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COL5A1 rs12722 polymorphism is not associated with passive muscle stiffness and sports-related muscle injury in Japanese athletes

Eri Miyamoto-Mikami^{1,2}, Naokazu Miyamoto³, Hiroshi Kumagai^{1,4}, Kosuke Hirata^{4,5}, Naoki Kikuchi⁶, Hirofumi Zempo^{1,7}, Noriko Kimura⁸, Nobuhiro Kamiya⁹, Hiroaki Kanehisa¹⁰, Hisashi Naito^{1,2,3} and Noriyuki Fuku^{1,2,3}

¹Institute of Health and Sports Science & Medicine, Juntendo University. ²Juntendo Advanced Research Institute for Health Science, Juntendo University. ³Graduate School of Health and Sports Science, Juntendo University.

⁴Research Fellow of Japanese Society for the Promotion of Science. ⁵Graduate School of Engineering and Science, Shibaura Institute of Technology. ⁶Department of Training Science, Nippon Sport Science University.

⁷Faculty of Health and Nutrition, Tokyo Seiei College. ⁸Graduate School of Sport and Exercise Sciences, Osaka University of Health and Sport Sciences. ⁹Faculty of Budo and Sport Studies, Tenri University.

¹⁰Faculty of Sport and Health Science, Ritsumeikan University.

Abstract

Background: Poor joint flexibility has been repeatedly proposed as a risk factor for muscle injury. The C-to-T polymorphism (rs12722) in the 3' untranslated region of the collagen type V $\alpha 1$ chain gene (COL5A1) is reportedly associated with joint flexibility. Flexibility of a normal joint is largely determined by passive muscle stiffness, which is influenced by intramuscular collagenous connective tissues including type V collagen. The present study aimed to test the hypothesis that the COL5A1 rs12722 polymorphism influences joint flexibility via passive muscle stiffness, and is accordingly associated with the incidence of muscle injury.

Methods: In Study 1, we examined whether the rs12722 polymorphism is associated with joint flexibility and passive muscle stiffness in 363 healthy young adults. Joint flexibility was evaluated by passive straight-leg-raise and sit-and-reach tests, and passive muscle stiffness was measured using ultrasound shear wave elastography. In Study 2, the association of the rs12722 polymorphism with sports-related muscle injury was assessed in 1559 Japanese athletes. Muscle injury history and severity were assessed by a questionnaire. In both Study 1 and Study 2, the rs12722 C-to-T polymorphism in the COL5A1 was determined using the TaqMan SNP Genotyping Assay.

Results: Study 1 revealed that the rs12722 polymorphism had no significant effect on range of motion in passive straight-leg-raise and sit-and-reach tests. Furthermore, there was no significant difference in passive muscle stiffness of the hamstring among the rs12722 genotypes. In Study 2, rs12722 genotype frequencies did not differ between the muscle injury and no muscle injury groups. Moreover, no association was observed between rs12722 polymorphism and severity of muscle injury.

Conclusions: The present study does not support the view that COL5A1 rs12722 polymorphism has a role as a risk factor for sports-related muscle injury, or that it is a determinant for passive muscle stiffness in a Japanese population.