Association between matrix metalloproteinase-3 polymorphism and anterior cruciate ligament ruptures

S. Malila1,2, P. Yuktanandana3, S. Saowaprut1, W. Jiamjarasrangsi4 and S. Honsawek1

1Department of Biochemistry, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand
2Graduate Program in Medical Sciences, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand
3Department of Orthopaedics, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand
4Institute of Orthopaedics, Lerdsin Hospital, Bangkok, Thailand
5Department of Preventive and Social Medicine, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand

Corresponding author: S. Honsawek
E-mail: Sittisak.H@chula.ac.th

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ABSTRACT. Anterior cruciate ligament (ACL) ruptures are considered to be the most severe joint injury in sports. However, the precise etiologies of ACL injuries are not fully understood. Recently, the gene encoding the matrix metalloproteinase-3 (MMP-3, stromelysin-1) was shown to be associated with anterior cruciate ligament ruptures. The 5A/6A polymorphism in the promoter of the MMP-3 gene affects the regulation of MMP-3 gene expression. We examined the association between polymorphism within -1612 of the MMP-3 gene and ACL rupture in an independent population. Eighty-six participants between 20 and 40 years of age with surgically diagnosed ACL ruptures and 100 healthy controls between 18 and 28 years of age without history of
ligament or tendon injuries were recruited for the study. All participants were genotyped for the MMP-3 polymorphism (-1612 5A/6A). Statistical analyses of genotype frequencies between patients and healthy controls were performed by the chi-square test. A significant difference was found between ACL rupture subgroups in terms of genotype association (5A+ (5A/5A, 5A/6A): 37.5% in contact sports vs 20% in non-contact sports; P = 0.02). In allelic association, there were significant differences (6A: 81.2% in contact sports vs 89.1% in non-contact sports, 5A: 18.8% in contact sports vs 10.9% in non-contact sports, P = 0.01). The 5A+ genotype of MMP-3 was represented in ACL ruptures in contact sport participants. We propose that this sequence variant is a specific genetic element that should be included in a multifactorial model to understand the etiologies and risk factors for ACL rupture.

**Key words:** Anterior cruciate ligament ruptures; Matrix metalloproteinase-3; Single nucleotide polymorphism