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## CLINICAL VIGNETTE

# Brugada Syndrome: Case Report with an Approach to Diagnosis

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### Case Report

Brugada syndrome is a genetic disorder characterized by specific abnormal electrocardiographic (ECG) patterns with an increased risk of life-threatening arrhythmias.<sup>1</sup> We report a 43-year-old male with history of hypertension, hyperlipidemia, and abnormal ECG referred to Cardiology for syncope evaluation. He had syncope at rest and regained consciousness and was back to his normal state after a few seconds. His ECG showed

NSR with ST/T abnormalities concerning for Brugada syndrome (Figure 1). His parents were from Thailand and there was no family history of sudden cardiac death. Echocardiogram and heart monitoring were unremarkable. He was referred to Electrophysiology and underwent a procainamide challenge test which was abnormal and he underwent successful placement of cardiac defibrillator. Genetic testing was deferred.

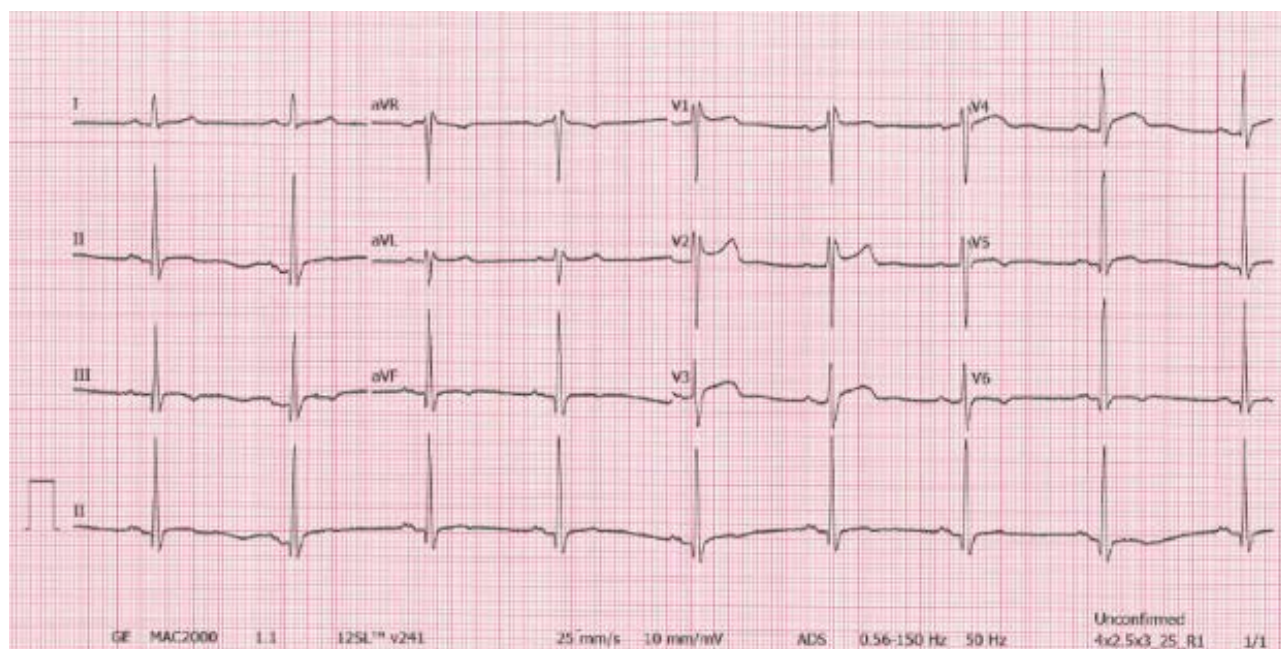


Figure 1. 12 lead ECG with sinus bradycardia with ST/T abnormalities consistent with Brugada pattern.

