GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Association between *C1GALT1* variants and genetic susceptibility to IgA nephropathy in Uygur

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Genet. Mol. Res. 14 (2): 5327-5333 (2015)
Received April 9, 2014
Accepted October 7, 2014
Published May 22, 2015
DOI http://dx.doi.org/10.4238/2015.May.22.3

ABSTRACT. Immunoglobulin A (IgA) nephropathy (IgAN) is a common form of primary glomerulonephritis characterized by diffuse glomerular mesangial IgA1 deposition leading to mesangial proliferation and chronic glomerular inflammation. Analyses of serum IgA1 from IgAN patients revealed abnormal galactosylation of the *O*-linked carbohydrate moieties of IgA that may result from altered activity in the core of 1 β1,3-galactosyltransferase (C1GalT1). To evaluate the association between *C1GalT1* single nucleotide polymorphisms (SNPs) and IgAN, we performed a case-control study on cohorts from the Uyghur population in China. A total of 180 IgAN patients and 180 healthy controls were recruited for the study. We sequenced 5 SNPs, including SNP1 (rs9639031), SNP2 (-527A/G), SNP3 (rs1008898), SNP4 (rs5882115), and SNP5 (rs1047763) in the *C1GalT1* gene in all eligible participants. The frequencies of the I allele and DI genotype of rs5882115 in IgAN patients were significantly higher than those in controls ($P < 0.05$). The frequency of haplotype GAGDA was significantly higher in patients than in controls (0.0719 *vs* 0.00, $P = 0.024$). Polymorphisms

in the *C1GALT1* gene were associated with genetic susceptibility to Uyghur IgAN.

Key words: *C1GALT1* gene; Gene polymorphism; IgA nephropathy; Uyghur

