



# Association between *PDCD1*, *CTLA4*, and *MECP2* gene polymorphisms and systemic lupus erythematosus in the Chinese Northern Han

H.R. Dong<sup>1\*</sup>, H.S. Li<sup>1\*</sup>, S.C. Wang<sup>1</sup>, Q.M. Balin<sup>1</sup> and P.Y. Chang<sup>2</sup>

<sup>1</sup>Clinical Laboratory Department, First Hospital of Hohhot in Inner Mongolia, Hohhot, China

<sup>2</sup>Department of Nuclear Medicine, Inner Mongolia Medical University Affiliated Hospital, Hohhot, China

\*These authors contributed equally to this study.

Corresponding author: P.Y. Chang

E-mail: changpeiye@126.com

Genet. Mol. Res. 14 (4): 17567-17573 (2015)

Received April 22, 2015

Accepted August 13, 2015

Published December 21, 2015

DOI <http://dx.doi.org/10.4238/2015.December.21.29>

**ABSTRACT.** Systemic lupus erythematosus (SLE) is an autoimmune disease that results in chronic inflammation of different organ systems. Several susceptibility loci for SLE have been suggested in different populations, but the nature of the susceptibility genes has yet to be determined. The programmed cell death 1 gene (*PDCD1*), the cytotoxic T-lymphocyte-associated protein 4 (*CTLA4*) gene, and the methyl-CpG-binding protein 2 gene (*MECP2*) are considered to be the candidate genes associated with SLE. We analyzed the role of *PDCD1*, *CTLA4*, and *MECP2* gene polymorphisms in Han patients suffering from SLE. Using a case-control study, 263 SLE patients and 263 healthy controls were collected from Chinese Northern Han people. Genomic DNA was prepared from peripheral blood leukocytes and the genotyping was performed using a polymerase chain reaction/ligase detection reaction assay. A statistically

significant difference was observed in the rs2239464 and rs2075596 polymorphisms of *MECP2* between SLE subjects and controls. The GG genotype in rs2239464 and the GG genotype in rs2075596 might protect against SLE. In contrast, no such association was found in the *CTLA4* or *PDCD1* polymorphisms. The rs2239464 and rs2075596 polymorphisms of *MECP2* might play a significant role in the development of SLE in the Northern Han of China.

**Key words:** Systemic lupus erythematosus; *PDCD1*; *CTLA4*; *MECP2*; Single nucleotide polymorphism