Asymptomatic, Nonsustained Ventricular Tachycardia in Myotonic Dystrophy Type 1 Detected with a Loop Recorder

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Cardiac involvement is a dominant feature of myotonic dystrophy type 1 (MD1) [1]. Cardiac involvement in MD1 manifests as impulse generation or impulse conduction disturbances or cardiomyopathy [1]. Since cardiac involvement predicts the outcome of these patients [2, 3], it is important to detect cardiac involvement as soon as possible after establishing the neurological diagnosis to initiate adequate cardiac therapy. Though ventricular tachyarrhythmias (VTs) are a frequent finding in MD1 patients [3–6], they are usually detected only after patients have become symptomatic. Asymptomatic, nonsustained VT is usually detected only by 24-hour ambulatory ECG or loop recording as in the following case.

Case Report

The patient is a 42-year-old, HIV-negative Caucasian male, in whom MD1 was diagnosed at age 31 years, based upon family history, clinical findings and molecular genetic investigations. Since age 6 years he developed slowly progressive dystarthish and since age 14 a foot drop bilaterally. Muscle enzymes were found to be elevated for the first time at age 31. Clinical neurologic investigation at age 42 years revealed severe dystarthish, bilateral ptosis, ophthalmoparesis, myopathic face, diffuse weakness of the upper limbs with distal predominance (M3+ to M4+), weakness for hip flexion bilaterally (M5–), and for foot extension and flexion bilaterally (M2 and M4–), hyperextensible joints, absent tendon reflexes, hypotonia, ataxia, and a positive Gower’s sign. Creatine kinase was 60 U/l (normal: <171 U/l). Hb A₁c was 6.4% (normal: 3.5–6.0%). Triglycerides and cholesterol were elevated. Ophthalmologic investigations revealed mild cataract and a CT scan of the brain showed moderate widening of the ventricles and a considerable falx menigioma. Nerve conduction studies disclosed polynuropathy. Needle electromyography showed typical abnormal spontaneous activity and a myogenic motor unit architecture. Analysis for mutations in the DMPK gene on chromosome 19q13.3 revealed a CTG-repeat expansion of 1,333 repeats in nuclear DNA from blood lymphocytes. The mutation was also found in his brother (1,333 repeats). His mother was clinically affected for hip flexion bilaterally (M5–), hyperextensible joints, absent tendon reflexes, hypotonia, ataxia, and a positive Gower’s sign. Creatine kinase was 60 U/l (normal: <171 U/l). Hb A₁c was 6.4% (normal: 3.5–6.0%). Triglycerides and cholesterol were elevated. Ophthalmologic investigations revealed mild cataract and a CT scan of the brain showed moderate widening of the ventricles and a considerable falx menigioma. Nerve conduction studies disclosed polynuropathy. Needle electromyography showed typical abnormal spontaneous activity and a myogenic motor unit architecture. Analysis for mutations in the DMPK gene on chromosome 19q13.3 revealed a CTG-repeat expansion of 1,333 repeats in nuclear DNA from blood lymphocytes. The mutation was also found in his brother (1,333 repeats). His mother was clinically affected as well, had developed left bundle-branch block, and had died at age 55 years from sudden cardiac death.

