Congenital long QT syndrome: a case report of LQT2 and LQT13 in a neonate

Rachel Bond, Andrew Blaufox, Bruce Goldner, and Apoor Patel
Hofstra North Shore, LIJ School of Medicine, Long Island, NY, USA

Long QT syndrome (LQTS) is a disorder of ventricular myocardial repolarization characterized by a prolonged QT interval on the electrocardiogram (ECG). At least 12 different genes in patients with congenital LQTS have been described, designated LQT 1 through 12. However, more recently at least 13 types of congenital LQTS have been identified.

The neonate, who was born prematurely, was noted to have increased ventricular ectopy on the monitor. An ECG obtained at the time demonstrated sinus bradycardia with QTc of 532 ms. An echocardiogram demonstrated a patent foramen ovale and propranolol was started. Genetic testing of the neonate revealed a pore, missense mutation in KCNH2 (LQT 2) and a nonsense mutation in KCNJ5 (LQT 13). Each parent underwent testing. Her mother was found to have a mutation in KCNH2 along with phenotypic evidence of LQT, prolonged QTc on ECG. Her father had a mutation in KCNJ5 but no phenotypic evidence of LQT.

LQT13 described in a Chinese family with a missense mutation (Gly387Arg) in the KCNJ5 gene. Our patient, not of Asian descent, was found to have a deleterious mutation in the gene KCNH2 and the already mentioned KCNJ5. There are no documented patients with deleterious mutations of both genes.

The full-length version of this report can be viewed at: http://www.escardio.org/communities/EHRA/publications/ep-case-reports/Documents/Congenital-long.pdf.