Objectives Background and objective: Dilated cardiomyopathy (DCM) is a group of heterogenic cardiomyopathy characterised by ventricular dilation and dysfunction. It is the third main culprit leading to heart failure and the most common cause of heart transplantation. Autoimmunity is one of the most important pathogenic mechanisms of DCM. Recent studies have reported two single nucleotide polymorphism (SNP) rs763361 and rs727088 within the last exon of CD226 gene were associated with multiple autoimmune diseases. However, it is still unknown whether these SNPs also affect patients’ susceptibility to DCM. In this study, we aim to investigate whether the SNPs rs763361 and rs727088 in CD226 gene are associated with DCM.

Methods Totally 308 DCM patients and 389 control subjects were recruited in this study. PCR-restriction fragment length polymorphism (PCR-RFLP) was used to genotype all the SNPs.

Results The frequency of T allele for SNP rs763361 was found to be significantly increased in the DCM group compared with the control group (44.8% vs 36.2%, OR = 0.700, 95% CI 0.564 to 0.869, p = 0.00120). Also, the frequency of TT genotype of SNP rs763361 in DCM patients compared with healthy controls were significantly different (14.9% vs 11.3%, p = 0.00076). Meanwhile, the frequency of G allele of SNP rs727088 was found to be significantly higher in the DCM group than the control group (33.3% vs 23.7%, OR = 0.621, 95% CI 0.491 to 0.786, p < 0.00100). The distributions of GG genotype at SNP rs727088 existed significantly difference between the DCM group and the control group (8.8% vs 3.3%, p = 0.00011).

Conclusions Our results indicate that the association of the CD226 gene SNPs with human DCM, and the allele T at SNP rs763361 and allele G at SNP rs727088 in CD226 gene may increase the risk of DCM.