

Timothy syndrome is a rare genetic disorder that affects the heart and several other organs. The most obvious manifestation is the abnormally long repolarization (QT interval) in the heart that predisposes patients to cardiac arrhythmias and sudden cardiac death. Unlike other forms of long QT syndrome, patients with Timothy syndrome also present with cardiac malformations, thickening of the cardiac walls (cardiac hypertrophy) and enlargement of the heart (cardiac dilatation). Timothy syndrome has been diagnosed in an extremely low number of individuals in the world because it has a high mortality, it is a complex disease, and few affected individuals reproduce.