Síndrome de Andersen Tawil Tipo1 (ATS1)

Dr. Andrés R. Pérez Riera

Nuestra colega Dra Li Zhang há realizado notables investigaciones en el campo de las canalopatias en especial en el síndrome de QT largo y la DAVD y descubrimientos especiales que muestran la gran capacidad de esta investigadora.
Ella descubrió con la paciencia de los Asiáticos que en los pacientes portadores del síndrome de Andersen Tawil tipo 1 (ATS1), los patrones de ondas T-U específicos del gen resultan de la disminución de IK1 debido a mutaciones de KCNJ2 que pueden ayudar al diagnóstico y la genotipificación directa. El QTc normal, el ECG distinto y otras características clínicas distinguen a ATS1 del síndrome de QT largo, y es mejor designarlo como ATS1 en lugar de LQT7. Genial observacion de Li publicado hace 15 anos en el Circulation.
Nosotros hicimos una revisión reciente
https://europepmc.org/article/med/32947483

Li Zhang 1, D Woodrow Benson, Martin Tristani-Firouzi, Louis J Ptacek, Rabi Tawil, Peter J Schwartz, Alfred L George, Minoru Horie, Gregor Andelfinger, Gregory L Snow, Ying-Hui Fu, Michael J Ackerman, G Michael Vincent Electrocardiographic features in Andersen-Tawil syndrome patients with KCNJ2 mutations: characteristic T-U-wave patterns predict the KCNJ2 genotype. Circulation . 2005 May 31;111(21):2720-6. doi: 10.1161/CIRCULATIONAHA.104.472498. Epub 2005 May 23.

Affiliations expand

LDS Hospital, 324 10th Ave, Suite 130, Salt Lake City, Utah 84103, USA.
PMID: 15911703 DOI: 10.1161/CIRCULATIONAHA.104.472498
Abstract
Background: The ECG features of Andersen-Tawil syndrome (ATS) patients with KCNJ2 mutations (ATS1) have not been systematically assessed. This

study aimed to define ECG features of KCNJ2 mutation carriers, to determine whether characteristic T-U-wave patterns exist, and to establish whether T-U patterns predict the ATS1 genotype.

Methods and results: In phase I, evaluation of T-U morphology in ECGs of 39 KCNJ2 mutation carriers identified characteristic T-U patterns: prolonged terminal T downslope, wide T-U junction, and biphasic and enlarged U waves. In phase II, ATS1 genotype prediction by T-U pattern was evaluated in the next 147 ECGs (57 other KCNJ2 mutation carriers, 61 unaffected family members, and 29 ATS patients without KCNJ2 mutations), with a sensitivity of 84% and specificity of 97%. Characteristic T-U patterns were present in 91% (87/96), in whom an enlarged U wave was predominant (73%). In phase III, QTc, QUc, and T- and U-wave duration/amplitude were compared in the 96 ATS1, 29 non-KCNJ2 ATS, and 75 normal subjects. In ATS1 patients, QUc, U-wave duration and amplitude, and QTc were all increased ($P<0.001$), but median QTc and interquartile range (IQR) were just 440 ms (IQR, 28 ms) compared with 420 ms (IQR, 20 ms) in normal subjects and 425 ms (IQR, 48 ms) in ATS non-KCNJ2 patients.

Conclusions: In ATS1 patients, gene-specific T-U-wave patterns resulting from decreased IK1 owing to KCNJ2 mutations can aid diagnosis and direct genotyping. The normal QTc, distinct ECG, and other clinical features distinguish ATS1 from long-QT syndrome, and it is best designated as ATS1 rather than LQT7.