Complete atrioventricular block in Duchenne muscular dystrophy

A. Fayssoil1*, D. Orlikowski2, O. Nardi2, and D. Annane2

1Cardiology, European Hospital Georges Pompidou, AP-HP, Paris, France; and 2Critical Care Unit, Hospital Raymond Poincare (AP-HP), University of Versailles SQY, 104 Boulevard Raymond Poincare, 92380 Garches, France

*Corresponding author. Tel: +33 66 785 5792. E-mail address: fayssoil2000@yahoo.fr

Duchenne muscular dystrophy (DMD) is an inherited myogenic disorder due to mutations in the dystrophin gene on chromosome Xp21.1. It is characterized by progressive muscle wasting and weakness of variable distribution and severity. Heart is involved leading to heart failure. Conduction abnormalities are unusual. We report a case of complete atrio-ventricular block in a DMD patient.

Case report

A 33-year-old man with Duchenne muscular dystrophy (DMD), on wheelchair, was admitted in hospital for dyspnea, bradycardia, and asthenia. Diagnosis was made in the childhood by genetic analysis, which found deletion exon 50–52 (dystrophin gene). His past medical history was pertinent for scoliosis, tracheotomy, and home ventilation for respiratory insufficiency. He had no cardiovascular risks factors. He had taken perindopril 2 mg and furosemide 20 mg daily for cardiomyopathy known for 13 years. On admission, body surface area was 1.5 cm², body temperature was 36.8°C, blood pressure was 126/59 mmHg, and heart rate was 40 bpm. Oxygen saturation was 100% with mechanical ventilation. Electrocardiogram (ECG) showed complete atrioventricular (AV) block (Figure 1). Echocardiography showed a global left ventricular dysfunction with estimated ejection fraction of 45%. Blood count and electrolytes were normal. A temporary transvenous pacing lead was inserted because of complete AV block and 24 h later, replaced by a dual-chamber permanent pacemaker. Patient was discharged home 5 days later without any complications.

Discussion

We report a case of complete symptomatic conduction AV block in a patient with DMD. DMD is an inherited myogenic disorder due to mutations in the dystrophin gene on chromosome Xp21.1. This is an X-linked disease: therefore, it affects one in 3500 live-born males.1 It is characterized by progressive muscle wasting and weakness of variable distribution and severity leading to immobility in the early teens. Clinical manifestations begin at the age of 3–4 years and affect particularly pelvic and shoulder girdles muscles. Pseudo-hypertrophy of the calves, lumbar lordosis, and kyphoscoliosis are commonly found. Patients are usually confined to wheelchair by the age of 12. Diagnosis is made by analysing dystrophin gene in DNA of peripheral lymphocytes and by performing muscular biopsy for immunostaining and western blot.1 Heart is involved leading to heart failure.1 Initially, cardiac involvement is segmentary with dystrophy involving the posterobasal segment of the left ventricular wall and progressively the whole heart. Traditionally, ECG discloses wide R waves in the right precordial leads, increase in the R/S ratio and Q waves in aVL, V5, and V6 because of the posterior wall involvement. Conduction abnormalities have been reported in the literature in DMD patients. The first case was reported by Benjaid in 1974.2 Pathophysiology remains complex. Dystrophin is a sarcolemmal protein that binds actin to extracellular matrix. Normally, dystrophin is expressed in heart and skeletal muscles. It is absent in DMD patients. Deficiency in dystrophin would result in breakdown of muscle membrane. This process would lead to loss of muscle.
enzyme and weak muscles. Cardiac involvement is due to progressive replacement of the cardiomyocytes and the Purkinje system by connective tissue or fat.\textsuperscript{1} Dystrophin is localized to the membrane surface of Purkinje fibres.\textsuperscript{3} AV block may be explained by the absence of dystrophin in electrical tissue that alters conduction. Cardiac autonomic nervous system disturbances are reported in DMD patients particularly reduced vagal activity and enhanced tone.\textsuperscript{4} Other abnormalities include a short PQ interval and a long QT interval. In a controlled series of 328 DMD patients, 62\% had conduction abnormalities such as a short PQ interval and a long QT interval by age 10 without clinical cardiac involvement.\textsuperscript{5}

Prophylactic pacemaker implantation is very problematic in view of the bad prognosis of the disease and the potential for serious deterioration in case complications occurred. Even a small pneumothorax may lead to frank respiratory insufficiency in these compromised patients because of their reduced ventilation reserve.

References