Clinical and Genetic Features of Long QT Syndrome in Korean population

Jue Seong Lee  
Jong-Il Choi  
Young Ran Jeon  
Yun Gi Kim  
Suk-Kyu Oh  
Ki Yung Boo  
Do young Kim  
Jaemin Shim  
Young-Hoon Kim

Introduction: Long QT syndrome is an inherited cardiac disease caused by abnormalities in cardiac ion channel. Untreated, symptomatic patients have a high mortality rate, with a mortality rate of 21% within 1 year of symptom onset. The aim of this study is to investigate the characteristics of LQTS in Korean population.

Methods: We studied the updated medical records from last year's data with confirmed LQTS from Korean Inherited Arrhythmia Registry and Korea University Hospital from January 2013 to June 2019.

Result: Total 61 LQTS patients were enrolled. The number of female patients was more prevalent with 48 patients(78.6%). Median age of diagnosis was 35.7±17 years old. Aborted cardiac arrest was the most common symptom at the time of diagnosis(30 patients, 49.1%), followed by presyncope or syncope(16 patients, 26.2%). Twelve patients had no symptoms at the time of diagnosis(19.6%). And total 57 patients had experienced syncope at least once(93.4%). Ventricular fibrillation was documented in 21 patients(34.4%) and ventricular tachycardia was documented in 13 patients(21.3%). Epinephrine test was performed in 18 patients, and 12 patients of them had positive result(66.7%). Mean corrected QT interval was 505.4±58.5ms. QTc longer than 480ms was 39 patients(63.9%) and The proportion of QTc≥480ms was higher in female patients(p=0.03) Unexplained sudden cardiac death(SCD) under 30 years of age among family members was found in 6 patients(9%) and under 50 years was found in 8 patients(13%). There was 47 patients(77%) taking beta-blocker and implantable cardioverter defibrillators were applied to 38 patients(62.3%). Genetic testing was done in 43 patients. 17 patients was confirmed genetic mutations(KCNQ1 6 patients, KCNH2 5 patients, SCN5A 4 patients, others 4 patients).

Conclusion: In Korean LQTS patients, most patients experienced syncope and nearly half of the confirmed LQTS patients(49.1%) were experienced cardiac arrest at the time of diagnosis. The diagnostic yield of major LQTS gene mutations were not higher than previously reported.