

ESR1 rs2234693 Polymorphism Is Associated with Muscle Injury and Muscle Stiffness.

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Abstract

Purpose: Muscle injury is the most common sports injury. Muscle stiffness, a risk factor for muscle injury, is lower in females than in males, implying that sex-related genetic polymorphisms influence muscle injury associated with muscle stiffness. The present study aimed to clarify the associations between two genetic polymorphisms (rs2234693 and rs9340799) in the estrogen receptor 1 gene (ESR1) and muscle injury or muscle stiffness.

Methods: In study 1, a questionnaire was used to assess the muscle injury history of 1311 Japanese top-level athletes. In study 2, stiffness of the hamstring muscles was assessed using ultrasound shear wave elastography in 261 physically active young adults. In both studies, rs2234693 C/T and rs9340799 G/A polymorphisms in the ESR1 were analyzed using the TaqMan SNP Genotyping Assay.

Results: In study 1, genotype frequencies for ESR1 rs2234693 C/T were significantly different between the injured and noninjured groups in a C-allele dominant (CC + CT vs TT: odds ratio, 0.62; 95% confidence interval, 0.43–0.91) and additive (CC vs CT vs TT: odds ratio, 0.70; 95% confidence interval, 0.53–0.91) model in all athletes. In study 2, hamstring muscle stiffness was lower in subjects with the CC + CT genotype than in those with the TT genotype; a significant linear trend (CC G CT G TT) was found ($r = 0.135$, $P = 0.029$). In contrast, no associations were observed between ESR1 rs9340799 G/A and muscle injury or stiffness.

Conclusions: Our results suggest that the ESR1 rs2234693 C allele, in contrast to the T allele, provides protection against muscle injury by lowering muscle stiffness.