A case of myotonic dystrophy presenting with ventricular tachycardia and atrial fibrillation

Ventrikül taşikardisi ve atriyal fibrilasyon ile seyreden miyotonik distrofi: Olgu sunumu

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Myotonic dystrophy type 1 (MD1) is an autosomal dominant disorder characterized by myotonia, progressive muscular weakness, cataract, and cardiac involvement. Cardiac involvement is common and includes conduction system abnormalities, supraventricular and ventricular arrhythmias, and less frequently, myocardial dysfunction and ischemic heart disease. A 54-year-old woman with a previous diagnosis of MD1 was admitted with palpitation, blood pressure of 157/118 mmHg, and a heart rate of 220 beat/min. Electrocardiography (ECG) showed ventricular tachycardia. Within minutes, hemodynamic collapse developed and electrical cardioversion was performed. Immediately following cardioversion, ECG showed atrial fibrillation, a slightly prolonged QT interval, and intraventricular conduction delay. After intravenous infusion of amiodarone, the rhythm converted to sinus. Transthoracic echocardiography showed significantly depressed left ventricular function, an ejection fraction of 25%, and normal coronary arteries. During electrophysiological study, atrium-His interval and His-ventricle interval were 120 msec was 54 msec, respectively, and monomorphic ventricular flutter was induced. An implantable cardioverter-defibrillator was placed. She was discharged in sinus rhythm.

Key words: Atrial fibrillation/etiology; defibrillators, implantable; electrocardiography; myotonic dystrophy/complications; tachycardia, ventricular/etiology.

Miyotonik distrofi tip 1 (MD1), miyotoni, ilerleyici kas güçsüzlüğü, katarakt ve kalp tutulumu ile seyreden otozomal dominant bir hastalıktır. Kardiyak tutulum sıktır ve daha çok ileti sistemi anormallikleri, supraventriküler ve ventriküler aritmiler şeklinde görülür. Daha az sıklıkta miyokart disfonksiyonu ve iskemik kalp hastalığı da görülebilir. Daha önce MD1 tanısı konmuş olan 54 yaşında kadın hasta, çarpıntı, 157/118 mmHg kan basıncı, 220 atım/dk kalp hızı ve elektrokardiyogramda ventrikül taşikardisi ile yatırıldı. Hastanın hemodinamik durumunun çok kısa sürede bozulması üzerine elektriksel kardiyoversiyon uygulandı. Kardiyoversiyonun hemen arkasından elektrokardiyogramda atriyal fibrilasyon, hafif uzamış QT intervali ve intraventriküler ileti gecikmesi gözlendi. İntravenöz amiodaron infüzyonundan sonra hasta sinüs ritmine döndü. Transtorasik ekokardiyografi incelemesinde sol ventrikül fonksiyonunun belirgin derecede zayıfladığı görüldü; ejeksiyon fraksiyonu %25 bulundu, koroner arterler ise normaldi. Yapılan elektrofizyolojik çalışmada, atriyum-His intervali ve His-ventrikül intervali sırasıyla 120 msn ve 54 msn ölçüldü ve uyarıyla monometrik ventriküler flutter oluşturuldu. Tedavi olarak kardiyak defibrilatör takılan hasta sinüs ritmiyle taburcu edildi.

Anahtar sözcükler: Atriyal fibrilasyon/etyoloji; defibrilatör yerleştirme; elektrokardiyografi; miyotonik distrofi/komplikasyon; taşikardi, ventriküler/etyoloji.

