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Commentary

Pathophysiology of a Rare Genetic Disorder: Short QT Syndrome

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DESCRIPTION

Short QT Syndrome (SQTS) is a rare genetic cardiac disorder characterized by a shortened QT interval on the Electrocardiogram (ECG), which is less than 300 milliseconds. It is associated with an increased risk of sudden cardiac death due to ventricular arrhythmias, including ventricular fibrillation and ventricular tachycardia. In individuals with SQTS, the heart's QT interval, which is the time it takes for the heart to recharge between beats, is shorter than normal.

This shortened QT interval can increase the risk of dangerous arrhythmias, including ventricular fibrillation, which can be life-threatening. SQTS is typically caused by mutations in specific genes that affect the ion channels in the heart, which are responsible for regulating the flow of electrically charged particles in and out of the heart cells. These mutations disrupt the normal electrical activity of the heart, leading to the shortened QT interval and increased risk of arrhythmias. This study discusses about the pathophysiology of SQTS, including the genetic mutations that lead to this condition and the underlying mechanisms that contribute to arrhythmogenesis.

Genetic basis of short QT syndrome

SQTS is a genetic disorder, and mutations in several genes have been identified to cause this condition. These genes encode ion channels or ion channel regulatory proteins that are involved in cardiac repolarization. Mutations in these genes can result in an increase in inward currents or a decrease in outward currents, leading to a shortened QT interval. The most commonly mutated genes in SQTS are *KCNH2*, *KCNQ1*, and *KCNJ2*, which encode the voltage-gated potassium channels hERG, Kv7.1, and Kir2.1, respectively.

Mutations in these genes result in an increase in the potassium current, leading to a shortened action potential duration and a shortened QT interval. Other genes that have been associated with SQTS include *KCNJ8*, *CACNA1C*, and *CALM1*, which encode the inward rectifying potassium channel Kir6.1, the L-type calcium channel Cav1.2, and the calcium-binding protein calmodulin, respectively.

Mechanisms of arrhythmogenesis

The shortened QT interval in SQTS is associated with an increased risk of ventricular arrhythmias, including ventricular fibrillation and ventricular tachycardia. Several mechanisms have been proposed to explain the arrhythmogenesis in SQTS.

Early after depolarizations

EADs are abnormal depolarizations that occur during the repolarization phase of the cardiac action potential. They result from an inward current that prolongs the action potential duration and can trigger a premature ventricular contraction or initiate a reentrant arrhythmia. In SQTS, the shortened action potential duration and increased potassium current can lead to a more positive resting membrane potential, which increases the likelihood of Early after depolarizations (EADs).

Triggered activity

Triggered activity occurs when a Delayed After Depolarization (DAD) triggers a depolarization that reaches the threshold for an action potential. DADs are caused by an increase in intracellular calcium, which activates the sodium-calcium exchanger (NCX), leading to a depolarizing current. In SQTS, the increased inward current can lead to an increase in intracellular calcium and an increased likelihood of triggered activity.

Reentry

Reentry is a common mechanism of arrhythmogenesis in cardiac disorders. It occurs when a propagating wavefront encounters an area of unidirectional block, allowing the wavefront to reenter the tissue and initiate a reentrant circuit. In SQTS, the shortened action potential duration and increased potassium current can lead to a decreased refractory period, which increases the likelihood of reentry.

Clinical implications

SQTS is a rare genetic cardiac disorder that is associated with an increased risk of sudden cardiac death due to ventricular arrhythmias. The diagnosis of SQTS is based on the presence of a

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a shortened QT interval on the ECG and the presence of symptoms or a family history of sudden cardiac death. Treatment options include lifestyle modifications, such as avoiding triggers that can precipitate arrhythmias, and the use of antiarrhythmic drugs or implantable cardioverter-defibrillators (ICDs) to prevent sudden cardiac death.

CONCLUSION

SQTS is a rare genetic cardiac disorder characterized by a shortened QT interval on the ECG. Symptoms of SQTS can vary widely, ranging from no symptoms at all to palpitations, fainting (syncope), dizziness, and sudden cardiac arrest. Diagnosis of SQTS usually involves an Electrocardiogram (ECG) to measure the QT interval, as well as genetic testing to identify any mutations in the associated genes.