CP3-04 Genetic Analysis of SCN5A in Korean Patients Associated with Complete Atrioventricular Block

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Recent studies showed that the genetic variation of SCN5A is related with atrioventricular block. But there was no study included Korean patients. To determine the complete atrioventricular block (CAVB) associated genetic variation in Korean patients, we investigated the genetic variation of the SCN5A in Korean patients with CAVB. We enrolled 10 patients with CAVB and 30 normal control. DNA was isolated from the each peripheral blood, and all the exons except untranslated region and exon-intron boundaries of the SCN5A gene were amplified by PCR and directly sequenced using an ABI PRISM 3100 Genetic Analyzer. When a variation was discovered in genomic DNA from AVB patients, confirmed whether the same variation existed in the control genomic DNA. A total of 7 genetic variations (5 known and 2 novel) were detected in 10 AVB patients. Of the 7 variations, 5 (G87A-A29A, IVS9-3C>A, A1673G-H558R, G3578A-R1193Q, T5457C-D1819D) have been reported in previous studies and 2 (C48G-F16L, G3048A-T1016T) were novel variations which have not been reported. Newly discovered 2 variations, non-synonymous change (C48G-F16L) and synonymous (G3048A-T1016T) were not found in 30 normal control group. In conclusion, we found 2 novel genetic variations (C48G-F16L, G3048A-T1016T) in the SCN5A gene in Korean patients with CAVB.

Keywords: SCN5A, atrioventricular block