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Effect of *RAGE* polymorphisms on susceptibility to and severity of osteoarthritis in a Han Chinese population: a case-control study

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ABSTRACT. Recent studies have revealed that the inflammatory process plays a role in the pathogenesis of osteoarthritis (OA). The S100 family and receptor for advanced glycation end products (RAGE)

participate in regulating inflammation, even in the production of matrix metalloproteinases (MMPs). MMP-1 degrades cartilage, which may result in OA development. Moreover, polymorphisms in *RAGE*, *S100A8*, and *MMP-1* have a marked effect on ligand binding and transcription regulating. In this study, we investigated the potential genetic contribution of the *RAGE*, *S100A8*, and *MMP-1* genes to OA. We performed a matched case-control association study and genotyped OA patients and healthy controls, who were analyzed by polymerase chain reaction-restriction fragment length polymorphism assays. A total of 207 patients were diagnosed with knee OA and underwent total knee replacement. The control group included 207 individuals who had standard X-rays of the knee joints to confirm K/L < 2 and were matched by age and gender. Single-nucleotide polymorphisms in *RAGE* (-429T/C, -374T/A, and 557G/A), *S100A8* (rs3795391A/G), and *MMP-1* (-1607 1G/2G, -755G/T, and -519A/G) were evaluated. *RAGE* -374T/A, S100A8 rs3795391A/G, *MMP-1* -1607 1G/2G, -755G/T, and -519A/G showed no significant difference between OA patients and healthy controls. *RAGE* -429T/C and 557G/A showed a significant association between OA patients and healthy controls (P = 0.016 and 0.047, respectively). In haplotype analyses, no *RAGE* and *MMP-1* haplotypes showed associations with OA. Our results suggest that the investigated polymorphism in the *RAGE* gene play a role in OA in the Han Chinese population.

Key words: Matrix metalloprotease-1; Osteoarthritis; Polymorphism; Receptor for advanced glycation end products; S100A8

