

## Long QT Syndrome 5 (KCNE1)

Long QT syndrome 5 (LQT5) is a heart condition caused by defects in the KCNE1 gene, which encodes for a protein involved in potassium channel regulation. Potassium channels in the heart muscles are important for maintaining a consistent heartbeat. Long QT refers to the elongation of the heartbeat, depicting an abnormal wave pattern on an electrocardiogram (ECG) seen with LQT patients. Such patients are at risk of episodes of increased heart rate, known as torsades de point, which may result in fainting or cardiac arrest. Death can sometimes occur, even in young people, so it is important that any fainting episodes are properly investigated. Treatments are by means of beta-blockers or implantable cardioverter-defibrillators (ICDs). Long QT syndrome types such as this, which only affect the heart, are also known as Romano-Ward syndrome.

Overall, it's estimated that about 1 in 2,000 people, suffer from LQT, some without knowing it. The condition does not seem to be more prevalent in any ethnic group. LQT5 (KCNE1 gene) only makes up <1% of the total cases of LQT, leading to less than 1 in 200,000 having the condition. The affected gene is inherited in an autosomal dominant manner, which normally is inherited from one parent who also has the condition.

### Sources

Alders, M. & Christiaans, I. (2003), "Long QT syndrome," in Pagon, R.A. et al., editors, *GeneReviews* [Internet]. See <http://www.ncbi.nlm.nih.gov/books/NBK1129/>

NIH, Genetics Home Reference: KCNE1 gene.  
See <http://ghr.nlm.nih.gov/gene/KCNE1>

NIH, Genetics Home Reference: Romano-Ward Syndrome.  
See <http://ghr.nlm.nih.gov/condition/romano-ward-syndrome>

Splawski, I. *et al.* (1997), "Mutations in the hminK gene cause long QT syndrome and suppress IKs function," *Nature Genetics*, 17, 338-340.  
See <http://www.ncbi.nlm.nih.gov/pubmed/9354802>