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82 THE *COL5A1* GENE AND RISK OF ACHILLES TENDON PATHOLOGY IN A BRITISH COHORT

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Background Achilles tendon pathologies (ATPs) such as Achilles tendinopathy and Achilles tendon ruptures have been identified as debilitating conditions resulting from either acute or repetitive overuse loading mechanisms. ATP is a multifactorial condition for which several genetic risk factors have been identified. Notably, the *COL5A1* rs12722 genetic variant has been associated with risk of Achilles tendinopathy in South African and Australian populations.[1,2] Further, the *COL5A1* rs71746744, rs16399 and rs1134170 variants were also associated with Achilles tendinopathy within combined South African and Australian populations.[3] The rs71746744 within the 3'-UTR of the *COL5A1* gene has been shown to be functional.[4]

Objective The objective if this study was to investigate if the *COL5A1* rs12722 and rs71746744 variants are associated with ATP in a British (UK) cohort.

Methods One hundred and thirty six (52 females and 84 males) participants diagnosed with Achilles tendon pathology (ATP group) and 131 (49 females and 82 males) asymptomatic healthy controls (CON group), were recruited for this case-control genetic association study. ATP, which includes non-insertional and insertional Achilles tendinopathy, as well as Achilles tendon rupture were diagnosed using MRI scans and ultra-sound imaging. This study was approved by the Research Ethics committees of the University of Northampton and the University of Cape Town and all participants gave written informed consent. SNP genotyping was performed using a fluorescence-based custom-made TaqMan (rs71746744) and restriction fragment length polymorphism assays (rs12722) from DNA extracted from saliva samples. The significance of all statistical testing was accepted at $p < 0.05$.

Results No significant genotype frequency distributions were detected in the UK cohort between the CON and ATP groups for the rs12722 ($p = 0.658$) and rs71746744 ($p = 0.319$) variants. However, when only the male participants were investigated, the TT genotype of the rs12722 ($p = 0.015$) and the INS/INS ($p = 0.031$) of the rs71746744 variant was significantly over-represented within the ATP groups.

Conclusion This study showed that both *COL5A*1 rs12722 and rs71746744 variants were significantly associated with risk of ATP within males. It is unknown why this association was not found in the female participants investigated.

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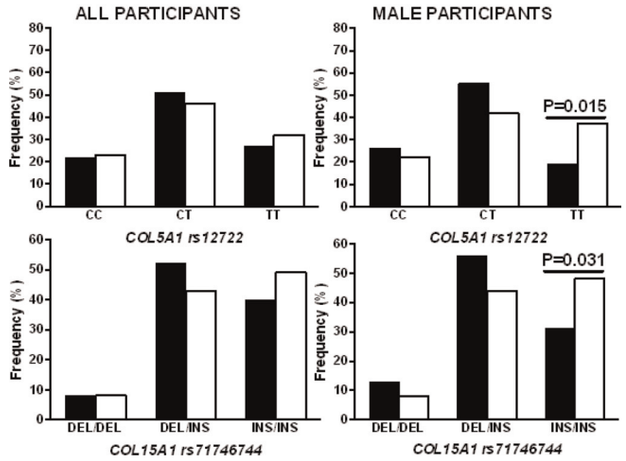

Abstract 82 Figure 1

83 INVESTIGATION OF ANGIOGENESIS ASSOCIATED GENES WITH ACHILLES TENDINOPATHY

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Introduction Genetic factors have been implicated with risk of Achilles tendinopathy. Angiogenesis plays an important role in extracellular matrix (ECM) remodelling following mechanical loading. It is proposed that dysregulation of this process may alter the mechanical properties of the tissue thereby increasing the risk of injury. Furthermore, increased levels of angiogenic cytokines and growth factors have been reported in injured Achilles tendons [Pufe, 2001] and after cyclic stretching of tendon fibroblasts [Petersen, 2004]. Vascular endothelial growth factor (VEGF) is a potent pro-angiogenic factor and the biological effects of VEGF are mediated through its receptor kinase insert-domain receptor (KDR). Recently, polymorphisms within the *VEGFA* and *KDR* genes were found to be associated with risk of anterior cruciate ligament (ACL) ruptures [Rahim, 2014]. Thus, the aim of this study was to investigate if genes encoding proteins involved in angiogenesis-associated signalling are associated with risk of Achilles tendon injuries.

Methods A genetic-association study was conducted on 120 control participants (CON), 91 participants with Achilles tendinopathy (TEN) and 44 participants with partial or complete ruptures of the Achilles tendon (RUP). All participants were genotyped for the functional *VEGFA* rs699947, *VEGFA* rs1570360, *VEGFA* rs2010963, *KDR* rs2071559 and *KDR* rs1870377 polymorphisms. Haplotypes were also inferred for *VEGFA* and *KDR*. Statistical analyses were conducted to determine any significant differences ($p < 0.05$) between the groups (CON vs. TEN and CON vs. RUP).

Results No independent significant differences were observed in the genotype frequency distributions of any of the polymorphisms between the CON vs. TEN groups. However, there was a trend towards significance ($p = 0.053$) in the genotype frequencies for the *VEGFA* rs699947 polymorphism when comparing

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Notes