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

Association of *TUSC3* gene polymorphisms with non-syndromic mental retardation based on nuclear families in the Qinba mountain area of China

M.J. Zhang[1]*, L.X. Xing[1]*, M. Cui[2], X. Yang[1], J.G. Shi[2], J. Li[3], K.J. Zhang[1], Z.J. Zheng[3], F.C. Zhang[1,3], J.L. Li[1] and X.C. Gao[1,3]

[1]Key Laboratory of Resource Biology and Biotechnology in Western China, Ministry of Education, Institute of Population and Health, College of Life Science, Northwest University, Xi'an, Shaanxi, China
[2]The Mental Health Center of Xi'an, Xi'an, Shaanxi, China
[3]Institute of Application Psychology, College of Public Management, Northwest University, Xi'an, Shaanxi, China

*These authors contributed equally to this study.
Corresponding authors: X.C. Gao / J.L. Li
E-mail: gaoxc@nwu.edu.cn / ljl@nwu.edu.cn

Genet. Mol. Res. 14 (2): 5022-5030 (2015)
Received August 8, 2014
Accepted December 10, 2014
Published May 12, 2015
DOI http://dx.doi.org/10.4238/2015.May.12.5

ABSTRACT. TUSC3 interacts with the protein phosphatase 1 and magnesium ion transport system, which plays an important role in learning and memory. Abnormal conditions of learning and memory are common clinical characteristics of mental retardation (MR). However, the association of *TUSC3* genetic polymorphisms with MR remains unknown. A total of 456 DNA samples including 174 nuclear families containing MR were collected in the Qinba mountain area of China. The genotypes of eight tag single nucleotide polymorphisms of *TUSC3* were evaluated with traditional genetic methods. Family-based association tests, transmission disequilibrium tests (TDTs), and

haplotype relative risk (HRR) analyses were performed to investigate the association between genetic variants of the *TUSC3* gene and MR. The genetic polymorphisms rs10093881, rs6530893, and rs6994908 were associated with MR (all P values <0.05) based upon the results of single-site TDT and HRR analyses. The haplotype block consisting of rs6530893 and rs6994908, harboring the sixth exon of *TUSC3*, was also associated with MR (all P values <0.05). This study demonstrated an association between genetic polymorphisms of the *TUSC3* gene and MR in the Qinba mountain area, the sixth exon of which might contribute to the risk of MR. However, further studies are needed on the causal mechanisms in this association.

Key words: Tumor suppressor candidate 3; Mental retardation; Nuclear family; Single nucleotide polymorphisms