted on autopsies.[7] No histological signs of coronary artery disease were found to explain the sudden cardiac death in these patients.[2]

Cardiac involvement presents with different manifestations

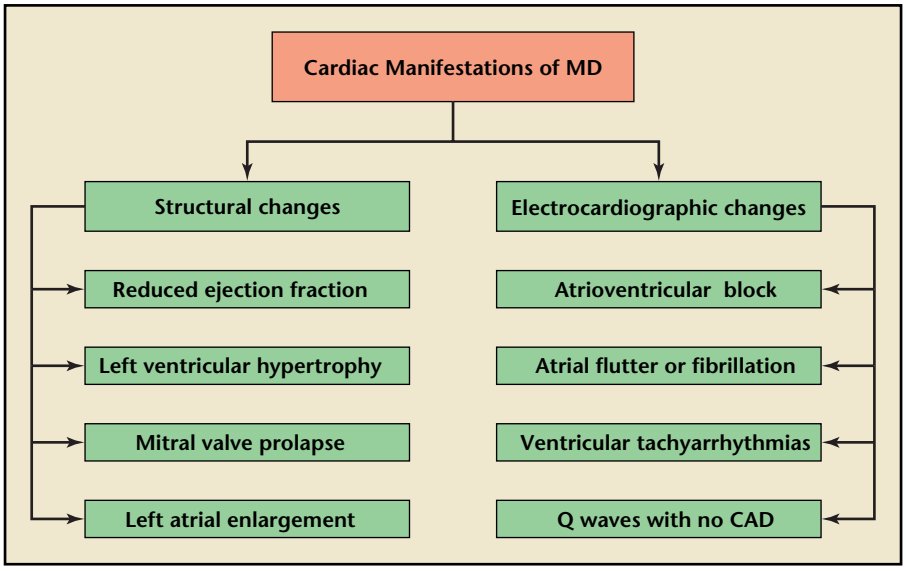


Figure 3. *Cardiac manifestations of MD. MD, myotonic dystrophy; CAD, coronary artery disease.*
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(Figure 3). Reduced left ventricular ejection fraction was seen in 50% of patients using transthoracic echocardiography.[8] The other echocardiographic abnormalities include left ventricular hypertrophy, mitral valve prolapse, and left atrial enlargement.[9]

The ECG changes include first-degree atrioventricular block and intraventricular conduction delay, with occasional progression to complete heart block.[10] An increased incidence of atrial fibrillation and ventricular tachyarrhythmias has also been reported, and in some patients, pathologic Q waves in the absence of coronary artery disease were identified.[11] The mechanism behind cardiac arrhythmia is thought to be related to fatty infiltrations causing delayed conduction in the His-Purkinje system. Pathological changes in the atrial muscular structure could establish substrate for reentry.[12] These histological changes might explain the failure of electrical cardioversion as long-term treatment of cardiac arrhythmias in MD patients.

Diagnosis

The diagnosis of this disease can be difficult. Sometimes, the first presentation can be cardiac arrhythmias. However, any new onset of atrial flutter or atrial fibrillation in a young patient without any underlying cardiac abnormality should be investigated for underlying myopathy.[12] In addition, after the diagnosis of MD in any patient is established, an important question would be raised about the risk of premature heart disease or even sudden cardiac death. So far, there are no proven criteria to predict these hazardous outcomes. Because of the progression in the myocardial fiber's changes, patients often have poor prognosis with advanced age and prolongation of the disease period. Male patients carry higher risk for acute cardiac events.[2]

Sabovic and colleagues[2] studied the relation between the expansion of CTG repeat in the DMPK gene and the cardiac involvement or sudden cardiac death in 63 MD patients, but they found no significant correlation. In young patients in whom cardiac complaints could be the first signs of MD, atrial flutter and ventricular tachycardia are more common than are conduction disorders and cardiomyopathy. These arrhythmias are more often associated with exercise, so a simple rest ECG is not sufficient to exclude them. Sports and physical exercise precipitates arrhythmias in over half of these patients.[11] Some authors strongly suggest that exercise testing with ECG monitoring be part of the routine evaluation of young patients with MD.[11] The delay in diagnosing asymptomatic arrhythmia might submit patients to serious complications, including sudden cardiac death. Not only should MD patients be screened for arrhythmias, but whenever MD is diagnosed, careful screening of other family members is important to identify affected members and establish their risk of sudden cardiac death. Screening should be followed by appropriate preventive methods.[11]

Any new onset of atrial flutter or atrial fibrillation in a young patient without any underlying cardiac abnormality should be investigated for underlying myopathy.

Conclusion

MDs are a clinically and genetically heterogeneous disease group. In the last few years, remarkable progress has been made in understanding the close and various relations between skeletal muscle disease and heart muscle disease. Cardiac involvement has been documented in a

number of primary MD cases, and is even the dominant feature in some of them. The myocardium can be affected in the form of a dilated cardiomyopathy, and the conduction system can show arrhythmias or conduction defects. Many patients with MD progress to terminal disease because of cardiac complications, such as sudden cardiac death and congestive heart failure. Patients with primary MD should be carefully investigated with an ECG, stress test, and an echocardiogram to identify preclinical cardiac involvement and to prevent life-threatening complications. There is a strong need for close collaboration between neurologists and cardiologists in order to provide optimal disease management for the affected MD patients. ■

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Main Points

- Myotonic dystrophy (MD) is the most common form of myopathy in adults, and it affects both skeletal and myocardial muscles. It is classified into 2 types according to clinical and genetic differences.
- MD does not present with muscle weakness, but muscle atrophy is the prominent sign. Type 1 usually affects the distal muscles, and type 2 affects the proximal muscles.
- Cardiac disease is an important cause of morbidity and mortality; it is responsible for approximately 30% of deaths in patients with MD.
- After the diagnosis of MD in any patient is established, an important question would be raised about the risk of premature heart disease or even sudden cardiac death.
- Not only should MD patients be screened for arrhythmias, but whenever MD is diagnosed, careful screening of other family members is important to identify affected members and establish their risk of sudden cardiac death.