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16 Years History of Presyncope: An Unexpected Presentation of Myotonic Dystrophy

16 Yıllık Presenkop Öyküsü: Myotonik Distrofinin Atipik Bir Prezentasyonu

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Öz

Miyotonik distrofi, erişkin çağın en sık görülen musküler distrofisidir. Hastalığın nörolojik bulguları tipik olmakla birlikte; kardiyak ve sistemik semptomların baskın olduğu olgularda tanı güçleşebilir. 16 yıldır süregelen presenkop atakları nedeniyle ilk olarak kardiyoloji bölümüne başvuran 46 yaşındaki erkek hasta, son dönemde belirginleşen yürüme güçlüğü şikayeti tanımlaması üzerine nöroloji bölümüne yönlendirilmişti. Detaylı nörolojik muayene ve elektrofizyolojik inceleme sonrasında hasta tip 1 miyotonik distrofi tanısı aldı. Mevcut olgu, sebebi bilinmeyen kardiyak ileti defekti, aritmi ve kardiyomiyopatiyle prezente olan hastalarda; özellikle de eşlik eden silik veya aşikar nörolojik şikayetler varlığında, nörolojik değerlendirmenin önemini ve gerekliliğini vurgulamaktadır. Bu tip olguların ayırıcı tanısında nöromuskuler hastalıklar da akılda tutulmalıdır ve doğru tanısal değerlendirme için multidisipliner yaklaşım esastır.

Anahtar Kelimeler: Miyotonik distrofi, kardiyak bulgu, presenkop

Abstract

Myotonic dystrophy is the most common muscular dystrophy of adulthood. Neurological manifestation of the disease is typical, however diagnosis could be challenge in patients presentig with predominant cardiac or systemic symptoms. 46 year old male was suffering from presyncope attacks for 16 years, and first examined by a cardiologist. Because of the recent complaints including walking difficulty, he was referred to neurology department. Following a detailed neurological examination and electrodiagnostic workup, he was finally diagnosed myotonic dystrophy type 1. The case highlights the necessitiy of neurological consultation in patients who present with conduction defects, arythmias or cardiomyopathies of unknown origin, accompanied by overt or subtle neurological symptoms. In such patients, neuromuscular disorders should be considered in the differential diagnosis; and in order to provide thorough diagnostic evaluation, multidisciplinary approach is essential.

Key words: Myotonic dystrophy; cardiac manifestation; presyncope

INTRODUCTION

Myotonic dystrophy is a multisystemic disorder characterized by muscle weakness and myotonia. It arises from abnormal expansion in CTG trinucleotide repeat and inherited in an autosomal dominant pattern. Cardiac, endocrine, ocular and gastrointestinal involvement usually occur during the disease course. The neurological manifestation of the disease is typical, however diagnosis can be delayed in patients presenting with cardiac or systemic findings. Here we present a case with 16 years history of presyncope episodes who was first evaluated by cardiology department and finally diagnosed myotonic dystrophy type 1 after neurological consultation.

CASE

46 year old male was suffering from presyncope attacks which were existing for 16 years. At the onset

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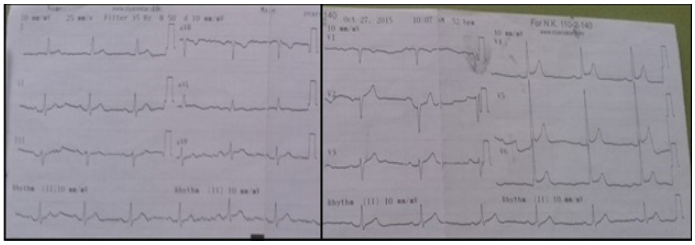

Figure 1. 12 lead electrocardiogram demonstrated sinus bradycardia with a heart rate of 52/min.

