Short QT Syndrome: A Fatal Faint

SNIGDHA KOLA, MD, INDRAJEET MAHATA, MD and ABRAHAM G. KOCHERIL, MD, FACC, FACP, FHRS

Department of Cardiology, University of Illinois College of Medicine, Urbana Champaign, IL

KEYWORDS. short QT syndrome, electrocardiogram, ventricular fibrillation

Introduction

Short QT syndrome (SQTS) is a rare channelopathy that increases the risk of tachyarrhythmias and sudden cardiac death. It was first described in 2000. The diagnosis is difficult as it is usually unmasked before or after an episode of ventricular tachyarrhythmia. Timely diagnosis and preventive measures along with genetic studies play a pivotal role in the course of patients.

Case report

A 20-year-old white female with a history of attention deficit disorder presented to the Emergency Room with an episode of loss of consciousness 1 month after she had delivered her first baby. Her family history was significant for unexplained sudden cardiac death in her mother at age 23. Physical examination was unremarkable. The metabolic panel was within normal limits, but the electrocardiogram (ECG) showed sinus bradycardia. She was extensively evaluated for neurogenic and cardiogenic causes of syncope with magnetic resonance imaging of the brain, echocardiogram, and the tilt table test, all of which were within normal limits.

An electroencephalogram showed an epileptogenic focus in the left temporal lobe with sharp discharges and she was started on antiepileptic medication. However, she continued to have episodes of syncope, and an insertable loop recorder was placed. Interrogation of this showed over 500 episodes of bradycardia correlating with some episodes of presyncope. A permanent pacemaker implantation was planned electively.

In the interim period, she had another episode of loss of consciousness with cyanosis and seizure activity requiring cardiopulmonary resuscitation and defibrillation. Her ventricular fibrillation was terminated by an automated external defibrillator shock, and the subsequent ECG tracing immediately after the termination of the arrhythmia was notable for a short QT interval of 280 ms. An ECG taken 8 h after onset of symptoms showed normal sinus rhythm.

Subsequently, she underwent insertable defibrillator placement. Genetic testing is planned.

Discussion

The genetic basis of SQTS has been found to be a gain of function mutation in the K (potassium) channels and loss of function mutation in the Ca (calcium) channels underlying the cardiac action potential. Based on these,
Figure 2: The patient sustained ventricular fibrillation (a); this was terminated by automated external defibrillator shock (b), followed by sinus rhythm with shortening of QT interval with a duration of 280 ms (c).

Figure 3: Following the first shock, the patient went into another episode of ventricular fibrillation (a); this was followed by a second shock (a) and the tracings immediately following this show shortening of the QT interval (b,c).
five different types of mutations have been identified and labeled SQT 1–5 (Table 1).3,4

Gain of function in K channels increases the outward K currents across the membrane,7 thereby shortening the duration of phases 1–3 of cardiac action potential. Loss of function of Ca channels decreases the inward currents across the membrane that are responsible for the plateau phase in the action potential (phase 2), thereby shortening it. These manifest as shortening of the QT interval in the surface ECG.

Conclusion

SQTS has been found to have an autosomal dominant pattern of inheritance.5 A rate-corrected QT interval of <320 ms6 is one of the proposed criteria, since a definitive consensus has not been established yet. Affected individuals are often noted to have a strong family history. The diagnosis of SQTS is difficult in view of it being masked when a patient is asymptomatic, and because clear parameters have not yet been determined. This channelopathy should be considered a possibility when an unexplained syncope is associated with a family history of sudden death.7–9 Implantation of a defibrillator is considered the mainstay of therapy; pharmacologic therapy is used when a defibrillator cannot be placed.

References


Table 1: SQT 1–5.

<table>
<thead>
<tr>
<th>Subtype</th>
<th>Mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td>SQT1</td>
<td>Gain of function of I(_{Kr})</td>
</tr>
<tr>
<td>SQT2</td>
<td>Gain of function of I(_{Ks})</td>
</tr>
<tr>
<td>SQT3</td>
<td>Gain of function of I(_{K1})</td>
</tr>
<tr>
<td>SQT4</td>
<td>Loss of function of I(_{Ca})</td>
</tr>
<tr>
<td>SQT5</td>
<td>Loss of function of I(_{Ca})</td>
</tr>
</tbody>
</table>