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QT and QU Interval Prolongation, Bidirectional Ventricular Tachycardia and Aborted Sudden Death. An Andersen-Tawil Syndrome

Konstantinos P. Letsas, MD, Michalis Efremidis, MD, Antonios Sideris, MD

*Second Department of Cardiology,
Evangelismos General Hospital of
Athens, Athens, Greece*

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ABSTRACT

A 42-year-old lady survived an episode of near-drowning and she was subsequently diagnosed with a rare genetic disease, recently classified as long QT 7 syndrome, for which she received an implantable cardioverter defibrillator. The features of this syndrome are herein described.

A rare constellation of symptoms, somatomorphic features, ECG findings and curious ventricular ectopic activity were fumbled upon a patient who survived a near-drowning episode.

CASE REPORT

A 42-year-old woman, who had experienced an episode of near-drowning during recreational swimming and had been successfully resuscitated, was referred to our department for evaluation. Physical examination revealed a short stature and micrognathia. Routine laboratory examinations including blood and urine tests, chest x-ray film and transthoracic echocardiography were normal. A standard 12-lead ECG on admission demonstrated sinus rhythm, a prolonged corrected QT interval (QTC) of 480 msec, a prolonged terminal T wave downslope and a prominent U wave with a wide T-U junction and a corrected QU interval (QUC) of 700 msec (Figure 1). Twenty-four-hour Holter monitoring showed frequent premature contractions in bigeminy and episodes of nonsustained ventricular tachycardia.

During electrophysiological study, an episode of symptomatic bidirectional ventricular tachycardia with a cycle length of 330 msec was recorded (Figure 2). A provocation test with face immersion in cold water induced complex ventricular ectopy in the form of an episode of nonsustained ventricular tachycardia as well as ventricular bigeminy. On the basis of this finding, one could indirectly assume that water and consequently swimming might represent a likely trigger for ventricular tachyarrhythmias in this patient. Causes of acquired long QT syndrome, such as drugs, electrolyte abnormalities, etc. were effectively ruled out. The aforementioned dysmorphic features (short stature, micrognathia) and ECG findings (T-U wave patterns, ventricular arrhythmias) suggested a high clinical probability of Andersen-Tawil syndrome, which

Address for correspondence:
Konstantinos P. Letsas, MD
25, 28th October Street
Athens 15 235, Greece
Tel: +302106843854,
Fax: +302107217687
e-mail: k.letsas@mail.gr



FIGURE 1. Precordial ECG leads showing prolonged QTC (480 msec) and QUC (700 msec) intervals, and characteristic T-U wave patterns.

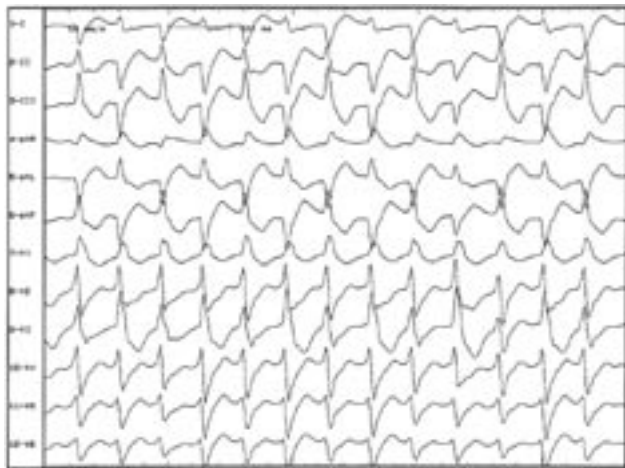


FIGURE 2. Bidirectional ventricular tachycardia recorded during electrophysiological study.

has been also proposed as LQT7. The patient was treated with an implantable cardioverter-defibrillator.

DISCUSSION

Andersen-Tawil syndrome (ATS) is a rare autosomal dominant disorder characterized by periodic paralysis, ventricular dysrhythmias and skeletal developmental abnormalities such as cleft palate, low set ears, micrognathia/retrognathia, hypertelorism, broad base of nose, short stature, clinodactyly/syndactyly/brachydactyly [1-4]. One gene, *KCNJ2*, has been identified so far and >20 mutations have been reported [5-

7]. This genotype has been labeled ATS1. *KCNJ2* encodes Kir2.1 protein, which is the critical α subunit of cardiac IK1, the inward rectifier potassium current. IK1 is responsible for maintaining resting membrane potential in atrial, His-Purkinje and ventricular cells. Reduced IK1 resulting from *KCNJ2* mutations alters late cardiac repolarization and leads to both distinctive T-U wave morphology and an increased propensity to ventricular arrhythmias.

A recent study [8] demonstrated genotype-specific T-U-wave patterns that included prolonged terminal T-wave downslope, a wide T-U junction, and biphasic and enlarged U waves. These T-U wave patterns were not seen in the ATS non-*KCNJ2* patients or normal subjects and could therefore be used to distinguish carriers of *KCNJ2* mutations from normal subjects, ATS patients without *KCNJ2* mutations and LQTS1 through LQTS3 with excellent sensitivity and specificity. Interestingly, our patient demonstrated almost all these characteristic T-U wave patterns.

Several features make the case herein described interesting. Firstly, QTC interval was well beyond the median QTC value of 440 msec in ATS1 patients (only 17% of ATS1 patients have a QTC >460 msec) [8]. Furthermore, despite the fact that ATS1 patients generally seek medical attention because of periodic paralysis rather than because of ventricular arrhythmia [8], our patient demonstrated complex ventricular ectopy, consisting of frequent PVCs mainly in bigeminy and bidirectional ventricular tachycardia. Moreover, syncope and cardiac arrest are infrequently seen in patients with ATS1 [8]. Finally, to our knowledge, this is the first report of a swimming-triggered cardiac event in a patient with Andersen-Tawil syndrome [9].

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