Musküler Distrofili
(Emery-Dreifuss Hastalığı) Bir Hastanın
Klinik İzlemi ve Aritmik Olayların Gelişimi

Mustafa KARACA M.D., Müge İldızlı DEMİRBAŞ M.D., Serdar BİÇEROĞLU M.D.,
Hasan YILMAZ M.D.
Atakalp Kalp Hastanesi, Kardiyoloji Bölümü, İzmir, Türkiye

ÖZET

Musküler distrofı (MD) kalıtımsal geçen bir hastalıktır. İsklelet kaslarının zayıflığına bağlı yorgunluk yaygın bir bulgudur ve kalp tutulumunu maskeleyebilir. MD hastalarında kalp tutulumunun prognostik önemi vardır ve hatta ilk klinik görünüm ani kardiyak ölüm olabilir. Bu yazida MD ve kalp tutulumu olan bir hastanın 14 yıl takibinde görülen aritmik olaylar sunulacaktır.

ANAHTAR KELİMELER
Muscular dystrophy, kalp tutulumu

Clinical Follow-up of a Patient with Muscular Dystrophy (Emery-Dreifuss disease) and the Progression of Arrhythmic Events

ABSTRACT

Muscular dystrophy (MD) is a genetically inherited disorder. Fatigue due to skeletal muscle weakness is a common symptom and can overshadow the cardiac involvement. Cardiac disease in the course of MD may have a prognostic significance and the first clinical presentation can be sudden death. In this report, overall arrhythmic events during a period of fourteen years were discussed in a patient with MD and cardiac involvement.

KEYWORDS
Muscular dystrophy, cardiac involvement

İLETİŞİM ADRESİ
Dr. Hasan YILMAZ
Atakalp Kalp Hastanesi, Kardiyoloji Bölümü, İzmir, Türkiye
**Introduction**

Muscular dystrophy (MD) is a genetically inherited disorder (1). Fatigue due to skeletal muscle weakness is a common symptom and can overshadow cardiac involvement. Cardiac disease in the course of MD may have a prognostic significance and the first clinical presentation can be sudden death (2). Cardiac involvement can lead to left ventricular systolic dysfunction, atrioventricular conduction defects supraventricular and ventricular arrhythmias (3,4). In this report, overall arrhythmic events during a period of 14 years in a patient with MD and cardiac involvement are discussed.

**Case**

The 37-years old male patient was referred to our cardiology clinic along with ventricular tachycardias (VT) causing hemodynamic instability. He had suffered from fatigue, dyspnea on exertion and discomfort in upper and lower extremities since his childhood. He had progressive muscle wasting of the lower extremities and difficulty in walking since his childhood. A diagnosis of muscular dystrophy was made in 1992 (at the age of 23) when he also suffered an unexplained syncope. He did not have any cardiac events for the next 8 years. He experienced sustained VT and syncope for which he was cardioverted several times and discharged on amiodarone 200 mg bid in the year 2000. He was stable for a year but developed recurrent sustained VT attacks in 2001 before he was referred to our clinic. From the family history his mother, grandmother and two male cousins had muscle diseases too. His mother died due to sudden cardiac death while she was 37 years old.

His cardiovascular examination was unremarkable. Neurologic examination showed marked muscular wasting and weakness of the shoulder girdle, proximal arm, proximal and distal leg muscles. Deep tendon reflexes of the lower legs were absent. No pathology was detected on pyramidal, extrapyramidal and cerebellar systems.

His ECG revealed sinus rhythm, first degree AV block (260 ms), left axis deviation (-30°), poor R wave progression in leads V1-3 (Figure 1). Echocardiography demonstrated global left ventricular dilatation (end diastolic dimension 59, end systolic dimension 40 mm) and mild left ventricular systolic dysfunction with an ejection fraction of 40%. Laboratory test results revealed that his serum CK level was 410 IU/L and LDH level was 850 U/L. Electromyogram (EMG) showed increased insertional activities, short duration polyphasic motor unit potentials, and an early recruitment or decreased interference pattern, suggesting a chronic myogenic disorder. Pathologic examination of the biopsy of the left gastrocnemius muscle revealed reduced myofibers with the replacement by diffuse fibroadipose tissue, marked variations in fiber size associated with atrophic or hypertrophic myofibers with marked splitting along with increased internal nuclei. These findings were consistent with Emery Dreifuss disease.

Following clinical evaluation, an electrophysiological study (EPS) was performed and revealed near normal intracardiac intervals (AH:151 ms, HV:50 ms, AV nodal Wenkebach time: 500 ms, 2:1 AV block time: 460 ms, SNRT max: 1120 ms, cSNRT: 270 ms). Sustained monomorphic VT with a cycle length of 250 ms and nonsustained polymorphic VT were induced by programmed ventricular stimulation with triple extrastimuli. Bundle branch reentry was excluded by demonstrating the absence of His deflection during VT. Based on the EPS, an endocardial cardioverter defibrillator (ICD) system, VENTAK PRIZM 2 VR1860 (Guidant Corp.,
Santa Clara, California) was implanted in July, 2001 to the left precordial region. The R wave amplitude, the pacing threshold and the pacing impedance at the time of implantation was 12 mV, 0.9 and 870 Ohms, respectively. The defibrillation threshold was 15 joules with a shock impedance of 42 ohms. After five days from ICD implantation, a fast VT (CL:280 ms) was detected and terminated successfully by ICD therapy.

During his follow-up, the degree of the AV nodal block progressively increased to third degree and the patient became pace dependant in 2005. Since then, he experienced several ventricular fibrillations which were successfully defibrillated into sinus rhythm (Figure 2). The patient has been currently doing well except from mild symptoms.

Discussion

Cardiac involvement in MDs is not rare and its signs and symptoms can be disrespected in overall symptomatology of the disease (1). Complete cardiac evaluation including echocardiography should be performed in patients with MD. Besides, patients with syncope should be evaluated thoroughly with noninvasive and invasive methods. Surface ECG serves as a good tool for this purpose. First degree AV block is a common finding as is the case in our pati-
ent. Less commonly atrial tachcardias and advanced AV blocks can be documented (2). In our patient first degree AV block progressed to complete AV block in four years of follow up after ICD implantation. Ventricular arrhythmias are reported in patients with MD but little is known about the exact incidence (5). While bradyarrhythmias can be managed with pacemaker implantation, ventricular tachyarrhythmias which might need further evaluation and require defibrillators (6). Therefore invasive electrophysiological studies should be carried out in patients with MD and ventricular arrhythmias. Bundle branch reentry tachycardia should be differentiated from other reentry VTs, since the first one can be ablated and cured during EPS (7,8). The patient presented in this report received appropriate shocks for VF five years after implantation of an ICD. First degree AV block that was present at the time of the diagnosis has evolved to complete AV block in four years. A pacemaker without ICD function would not prevent the patient from a cardiac arrest in his follow-up.

Cardiac involvement and its catastrophic results are the main reason of death in many patients with MD. Each patient with the diagnosis of MD should be evaluated by a cardiologist as for cardiac involvement and arrhythmic events. Patients in need of permanent pacemakers should also undergo invasive and non-invasive diagnostic techniques for the purpose of identifying ones at risk of fatal ventricular arrhythmias. In conclusion, the current status of ICD implantation for the prevention of sudden death in patients with MD and cardiac involvement should be reevaluated.

**FIGURE 2**

Ventricular fibrillation successfully defibrillated into sinus rhythm by an appropriate ICD shock.
REFERENCES


