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Update on Long QT Syndrome

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Abstract

Congenital long QT syndrome (LQTS) is the Rosetta stone in which the association between genotype and phenotype is most evident. Congenital LQTS is characterized by the prolongation of the QT interval and Torsade de Pointes (TdP) leading to syncope or sometimes sudden cardiac death. Genotype-Phenotype Correlation has been investigated in the International LQTS Registry. We previously investigated the clinical course of 858 LQT2 patients with *KCNH2* mutations derived from the International registry of U.S., the Netherlands, and Japan. The LQT2 patients with missense mutations located in the pore region (S5-loop-S6) showed the greatest risk for arrhythmic cardiac events than those with missense mutations in other regions. We conducted the Japanese Congenital LQTS Multicenter Registry including 1124 congenital LQTS patients, who were genotyped as LQT1, LQT2 or LQT3 syndrome (616 probands and 508 family members). Our findings suggest that pathogenic variants in the pore region were associated with higher arrhythmic events than were other variants in each genotype, while sex-associated differences were observed in LQT2 but not in those with LQT1 and LQT3. Risk for cardiac events in LQTS appear to vary according to genotype, variant site, age, and sex.