Condition: Long QT syndrome

Inheritance:

Genetically heterogeneous.

Genetic etiology:

Most are due to mutation in various genes encoding sodium and potassium ion channels. Romano Ward syndrome (*LQT 1-6)* due to mutations in *KCNQ1, KCNH2, SCN5A, KCNE1,* KCNE2, encoding potassium or sodium channels. Jervell and Lange-Nielsen syndrome due to mutation in *KCNQ1 (LQT1)* or *KCNE1 (LQT2),* both potassium channels. Andersen-Tawil due to mutations in *KCNJ2* encoding inward rectifier potassium channel.

Frequency:

Romano-Ward: 1/7,000; Jervell and Lange-Nielsen: 1.6 – 6/million; Andersen-Tawil syndrome: rare.

Clinical features:

Long QT syndrome is a disorder in which there is prolonged ventricular repolarization, leading to cardiac arrhythmia. There are three major syndromes, each of which is itself genetically heterogeneous. Romano-Ward is isolated long QT and is dominantly inherited. Jervell and Lange-Neilsen syndrome is long QT with sensorineural deafness and is autosomal recessive. Andersen-Tawil syndrome includes long QT, dysmorphic features, and periodic muscle weakness and is autosomal dominant.

Management:

Treatment with beta blocker medication or insertion of cardiac pacemaker.

Genetic counseling:

Based on establishing correct diagnosis; genetic testing available for some of the responsible genes. Individuals at risk should be carefully evaluated, since sudden death is possible in untreated individuals.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.