Cardiopathy in a patient with Emery-Dreifuss muscular dystrophy

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Emery-Dreifuss muscular dystrophy (EMD) was first described in 1961 by Dreifuss and Hogan. [1] It is a rare X-linked recessive disease characterized by variable slowly progressive muscle wasting and weakness, early contractures of the elbows, Achilles tendons and postcervical muscles. Cardiac manifestations are often severe, including dilated cardiomyopathy, complete heart block and sudden death. Until the report of Emery and Dreifuss in 1966 the cardiac manifestations were not recognized. [2] Later the gene was found to be located in the Xq28 region. Atrial arrhythmia that often progress to atrial standstill with bradycardia are the pathognomonic, unusual electrophysiological disorder in the disease. In symptomatic patients pacemaker implantation due to bradycardia is an accepted therapy although risk of sudden death in young adulthood remains very high. [3, 4]

**Case report**

A 25 year old man with the diagnosis of EMD was admitted to our hospital because of palpitations and dizziness. During the past year he had complained of gradually progressive exertional dyspnoea and fatigue. He denied a history of angina or syncope. He is a member of a known family with EMD living in lower Bavaria (Germany), described by Rotthauwe in 1972. [5] In this family, 9 of 17 affected males died suddenly at ages between 37 and 59 years, and several of them had advanced cardiomyopathy. His motor and mental development was not delayed. The disability of a mild muscular weakness in the upper arms, shoulders and lower legs, as well as contractures of elbows started in early childhood. There had been no progression of the motor disability since he was a teenager.

Physical examination on admission showed contractures at the elbows and muscle wasting in the shoulder girdle, biceps and leg (calf and dorsiflexors) muscles. The blood pressure was 125/75 mmHg and the heart rate was regular at 44 beats/minute (bpm). Jugular venous pressure was at the sternal angle without hepatojugular reflex. Auscultation revealed a 2/6 holosystolic murmur along the lower left sternal margin without any radiation. Lung auscultation was normal. In the laboratory tests, serum creatine phosphokinase level was elevated to 1493 units per litre (< 270 units per litre in normal subjects). A twelve-lead ECG showed no evidence of a p-wave in any lead. There was a junctional rhythm with a ventricular rate of 40 bpm. There were no T-wave changes (Figure 1). Holter monitoring showed a minimal heart rate of 35 bpm, but neither sinus pauses longer than 2000 ms nor ventricular arrhythmia were registered. Maximal heart rate was 91 bpm. An event recording study during 7 days showed several asymptomatic cardiac arrests of up to 3700 msec duration and one episode of bradycardia at a rate of 28 bpm. In a bicycle exercise test the patient achieved 210 Watt in steady...
Discussion

Emery-Dreifuss muscular dystrophy (EMD) is a X-chromosomal recessive disease with rather mild involvement of skeletal muscles and progressive cardiopathy, which limits life expectation to medium adulthood. Cardiac findings in the disease are unique. The cardiopathy is characterized by a marked loss of myocardium and its replacement by adipose and fibrous tissue starting in both atria and the conducting system and progressing to both ventricles. This process results in a dilatation of right and later left atrium, proceeding to dilatation of ventricles.

The rhythm disorders begin with a decrease and finally the loss of the p-wave in the ECG in childhood and progressive atrio-ventricular block with slow junctional rhythm. [6]

Electrical and mechanical atrial standstill with very thin and pale atrial walls is the pathognomonic finding. The increased risk of systemic embolism is obvious and comparable to the situation in atrial fibrillation. Several cases with either cerebral or peripheral embolic events have been described. [7] The progression to the ventricles leads finally to conges-tive heart failure and its complications. Death due to terminal heart failure is common but sudden death appears to be more frequent. Whether this occurs due to asystoly or ventricular tachyarrhythmia is unknown. Despite anti-bradycardia therapy with pacemakers several affected patients have died suddenly. [8]

Our patient is in a progressive phase of the pathognomonic heart disease with dilatation of all four chambers and atrial standstill, left ventricular dysfunction, secondary mitral and tricuspid regurgitation due to dilatation of the annulus. He has a junctional rhythm with 40 beats per minute (bpm) and had several asymptomatic pauses up to 3700 msec and bradyarrhythmias down to 28 bpm. Worldwide about twenty families exist with EMD, and some single cases have also been described. [7, 8] Our patient is member of a known family with 17 males affected with EMD in 3 generations. Nine of them died suddenly and five other have had partial or complete atrioventricular block.

Permanent ventricular pacing improves symptoms and prolongs the life span in patients with sinus bradycardia, sinus pause or heart block. [3, 8] Because of the complete loss of myocardium in the atria, atrial pacing is not of aid. Therefore, in view of the chronotropic incompetence and its expected progression, our patient received a ventricular pacing system with a rate response option. [9] Little is known about the aetiology of fatal accidents in these patients. About 40 % of EMD patients died suddenly, many of them without preceding cardiac symptoms. [10] Whether the risk of sudden death bases on asystoly or on tachyarrhythmias seems to be unknown. [5]

This fact and the excessively high sudden death rate in our patient’s family gave us the indication for implantation of a prophylactic cardioverter and defibrillator in combination with the VVIR-pacemaker. To reduce the cardiac remodeling, ACE inhibitor therapy was started, in view of its therapeutic effects in dilated cardiomyopathy of any aetiology. The importance of prophylactic long-term oral anticoagulation for the prevention of systemic embolism originating from the dilated and still standing left atrium is obvious.

During two years of follow-up the patient has remained stable and the patient oligosymptomatic without any syncpe or shock.

References

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