Interleukin-21 Receptor Gene Polymorphisms in Kawasaki disease

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Introduction: It has been known that interleukin-21 receptor (IL-21R) gene polymorphism is related with the development of systemic vasculitis. We explored the polymorphisms of IL-21R gene in patients with Kawasaki disease (KD).

Materials and methods: We genotyped the promoter region of IL-21R gene (-2500 bp to +1 bp) in 100 patients with KD and 100 healthy controls who are all Koreans.

Results: We found the 5 single nucleotide polymorphisms (SNPs) of which minor allele frequency > 0.01 in the promoter region of IL-21R gene. Those are -1681 G>T (chromosome site 27411802), -380 G>A (27413104), -332 G>C (27413151), -237 A>T (27413246), and -53 G>A (27413430). There is no significant difference in minor allele frequency of each SNP between patients with KD and healthy controls except -237 A>T. Twenty five patients with KD had more than 1 SNP while 7 healthy controls had. There were more individuals who have any SNPs of IL-21R gene in patients with KD than the controls (odds ratio: 3.0, 95% CI: 1.6-5.6, P = 0.0005). There was no significant correlation between IL-21R gene polymorphisms and the serum level of IL-21 in patient with KD. The serum level of total IgE was not affected by the existence of IL-21R gene polymorphisms in this study.

Conclusion: Our data suggest that the genetic susceptibility profile for KD may include IL-21R gene.