

Interleukin-21 receptor gene polymorphisms in Kawasaki disease

Hae Yong Lee¹, Mi-Hyun Kim¹, Yeong Ro Lee², Dong Hoon Lee², Kiho Bae², Sang Baek Koh³, Mee Kyung Namgoong¹, Byung Ho Cha¹, Baek Keun Lim¹, Jin-Kyong Chun¹

¹Departments of Pediatrics and ³Preventive Medicine, Wonju College of Medicine, ²Division of Biological Science and Technology & Yonsei-Fraunhofer Medical Device Lab, College of Science and Technology, Yonsei University, Wonju, South Korea

Purposes

- 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.
- We searched any single nucleotide polymorphisms by comparing with reference sequences of *IL-21R* gene
- (NG_012222.1/NC_000016.9).

Conclusions

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

Results

Identified SNPs in promoter region of interleukin-21 receptor (*IL-21R*) gene.

Chr position*	Position†	SNPs	MAF in subjects‡
27411802	-1681	G→T	0.15
27413104	-379	G→A	0.06
27413151	-332	G→C	0.145
27413246	-237	A→T	0.075
27413430	-53	G→A	0.04

*: reference sequence: NC_000016.9

†: from the starting transcription site (+1); Chr position:

27413483

‡: minor allele frequency in controls and patients

Comparison in terms of allele frequencies of the SNPs in promoter region of *IL-21R* gene between patients with KD and healthy controls.

SNPs	KD	Control	OR (95% CI)	P
-1681 G>T	11/51 (21.6)	12/100 (12.0)	2.0 (0.8-5.0)	0.15
-379 G>A	9/100 (9.0)	3/100 (3.0)	3.2 (0.8-12.1)	0.13
-332 G>C	16/100 (16.0)	13/100 (13.0)	1.3 (0.6-2.8)	0.68
-237 A>T	15/100 (15.0)	0	36 (2.1-618)	< 0.0001
-53 G>A	5/100 (5.0)	3/100 (3.0)	1.7 (0.4-7.3)	0.72

The difference of frequencies of individual who has any SNPs between patients with KD and healthy controls

	Patients with KD	Healthy Controls	OR (95% CI)	P
Any SNPs	45	21	3.0 (1.6-5.6)	0.0005
No SNP	55	79		