Myotonic dystrophy type 1 (MD1) is an autosomal dominant disorder caused by the mutational expansion of a repetitive trinucleotide sequence in the 3'-untranslated region of the myotonic dystrophy protein kinase gene on chromosome 19q13.3.[1] This disorder is characterized by myotonia, progressive muscular weakness, cataract, and cardiac manifestations. Cardiac involvement is common and involves conduction system abnormalities, supraventricular and ventricular arrhythmias, and less frequently, myocardial dysfunction and ischemic heart disease.[2]

CASE REPORT

A 54-year-old woman presented to the emergency room with palpitation, blood pressure of 157/118 mmHg, and a heart rate of 220 beats per min. Electrocardiography (ECG) showed ventricular tachy-

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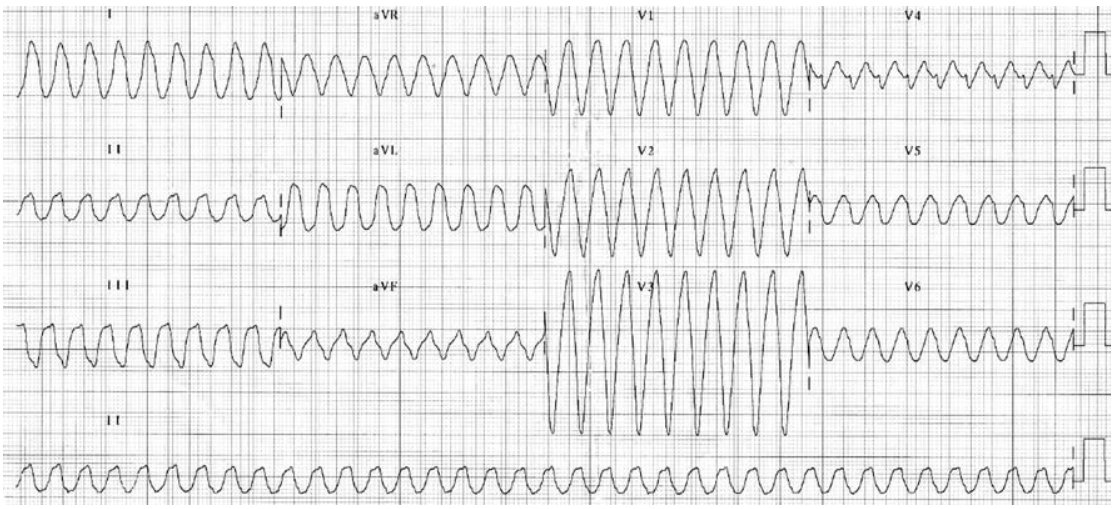

Figure 1. Admission electrocardiogram showing ventricular tachycardia during palpitation.

cardia (VT) (Fig. 1). Within minutes, hemodynamic collapse developed and electrical cardioversion with 200 joules was immediately performed. Immediately following cardioversion, ECG showed atrial fibrillation (AF), a slightly prolonged QT interval, and intraventricular conduction delay (Fig. 2). After intravenous infusion of amiodarone, the rhythm converted to sinus rhythm.

The patient had a 12-year history of myotonic muscular dystrophy for which she had been treated with mexiletine. Two months earlier, she was treated for atrial flutter with propafenon and amiodarone, but this rhythm persisted.

Physical examination showed ptosis, and cataract, and manual strength test of symmetric upper and lower extremities showed muscular weakness of grade 4/5. Electroneuromyographic assessment was compatible with MD. Transthoracic echocardiography showed significantly depressed left ventricular function and an ejection fraction of 25%. No signs of dyssynchrony were found. On cardiac catheterization, pulmonary capillary wedge pressure was 23 mmHg, left ventricu-