### Discussion

Brugada syndrome is a rare genetic disorder that affects the electrical system of the heart, predisposing individuals to life-threatening arrhythmias.<sup>2</sup> It was first described by the Brugada brothers in the 1990s and has since gained recognition worldwide. This syndrome is an autosomal dominant genetic disorder with variable expression, characterized by specific electrocardiographic (ECG) patterns, an increased risk of sudden cardiac death (SCD), and a distinct clinical presentation.<sup>2</sup>

The hallmark ECG finding in Brugada syndrome is the presence of a characteristic pattern known as the "Brugada pattern." It is typically observed in the right precordial leads (V1 to V3) as a coved-type ST segment elevation followed by a negative T wave. This ECG pattern can be dynamic and may not always be present, making diagnosis and risk stratification challenging.<sup>3</sup>

The clinical presentation of Brugada syndrome varies from asymptomatic individuals to those who have experienced syncope or life-threatening ventricular arrhythmias, including

ventricular fibrillation (VF) or ventricular tachycardia (VT).<sup>2</sup> These arrhythmias often occur during rest or sleep and can lead to sudden cardiac death if left untreated. The age of symptom onset ranges from infancy to late adulthood, with most cases manifesting between the third and fourth decades of life.

Brugada syndrome has a strong genetic component, with mutations in several genes, including SCN5A, implicated in its pathogenesis. SCN5A encodes the alpha subunit of the cardiac sodium channel, and mutations in this gene lead to altered sodium channel function, resulting in abnormal cardiac repolarization and increased vulnerability to arrhythmias. However, not all individuals with Brugada syndrome have identifiable mutations, suggesting involvement of additional genetic and environmental factors. The majority of affected individuals are Asian descent, with the highest prevalence in Southeast Asia. The Brugada ECG pattern is more common in men than in women with some studies reporting 2-9 times more likelihood in men.<sup>1</sup>

Diagnostic evaluation initially involves obtaining a detailed clinical history, including a thorough assessment of symptoms, such as syncope or palpitations, and a family history of sudden cardiac death or Brugada syndrome. The presence of these symptoms and a positive family history raise suspicion and warrant further investigation.

Electrocardiography (ECG) plays a pivotal role in diagnosing Brugada syndrome. A 12-lead ECG is obtained with particular attention to leads V1 to V3, where the characteristic Brugada pattern is typically observed. The presence of a coved-type ST segment elevation followed by a negative T wave in these leads is suggestive of Brugada syndrome. It is important to recognize that the Brugada pattern may not always be present and can be dynamic, making serial ECG recordings necessary for accurate diagnosis.

Genetic testing can provide valuable information in confirming the diagnosis of Brugada syndrome.<sup>4</sup> Mutations in specific genes, particularly SCN5A, are associated with the syndrome. However, it is important to realize that genetic testing is not always definitive, as some individuals with Brugada syndrome may not have identifiable mutations. A negative genetic test does not exclude the diagnosis.

Additional tests may further assess the risk of arrhythmias in individuals with Brugada syndrome.<sup>2</sup> These include exercise stress testing and the procainamide challenge test, which can unmask the Brugada pattern in some individuals, and programmed electrical stimulation (PES), which evaluates the heart's susceptibility to arrhythmias. These help risk stratification and inform the decision regarding implantable cardioverter-defibrillator (ICD) placement.

Given the potential familial nature of Brugada syndrome, evaluation of family members is crucial. ECG screening should be considered for first-degree relatives of affected individuals,

as they may exhibit the Brugada pattern or have an increased risk of developing symptoms over time.<sup>5</sup>

Management of Brugada syndrome involves a combination of lifestyle modifications and medical interventions. Individuals with the syndrome should avoid factors that can exacerbate the arrhythmias, such as certain medications, fever, and excessive alcohol consumption.<sup>6</sup> Medications like sodium channel blockers (e.g., flecainide, ajmaline) may be used to unmask the ECG pattern and guide risk stratification. However, their role in long-term management is still under investigation.

In conclusion, Brugada syndrome is a complex and potentially life-threatening cardiac disorder characterized by specific ECG patterns and an increased risk of sudden cardiac death. Prompt diagnosis, risk stratification, and appropriate management are needed to reduce the risk of arrhythmias and improve outcomes in affected individuals. Further research is needed to better understand the underlying mechanisms and develop targeted therapies for this challenging condition.

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