Brugada syndrome: Unmasking a silent killer

Learn to recognize this genetic disorder that can cause sudden death in apparently healthy young people.

By Leslie Foran Lee, RN, and Nancy Felmlee, BSN, RN

DEALING WITH THE DEATH of an apparently healthy young person is one of the challenges we face in healthcare. A person may just not wake up one morning, or die suddenly while resting on the sidelines after the big game. This mystery of sudden death has baffled healthcare providers for centuries. One of the links to this mystery is Brugada syndrome, or unexplained sudden cardiac death syndrome, a genetic disorder that causes syncope or death in young people with no apparent cardiac history. Since the syndrome was described by Pedro and Josep Brugada in 1992, Brugada syndrome has attracted great interest because of its high incidence in many parts of the world and its association with a high risk of sudden death, especially in men ages 30 to 50.1

The syndrome is characterized by ST-segment elevation in the right precordial leads (V1, V2, and V3) and a high incidence of sudden death. Many different clinical situations or drugs can exacerbate or unmask a Brugada-like ECG pattern.2

Risks and mortality

Although the genetic mutation responsible for Brugada syndrome occurs equally in men and women, the prevalence of the syndrome is 8 to 10 times higher in men.1

Recent studies have shown that, for unknown reasons, Brugada syndrome is far more common in Asian men. They are healthy individuals with no apparent cardiac history. After history and physical exam of these men, it was found that there was a family history of either syncope or sudden death. The syncopal episodes or sudden deaths followed rigorous exercise, or occurred during sleep, or were exacerbated by a fever. In the last several years, risk stratification has become the preferred way to accurately identify and treat these patients. The risks are the presence of symptoms before diagnosis, a spontaneous type 1 ECG at baseline, inducible ventricular dysrhythmias in an electrophysiology (EP) lab, and being male.3 (See A closer look at Brugada syndrome for details on ECG patterns found with the syndrome.)

Pathophysiology

Brugada syndrome is an ion channel disorder, which results in the abnormal electrical activity in the epicardial cells of the right ventricle. How this actually occurs is under investigation, but a familial occurrence is noted to be present in about half the patients with Brugada syndrome, suggesting a genetic component to the disease. Also suggestive of a genetically determined disease is the clustering of the first onset of symptoms at ages 30 to 50. Recent studies show a confirmed genetic association in 10% to 30% of patients, with mutations in the SCN5A gene that encodes the cardiac sodium channel. The result is a loss of proper function of the sodium channel and a predisposition to ventricular fibrillation (VF).4

Because the sodium channel plays a key role in conducting cardiac impulses, reviewing it can help you better understand the pathophysiology of Brugada syndrome. The sodium channel has two main functions: moving sodium ions across the pores in the cell membrane and gating (opening or closing in response to changes in membrane potential).5 The cardiac sodium channel opens and closes rapidly at the onset of the cardiac action potential. In Brugada syndrome, the channel is perpetually closed.6

Genetic defects in the membranes’ ion channels can disrupt the delicate balance of dynamic interactions.
between the ion channels and the cellular environment, leading to altered cellular function. A single mutation in the sodium channel is enough to cause Brugada syndrome and contribute to the genesis of cardiac dysrhythmias.5

**Recognizing the problem**

About 8% of asymptomatic patients with Brugada waves on ECG have subsequent cardiac events.3 Although this number will undoubtedly grow as research continues, many clinicians feel it’s not possible to justify genetic studies, endocardial biopsies, magnetic resonance imaging, EP studies, and coronary arteriography in every patient in whom this syndrome is suspected.7

Brugada syndrome can be diagnosed based on the patient’s ECG and family history. (Other tests may be done to rule out other cardiac conditions.) Three types of ECG patterns in the right precordial leads are recognized, and often all three are present in the patient. However, Brugada syndrome can only be definitively diagnosed if the patient has the type 1 ECG pattern in more than one right precordial lead (V1–V3), in the presence or absence of a sodium channel blocking drug, such as flecainide or procainamide, and one of the following:

- documented VF
- documented polymorphic ventricular tachycardia (VT)
- family history of sudden death at an age younger than 45 years
- the presence of coved-type ECG in family members
- inducible ventricular dysrhythmias with programmed electrical stimulation
- syncope
- nocturnal agonal respirations.3

The right precordial ST-segment elevation associated with this syndrome can be mistaken for other cardiac problems, including acute pericarditis, acute myocardial ischemia or infarction, and Prinzmetal angina. (See *What’s up with the ST segment?* for details on differentiating Brugada syndrome from other conditions.)

If you’re caring for a patient who had a syncopal episode but has no history of cardiac disease, obtain an extensive history and perform a careful physical assessment. If you suspect Brugada syndrome, place the right precordial leads slightly higher on the patient’s chest to capture a complete right bundle-branch block or type 1 ECG pattern. Place leads V1 and V2 at the third intercostal space to the right and the left of the sternal border, instead of at the fourth intercostal space.6

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**A closer look at Brugada syndrome**

The waveform shows the coved-type ST-segment elevation in leads V1 and V2 that are characteristic of type 1 ECG patterns you may see in a patient with Brugada syndrome. Diagnostic criteria for Brugada syndrome, based on ST-segment abnormalities in leads V1 through V3, are outlined in the table.

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Assess and support the patient’s airway, breathing, and circulation. Make sure he has a patent airway and have resuscitation equipment readily available. Place the patient on a cardiac monitor, establish vascular access, and administer supplemental oxygen as indicated. Document and inform the healthcare provider of any cardiac rate or rhythm abnormalities.

Because the ECG pattern in Brugada syndrome patients can be dynamic, you may not see the characteristic hallmark signs. To unmask the ECG pattern and aid in diagnosis, patients may be given an infusion of sodium channel blockers to increase the sodium channel dysfunction. Commonly used drugs include flecainide and procainamide. The patient undergoing the pharmacological challenge test should have continuous ECG monitoring, and emergency equipment and supplies need to be immediately available. The study should be terminated when the test is positive, if the patient develops premature ventricular beats or other dysrhythmias, or the patient’s QRS complex widens to 130% or more of baseline. Monitoring the patient after testing is an important nursing implication. Watch for changes in vital signs, cardiac dysrhythmias, near-syncpe, or syncope, and intervene as appropriate.

The patient may need a cardiac EP study, depending on his clinical status. An EP study may not be needed for a patient who has classic Brugada waves after VT has reverted to normal. (This patient will need an implantable cardioverter-defibrillator [ICD]).

An asymptomatic patient with Brugada waves in a routine ECG may need an EP study; if VT can be induced, the patient should receive an ICD.

**Treating Brugada syndrome**

A patient whose Brugada syndrome isn’t treated has a very poor prognosis. One-third of patients who suffered syncopal episodes or were successfully resuscitated after sudden cardiac death develop a new episode of polymorphic VT within 2 years.

No drugs have reduced the VT or VF in patients with Brugada syndrome. Because the dysrhythmias are ventricular in nature, the primary treatment is ICD implantation, although the treatment is controversial because an ICD carries the risk of inappropriate shocks. Educate the patient and family members about Brugada syndrome and cardiopulmonary resuscitation. Genetic counseling also is recommended.

**On the alert**

By understanding Brugada syndrome, taking a careful patient history, and being able to differentiate ECG abnormalities, you may be able to recognize this syndrome in a patient and help him get appropriate care.

**References**