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Association between *IL-4* gene polymorphisms, IL-4 serum levels, and ankylosing spondylitis

X.L. Liu[1], J.K. Ren[2] and Y.L. Su[1]

[1]Clinical laboratory, Shangluo Central Hospital, Shangluo, China
[2]Clinical laboratory, Shanxi Provincial People's Hospital, Xi'an, China

Corresponding author: X.L. Liu
E-mail: liuxiaoliang6003@163.com

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ABSTRACT. We aimed to investigate the effect of two common polymorphisms in interleukin-4 (*IL-4*) on serum IL-4 levels and the development of ankylosing spondylitis (AS) in the Chinese population. A total of 420 inpatients and outpatients diagnosed with AS were enrolled as the case group, and 330 healthy volunteers were selected as the control group. *IL-4* rs2243250 and rs2227282 genotype frequencies in the latter were consistent with Hardy-Weinberg equilibrium (both $P > 0.05$). The TC+TT genotypes and T allele of rs2243250 were strongly associated with elevated AS risk [CC *vs* TC+TT: odds ratio (OR) = 2.378, 95% confidence interval (CI) = 1.746-3.239, $P < 0.001$; C *vs* T: OR = 2.588, 95%CI = 2.007-3.337, $P < 0.001$]. Moreover, the rs2227282 GG genotype and G allele may also correlate with increased risk (CC *vs* GC: OR = 1.555, 95%CI = 1.130-2.141, P = 0.007; CC *vs* GC+GG: OR = 1.833, 95%CI = 1.357-2.476, $P < 0.001$; C *vs* G: OR = 1.403, 95%CI = 1.086-1.811, P = 0.009). In addition, serum IL-4 concentrations were significantly lower in AS patients carrying the rs2243250 TT genotype compared to those with the CC and TC

genotypes (both $P < 0.05$). Similarly, patients carrying the rs2227282 CC genotype demonstrated higher serum IL-4 levels than those with the GC and GG genotypes (both $P < 0.05$). Our study provides evidence that *IL-4* polymorphisms associated with diminished serum IL-4 levels may be partially responsible for AS development in the Chinese population.

Key words: Interleukin-4; Ankylosing spondylitis; Serum levels; Single nucleotide polymorphism; *IL-4* rs2243250; *IL-4* rs2227282