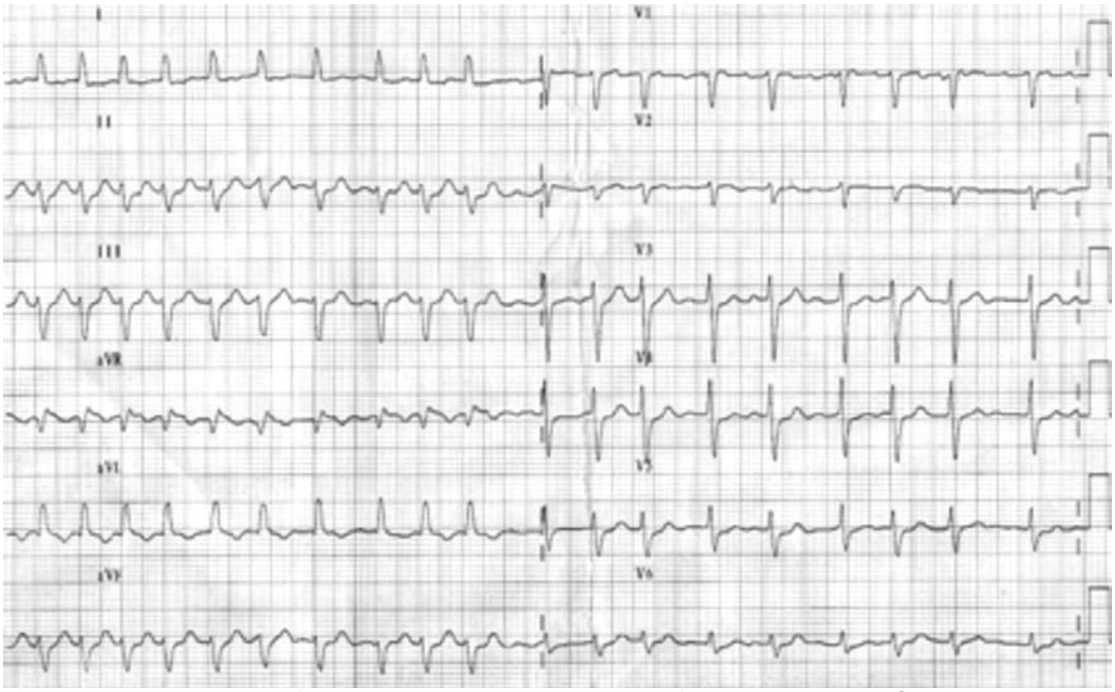

Figure 2. Electrocardiogram following cardioversion showing atrial fibrillation, prolonged QT interval, and intraventricular conduction delay.

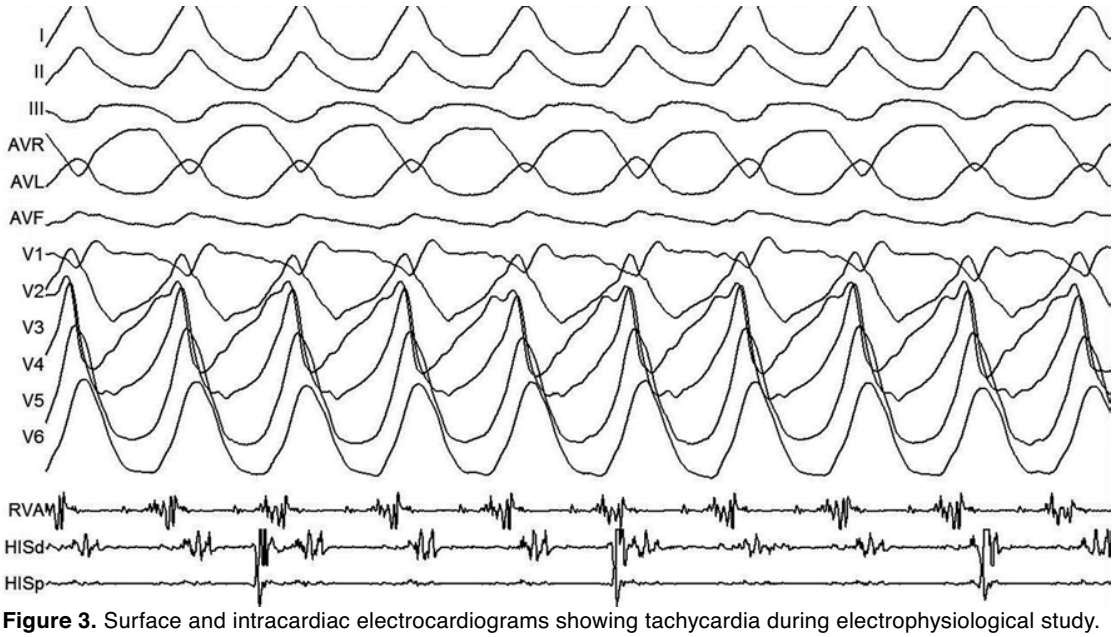

Figure 3. Surface and intracardiac electrocardiograms showing tachycardia during electrophysiological study.