His medical history was negative for arterial hypertension, diabetes, angina pectoris, coronary heart disease, dyspnea, leg edema, syncope, or palpitations. The patient was first investigated for cardiac involvement at age 31 years, revealing normal P-Q interval (0.18 ms) and ST elevation on ECG and slight tricuspid insufficiency on echocardiography. At age 36 years cardiologic investigation revealed tricuspid insufficiency, an E/A ratio <1, and episodes of bradycardia (HR <50/min) on 24-hour ambulatory ECG. At age 41 years, physical examination was normal. Blood pressure was 120/70 mm Hg. The ECG showed sinus rhythm, an AV block I (P–Q interval 0.21 ms) and absent R progression between V1 and V4. Echocardiography was normal. Since 24-hour ambulatory ECG revealed a P–Q interval of 200 ms and 204 episodes of bradycardia and since his mother had died from sudden cardiac death, it was decided to implant a loop recorder (Reveal Plus, Model 9526, Medtronic Inc.). During the first 1.5 years no abnormal electrical activity could be recorded although interrogation was carried out every 3 weeks. After almost 2 years, however, interrogation of the loop recorder revealed a VT of 1-min duration following an initializing R-on-T phenomenon (fig. 1) during sleep without obvious clinical manifestations. Following the detection of the VT the patient received an implantable cardioverter defibrillator (ICD) at age 42. Shortly before implantation of the ICD, recurrent VTs were recorded also during continuous ECG monitoring in the hospital where the implantation of the ICD was scheduled. In addition to the ICD he was regularly taking candesartan. During a follow-up of 3 months, 2 discharges were registered because of VTs.
Discussion

Cardiac involvement in MD1 includes impairment of impulse generation, including sinus bradycardia, atrial and ventricular ectopic beats, atrial fibrillation, atrial flutter, VT, torsade de pointes, or ventricular fibrillation, or impairment of the cardiac conduction system, including atrioventricular block, left or right anterior hemiblock, Q-T prolongation, or bundle-branch block [6–9]. The prevalence of inducible VTs in MD1 patients is 18% [10]. Myocardial involvement in MD1 is rare and includes myocardial fibrosis [11], fatty infiltration [11], myocardial thickening [11], or left ventricular hypertrabeculation, also known as noncompaction [12], resulting in left ventricular systolic or diastolic dysfunction [13]. The most commonly reported VT is bundle-branch reentry [5, 14] followed by fascicular VT [15, 16]. The pathoanatomic substrate of VT is fatty infiltration, interstitial fibrosis, or atrophy and disarrangement of cardiomyocytes [6, 11]. The alterations result in HisPurkinje conduction delay [5, 17]. VT can be most easily detected on 24-hour ambulatory ECG or loop recording. The latter has only rarely been carried out in patients with MD1 so far [18]. Though some authors recommend the implantation of ICDs for documented VTs in MD1 patients [6], ICDs have only occasionally been implanted for the prevention of sudden cardiac death in patients with MD1 [19, 20]. In addition to the implantation of an ICD, ablation of the right bundle has been recommended [13]. Before the availability of ICDs the prevalence of sudden cardiac death was up to 33% in MD1 patients [8, 11, 21]. Despite ablation, however, some patients with MD1 experience sudden cardiac death.

The presented patient is remarkable for dysarthria as the initial manifestation of MD1, for only mild cognitive impairment despite a CTG repeat number of 1333, for the late development of cardiac involvement despite considerable repeat expansion, and for the development of asymptomatic, nonsustained VTs, which lastly prompted the implantation of an ICD. Since sudden cardiac death is a prominent feature of cardiac involvement in MD1 [8, 11, 21], the patient might profit from the implantation of an ICD, particularly in the light of his mother’s sudden cardiac death and the recurrence of VTs. However, few data are available about the long-term
benefit of ICDs implanted to prevent sudden cardiac death in patients with MD1. Whether sudden cardiac death could have been prevented in his mother if she had received an ICD and whether the presented patient will profit from the ICD also on the long run remains questionable. Administration of amiodarone, which was planned by the treating physicians in addition to the ICD, was not recommended to the patient in addition to the ICD, was not recommended to the patient, since previous reports showed that amiodarone can have an arrhythmogenic effect in MD1 patients, leading even to cardiac arrest [22].

This case shows that loop recording may be helpful for the detection of asymptomatic, nonsustained VTs in MD1. Implantation of a loop recorder and close interroga
tion is strongly recommended in MD1 patients with documented bradycardia, syncope, or a family history positive for sudden cardiac death. Implantation of an ICD may prevent sudden cardiac death in MD1 patients.

References

therapy of the arrhythmias in 3 cases of myotonic dystrophy (Steinert’s disease). Cardiologia 1996; 41:1209–1214.