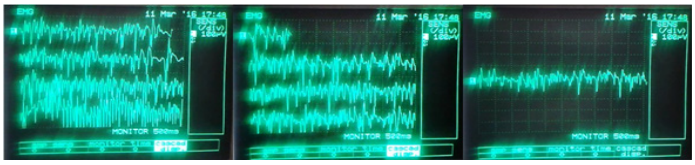

Figure 2. Needle EMG of the right anterior tibial muscle detected frequent myotonic discharges.

of episodes, he was evaluated by a cardiologist and recommended lifestyle changes. His past history was unremarkable except these presyncope attacks. The patient's attacks had worsened throughout the previous year and he was reevaluated by another cardiologist. Resting electrocardiogram (ECG) revealed sinus bradycardia with a heart rate of 52/min (Figure-1). Transthoracic echocardiography did not show any structural cardiac abnormality. 24 hours ECG recording was performed, which detected sinus bradycardia with a minimum heart rate of 36/min and 240 ventricular extrasystoles.

Since he complained about fatique and difficulty in walking recently, he was consulted to neurology department. The neurological examination revealed bilateral crural atrophy, weakness of ankle dorsiflexion and diminished tendon reflexes in the lower extremities. The family history was remarkable for consanguineous marriage. Hemogram, oral glucose tolerance test, vitamin B12 and thyroid stimulating hormone (TSH) levels were normal. Serum biochemistry showed mild elevation of aspartate aminotransferase (SGOT), alanine aminotransferase (SGPT) and lactate dehydrogenase (LDH) levels (59 u/l, 125 u/l, 227 u/l respectively; normal range: 5-34 u/L, 0-55 u/l, 125-220 u/L respectively). Creatinine kinase (CK) level was 135 u/L (normal range:30-200 u/l). Brain magnetic resonance imaging (MRI) showed ischemic gliotic changes on T2 and FLAIR sequences. After referral to our electrophysiology laboratuary with the early diagnosis of hereditery neuropathy, an electromyography (EMG) was performed. Nerve conduction study was normal, however needle EMG revealed myotonic discharges in both proximal and distal muscle groups of upper and lower extremities (Figure-2)

The neurological history was elaborated afterwards and, he was questioned whether he had experienced muscle stiffness at any time period. He admitted that he had difficulty in relaxing a clenched fist during adolescence. As he grew up, the symptom characteristics did change and distal muscle weakness became prominent. This time he had difficulty while using a key or opening a jar cap.

Bilateral temporal atrophy, facial weakness, high arched palate, diminished pharyngeal reflex, weakness of neck extension and both dorsal-plantar flexion of ankle were remarkable findings in the repeated neurological examination. Myotonic phenomenon was demonstrated.

Based on the typical clinical and electropysiological findings, he was diagnosed myotonic distrophy type 1.

DISCUSSION

Myotonic dystrophy type 1 was first identified by Hans Steinert in 1909. It is the most common muscular dystrophy of adulthood with an incidence of 1/8000 (1). According to the age of onset and severity of the symptoms; four grades of clinical categories were defined; congenital, childhood, adulthood (classical) and late onset (mild) type (2). Cardiac involvement is one of the most frequent systemic manifestatios of the disease and considered to arise from myocardial fibrosis. The clinical picture can range from asymptomatic ECG changes (such as PR, QRS and QT prolongation) to overt symptoms (such as presyncope and syncope, cardiac failure, sudden cardiac death) (3). Myocardial fibrosis results in degeneration of the conduction system which is assumed to serve as a basis for re-entrant arrhythmias and may also cause conduction abnormalities, as well as systolic ventricular dysfunction. (4-7).

Cardiac mortality in myotonic dystrophy type 1 is known to be as high as 30% and could occur as a result of progressive left ventricular dysfunction, ischemic heart disease, pulmonary embolism or sudden unexpected cardiac death (6). This type of patients, even the asymptomatic ones who demonstrate resting ECG abnormalities or prolonged HV intervals during

electrophysiological studies are known to benefit from permanent pace maker implantation (8).

CONCLUSION

In conclusion, the patients presenting with conduction defects, arythmias or cardiomyopathies of unknown origin, accompanied by overt or subtle neurological symptoms may need further neurological evaluation. In such patients, neuromuscular disorders should be considered in the differential diagnosis and in order to provide appropriate diagnostic workup, multidisciplinary approach is essential.

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