TOPIC: SHORT QT SYNDROME (SQTS)

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Introduction

Short QT syndrome (SQTS) is a rare inherited arrhythmogenic disorder characterized by abnormally short QT interval on electrocardiogram (ECG) secondary to a cardiac channelopathy in the absence of structural heart disease.[3] Defective function of the potassium-calcium ion channels in SQTS patients facilitates an increased risk of ventricular and atrial arrhythmias, which can lead to symptoms such as syncope or cardiac arrest.[2]

Having just been recognized as a new clinical entity in 2000 by Gussak et al., SQTS is a relatively novel pathology that has since been extensively investigated in regards to diagnosis and treatment.[1,3] This inheritable channelopathy of the heart is typically defined as QTc ≤330 ms or QTc interval <360 ms, measured via electrocardiogram (ECG), in addition to one or more of the following criteria: history of cardiac arrest, syncope, or family history of SQTS/sudden cardiac death (SCD) at 40-years-old or younger.[1,2,3] Implantable cardioverter defibrillators are often recommended for these patients. Quinidine a class Ia antiarrhythmic agent, has been shown to prolong the QT interval and reduce arrhythmia susceptibility in SQTS patients.[2] Atenolol, a beta-adrenergic receptor blocker, is commonly used to manage tachyarrhythmias. However, its effect on patients with SQTS remains unclear. We present a case of a patient with SQTS who experienced increased ICD firings and QTc prolongation following initiation of Atenolol.

Case Description

This is a case of a 24-year-old male with a past medical history of short QT syndrome with prior cardiac arrest and ventricular fibrillation, status post Boston Scientific subcutaneous ICD placement and cardiac sympathectomy, who presented with complaints of increased palpitations and ICD firing. He was complaint with his follow up visits with his electrophysiologist and has been on Quinidine. Prior to his admission, the patient noted experiencing three episodes of ICD firing since being started on Atenolol. He has been taking Quinidine as scheduled without missing any doses and previously only experienced ICD firing if he missed a dose. He attributes the increase in ICD firings to his recently added medication, Atenolol.

On physical exam, the patient complained of only mild chest pain but reported no other symptoms. ECG revealed QT/QTc of 428/409 ms and sinus bradycardia. Chest X-ray showed no radiographic evidence of acute cardiopulmonary disease. Troponin, CBC, CMP, Magnesium, Phosphorus, and TSH were within normal limits. Atenolol was discontinued and the patient was admitted for observation.

Discussion

SQTS is a rare autosomal dominant disorder that has been found mostly in healthy and young people but is often underdiagnosed.[2] Population-based studies have shown that approximately 0.5% of adults have a QTc <340 ms, making it extremely rare to find patients who fit the diagnostic criteria for SQTS.[3]

SQTS is associated with various, inherited mutations that affect the function of ion channels responsible for regulating cardiac action potentials.[2] Both gain-of-function mutations of potassium channels genes and loss-of-function mutations of calcium channel genes have been observed as causes of SQTS.[3] These mutations result in an abbreviated repolarization phase during a cardiac action potential, ultimately leading to a shortened QT interval as well as both atrial and ventricular tachyarrhythmias.[3] SQTS is identified as a heterogenous disease from both a phenotype and genotype standpoint.[2]

Clinical presentations of SQTS can range from asymptomatic to more serious life-threatening complications.[1] Common presenting symptoms associated with SQTS include arrhythmia, syncope, atrial and ventricular fibrillation, and cardiac arrest.[2,3]

12-lead electrocardiogram (ECG) remains the gold standard study for diagnosing SQTS with the hallmark finding of abnormally short QT/QTc.[1,2,3] Diagnosis of SQTS is currently defined as having a QTc ≤330 ms or QTc interval <360 ms in addition to at least one of the following criteria: no abnormal heart structures, history of cardiac arrest, syncope, or family history of either SQTS or sudden cardiac death (SCD) at 40-years-old or younger.[1,2,3] Genetic testing can also confirm diagnosis, as there are three genes encoding potassium channels that have definitive pathogenic variants: *KCNQ1*, *KCNH2*, and *KCNJ2*.[1]

SQTS can lead to fatal arrhythmias causing sudden cardiac death or premature expiration in patients with SQTS; however, some SQTS patients may never experience or report symptoms.[1,3] The prognosis of those with asymptomatic SQTS has yet to be defined, as there have been reported individuals with QTc <320 who have reached adulthood without developing experiencing any serious arrhythmias.[3]

First-line treatment is implantable cardioverter defibrillator (ICD); however, there have been several reports of inappropriate shock due to sinus tachycardia, atrial fibrillation, as well as the oversensing tall and narrow T waves.[2] Quinidine has been shown to effectively prolong QT interval and ventricular refractory periods, serving as an alternative or adjuvant pharmacological therapy to ICD placement.[2]

The introduction of Atenolol in this patient with short QT syndrome and a history of cardiac arrest and ventricular fibrillation was associated with an escalation in ICD firings and a prolongation of the QTc interval. This observation suggests a potential proarrhythmic effect of Atenolol in patients with SQTS. Beta-blockers are commonly used in the management of various cardiac arrhythmias, including tachyarrhythmias. However, their effects on patients with SQTS, characterized by a short QT interval and an increased susceptibility to ventricular arrhythmias, require careful consideration.

Individualized therapy, close monitoring, and a multidisciplinary approach involving electrophysiologists, cardiologists, and pharmacologists are essential in the management of these patients.

References

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