GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

H558R polymorphism in *SCN5A* is associated with Keshan disease and QRS prolongation in Keshan disease patients

S. Jiang[1], F.L. Li[2], Q. Dong[2], H.W. Liu[2], C.F. Fang[2], C. Shu[2], H. Cheng[2], J. Cui[2], H.X. Ma[3], D.Q. Chen[4], H. Li[2]

[1]Department of Pharmacology, Institute of Medical Sciences, Shanghai Jiao Tong University School of Medicine, Shanghai, China
[2]Department of Biochemistry and Molecular Biology, Harbin Medical University, Harbin, China
[3]Daqing First Hospital, Da Qing, China
[4]Qiqihar First Hospital, Qiqihar, China

Corresponding author: H. Li
E-mail: lihui@ems.hrbhmu.edu.cn

Genet. Mol. Res. 13 (3): 6569-6576 (2014)
Received October 31, 2012
Accepted July 18, 2013
Published August 28, 2014
DOI http://dx.doi.org/10.4238/2014.August.28.1

ABSTRACT. Keshan disease (KSD), a potentially fatal cardiomyopathy, has very high incidence in some selenium-poor regions of China. KSD may be accompanied with a variety of arrhythmia, which is associated with mutations in the gene coding for cardiac voltage-gated sodium channel (*SCN5A*). The molecular mechanism of KSD is still largely obscure. We aimed to determine the association between the H558R polymorphism of *SCN5A* and KSD. We recruited 71 patients with KSD and 80 geographical region-matched control subjects in our study. Vital sign and electrocardiographic (ECG) measurements were performed for heart rate, systolic pressure, diastolic pressure, PR interval, QT interval, QRS duration, ST-T changes and complete right bundle branch block (CRBBB), and H558R polymorphism was genotyped using the polymerase chain

reaction single-strand conformation polymorphism (PCR-SSCP) method and sequencing. A significant association was found between the H558R polymorphism of exon 12 and KSD. Allele C carriers had a decreased risk for KSD with an odds ratio of 0.332 [95% confidence interval (CI), 0.160-0.692] as well as for QRS prolongation in KSD patients with an odds ratio of 0.089 (95%CI, 0.022-0.361). Our results provide support to the association between H558R polymorphism and the decreased risk for KSD. H558R polymorphism might increase susceptibility to KSD, and *SCN5A* containing the polymorphism might be a predisposing gene for QRS prolongation.

Key words: Keshan Disease, *SCN5A*, Polymorphism, H558R, QRS Duration

