Congenital Long QT Syndrome

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PATHOGENESIS

Inherited Disorder of Myocardial Repolarization
Dead Red-polar-bear with Red-hair
Congenital long QT syndrome is a hereditary cardiac disease which leads to abnormally long repolarization of the heart cells due to ion channel defects.

Potassium (K+) Channel Mutations
Mutant coming out of Banana Gate
The most common ion channel defects that cause congential long QT syndrome are loss-of-function mutations affecting the potassium (K+) channels.

SYMPTOMS

Prolonged QT Interval
Stretched QT-heart
The QT interval is the measure of ventricular depolarization followed by repolarization. In patients with congenital long QT syndrome, some of the cardiac myocytes may have an ion channel mutation, leading to an abnormally slow repolarization, leading to prolonged QT interval.

Torsades de Pointes
Tortoise with Points
Torsade de pointes is an arrhythmia characterized as a polymorphic ventricular tachycardia with gradual changing amplitudes and twisting of the QRS complexes around the isoelectric line. Patients with congenital long QT syndrome are most likely to develop torsades as a result of the prolonged repolarization and early afterdepolarizations. These early afterdepolarizations lead to additional action potentials that trigger activity and lead to development of torsades de pointes.

Increased Risk of Sudden Cardiac Death (SCD)
Up-arrow-risk and Dead Heart-Skeleton-guy on Spikes
Sudden cardiac death (SCD) is the sudden cessation of cardiac activity with hemodynamic collapse. This can be caused by sustained ventricular tachycardias such as torsades de pointes.

TYPES OF SYNDROMES

Romano-Ward Syndrome
Roman with Award
Romano-Ward syndrome is the most common form of congenital long QT syndrome. This type is inherited in an autosomal dominant manner and has no sensorineural deafness.

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Autosomal Dominant

Domino-podium

Romano-Ward syndrome has an autosomal dominant pattern. You need only one mutated gene to be affected by this type of disorder. A person with an autosomal dominant disorder has a 50 percent chance of having an affected child with one mutated gene (dominant gene) and a 50 percent chance of having an unaffected child with two normal genes (recessive genes).

Pure Cardiac Phenotype (No Deafness)

Roman with Award on top of Dead Stretched QT-heart

Patients with Romano-Ward syndrome only present cardiac related symptoms.

Jervell and Lange-Nielsen Syndrome

Gerbil having Lunch-in-the-sun

Jervell and Lange-Nielsen syndrome is another congenital long QT syndrome that is characterized by abnormalities regarding the electrical system of the heart but this syndrome also presents with deafness present at birth.

Autosomal Recessive

Recessive-chocolate

Jervell and Lange-Nielsen syndrome demonstrates an autosomal recessive inheritance pattern. This means that both genes in a pair must be abnormal to cause the disease.

Sensorineural Deafness

Ear breaking Sensor-nerve Headphones

Patients with Jervell and Lange-Nielsen syndrome are characterized by congenital profound bilateral sensorineural hearing loss, along with prolongation of the QT interval.

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