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Lack of association between rare mutations of the *SIAE* gene and rheumatoid arthritis in a Han Chinese population

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ABSTRACT. The function of rare genotypes encoding defective variants of sialic acid acetylesterase (SIAE) in some autoimmune diseases, including rheumatoid arthritis (RA), is ambiguous. We determined whether mutations in the *SIAE* gene are responsible for RA in a Han Chinese population. DNA was prepared from the venous leukocytes of 444 RA patients and 647 normal controls. The coding regions and adjacent intron sequences of *SIAE* were amplified by polymerase chain reaction. The products were

then subjected to sequencing analysis. The detected variations were further evaluated in the normal controls and available family members by sequencing. Seven variants of RA were identified in this study, including four known single nucleotide polymorphisms SNPs (rs7941327, rs7941523, rs1942663, and rs12282107) and three novel SNPs. The genomic positions of the three novel SNPs are chr11:124013712, chr11:124023268, and chr11:124044505. No significant differences in the seven SNPs of *SIAE* were observed between patients with RA and controls in this cohort ($P > 0.05$). Three novel variations and four known SNPs in *SIAE* were detected in the Chinese RA patients and normal controls. Our results imply that *SIAE* does not play a major role in RA in this population.

Key words: Rheumatoid arthritis; Sialic acid acetylesterase; Sequencing; Single nucleotide polymorphism; Han Chinese population