Phenotype and Genotype Relationship between Inherited Cardiac Arrhythmia and Epileptic Syndrome

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Inherited channel disorders are associated with paroxysmal dysfunction of excitable tissues and manifest as diseases of the heart and brain. Because of similar clinical manifestations, inherited cardiac arrhythmias can be misdiagnosed as epilepsy. The relations between inherited cardiac arrhythmias and epilepsy are not completely understood. The purpose of this study was to test the hypothesis that similar channelopathies can underlie both inherited cardiac arrhythmias and epilepsy with genetic sequence analysis.

Methods: Sequence alignments of the amino acid in the genes associated with inherited cardiac arrhythmias and epilepsy were obtained in the Entrez retrieval system and sequence homology between BrS, LQTS and epilepsy was determined by using the Genetyx program. Results: Minimum number of amino acid (aa) sequences was 200 for the analysis of homology. The KCNQ1 mutation showed homology 38.3%/614aa with the KCNQ2 and 39.6%/520aa with KCNQ3. The SCN5A mutation showed homology 46.9%/1959aa with SCN1A and 56.3%/2032aa with SCN2A, respectively. No significant sequence similarities were found between other gene mutations. Conclusions: These results indicate that the pathoetiologies of LQTS, BrS and epilepsy could partly overlap through the α subunit channels and have raised the possibility of a link between cardiac and neural channelopathies.

Keywords: channelopathy, Brugada syndrome, Long QT syndrome