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Scientists discover KCNH2 gene mutation in patients with long QT syndrome

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Researchers from the Hospital Virgen de las Nieves of the University of Granada have identified the most frequent mutations in the gene KCNH2 in patients with long QT syndrome.

Long Qt syndrome is a disorder of cardiac ionic channels that approximately affects one in every 2,500 people and may cause torsade de pointes episodes, which can trigger sudden death. This condition usually affects children and adolescents, and it is occasionally mistaken for convulsions, leading to a misdiagnosis of epilepsy.

So far, hundreds of mutations have been found in twelve genes of sodium and potassium channels. Thus, approximately 75% of the mutations in cases of LQTS are located in three genes: KCNQ1, the most frequent in other sectors of the population (potassium channel), KCNH2 (potassium channel) and SCN5A (sodium channel).

To carry out this study, researchers selected nine patients who met the diagnostic criteria for long QT syndrome, and four patients with ventricular fibrillation (cardiac arrest produced in the absence of any identifiable causes). These patients and their first -degree relatives were examined in the Arrhythmia Assessment Unit of the Hospital Virgen de las Nieves in Granada, Spain.

Genetic Study

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