

Long QT syndrome (LQTS) is an autosomal dominant disorder, caused by abnormalities of the heart's electrical conduction system, and is characterized on the electrocardiogram (a test that records the electrical activity of the heart) by prolongation of the QT interval that corresponds to prolongation of the recovery phase or repolarization of the heart muscle (ventricular myocardium) after each heartbeat. QT prolongation predisposes those affected to an increased risk of life threatening sudden alterations in the cardiac rhythm, (termed arrhythmias), specifically torsade de pointes (TdP) or ventricular fibrillation (VF). These arrhythmias can lead to sudden loss of consciousness (syncope), cardiac arrest and potentially cause sudden cardiac death. The severity of cardiac symptoms varies greatly from one person to another, even among family members who carry the same rare genetic variant. Some individuals may have no apparent symptoms (asymptomatic) for their entire lives, whilst others develop arrhythmias resulting in episodes of syncope, and cardiac arrest, at a young age. Several different factors are known to trigger the onset of symptoms including physical activity, excitement and fright, although cardiac events may occur while asleep or at rest. Long QT syndrome affects males and females in equal numbers and has been identified in all ethnic groups. The exact incidence and prevalence of the disorder is not known. It is estimated to occur in approximately 1 in 2,000 live births from a clinical and genetic study of 44,500 newborns (neonates).