lar end-diastolic pressure was 19 mmHg, and systolic, diastolic, and mean pulmonary artery pressures were 45, 20, and 30 mmHg, respectively. Cardiac index was 2.03 l/min/m^2 according to the Fick method. Coronary arteries were normal. Intracardiac electrocardiogram was obtained during electrophysiological study (EPS). On EPS, atrium-His interval was 120 msec and His-ventricle (H-V) interval was 54 msec, and monomorphic ventricular flutter was induced by a single programmed stimulus from the right ventricular apex (at a coupling interval of 260 msec) (Fig. 3). Ventricular flutter was terminated by overdrive pacing. An implantable cardioverter-defibrillator (ICD) (VVIR mode, Ovatio VR 6250, Ela Medical, Plymouth, MN, USA) was implanted. The patient was discharged in sinus rhythm and on medical treatment.

DISCUSSION

Conduction system abnormalities are commonly observed in MD1. Fibrosis and fatty infiltration are observed together in the conduction system and may be a possible underlying mechanism of the development of conduction system defects.[3,4] The most frequent involvement is in the His-Purkinje system, but any part of the conduction system may be affected.[2] A long PR interval and/or a wide QRS complex may accompany delayed impulse propagation along the conduction system. Late potentials which result from delayed myocardial activation usually associated with abnormal tissue predict ventricular arrhythmias.[2] A long PR interval (220 msec) and wide QRS (124 msec) were present in our patient.

In patients with MD1, a pacemaker should be implanted according to the recommended guidelines.[5] Asymptomatic atrioventricular conduction delay, especially in the presence of a prolonged H-V interval, represents one of the major therapeutic challenges in MD1, as data on the rate of progression to complete atrioventricular block are inconsistent. The presence of a prolonged H-V interval exceeding 70 msec may require prophylactic pacemaker implantation, even in the absence of symptoms.[6]

Tachyarrhythmias can occur in MD1 patients. Supraventricular tachyarrhythmias are common and may be asymptomatic.[2] The most common arrhythmias are atrial flutter and AF.[2] Atrial fibrillation was present in our patient.

Ventricular arrhythmias are also frequent in MD1. Monomorphic or polymorphic VT and ventricular fibrillation (VF) have been reported.[7] Monomorphic VT may be associated with re-entry around areas of fibro-fatty degeneration of the myocardium, bundle branch re-entry (typical), or triggered activity.[2]

During EPS, VF can be induced in the form of unsustained or sustained polymorphic VT, VF, or both sustained and unsustained monomorphic VT.[2] Sustained monomorphic ventricular flutter was induced in our case.

Treatment of ventricular arrhythmias in MD1 is difficult. Implantation of an ICD should be considered to treat VT because massive fatty fibrosis in cardiac muscle is often responsible for VT and numerous phar-

macological treatments have been found not to improve the condition.[8] There are also several reports of successful catheter ablation of VT in MD1 patients.[8,9]

Sudden death accounts for 2% to 30% of mortality in MD1 patients, possible mechanisms being ventricular asystole, degeneration of VT, VF, or electromechanical dissociation.[2] Groh et al.[10] investigated the predicting factors of sudden death in 406 adult patients with MD during a mean of 5.7 years follow-up period. They found that the presence of a severe abnormality on the ECG (PR interval ≥240 msec, QRS duration ≥120 msec, or second-degree or third-degree atrioventricular block) and a diagnosis of atrial tachyarrhythmia predicted sudden death, representing a 3.3-fold and 5.1-fold increased risk, respectively. Our patient had prolonged QRS duration (124 msec), AF, symptomatic VT, and inducible ventricular flutter. We implanted an ICD to prevent sudden cardiac death. We believe that implantation of an ICD is necessary in DM1 patients with symptomatic VT.

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