Association study between matrix metalloproteinase-9 gene (MMP9) polymorphisms and the risk of Henoch-Schönlein purpura in children

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ABSTRACT. Henoch-Schönlein purpura nephritis (HSPN), the most serious long-term complication of Henoch-Schönlein purpura, is one of the most common renal diseases in children. Matrix metalloproteinase-9 (MMP-9) is implicated in the pathogenesis of renal diseases. Genomic DNA was isolated from the venous blood leukocytes of 220 unrelated patients with HSPN and 205 unrelated healthy individuals. To identify markers contributing to genetic susceptibility to HSPN, this study examined the potential association between HSPN and four single nucleotide polymorphisms of the MMP-9 gene (MMP9) (rs17576, rs3918254, rs3787268, and rs2236416) by using the MassARRAY system. The allelic or genotypic frequencies of the rs17576 (exon 6) and rs3918254 (intron 6) polymorphisms in HSPN were significantly different from those in the healthy controls. The HSPN subjects had a significantly higher frequency of the G allele of rs17576 ($\chi^2 = 8.416$, P
= 0.004, OR = 1.556, 95% CI = 1.153-2.100) and a significantly lower frequency of the A allele of rs2236416 ($\chi^2 = 10.363, P = 0.001, \text{OR} = 0.545, 95\%CI = 0.375-0.791$). Linkage disequilibrium was observed in two blocks ($D' > 0.9; r^2 > 0.8$ in control). In block 1, significantly more G-C haplotypes ($P = 0.011$) and significantly fewer A-C haplotypes ($P = 0.008$) were found in the HSPN subjects. In block 2, significantly more G-G haplotypes ($P = 0.016$) and significantly fewer A-G haplotypes ($P = 0.006$) were found in the HSPN subjects. These observations suggest that the rs17576 and rs3918254 polymorphisms of $MMP9$ are associated with HSPN.

**Key words:** Henoch-Schönlein purpura nephritis; Matrix metalloproteinase-9; Single nucleotide polymorphisms