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LETTER TO THE EDITOR

Bilateral consecutive rupture of the quadriceps tendon in a man with BstUI polymorphism of the COL5A1 gene

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Dear Editors,

I write to you regarding the paper by Longo et al. [1] "Bilateral consecutive rupture of the quadriceps tendon in a man with BstUI polymorphism of the COL5A1 gene."

The impression given by the authors is that they have identified a possible association between the presence of a variant in the *COL5A1* gene and rupture of the quadriceps tendon. However, the data presented in the paper do not support such a proposition. The authors have investigated a single individual and demonstrated that he harbours a specific variant (rs12722; NM_000093.3:c.*267C>T) in *COL5A1*. This variant lies in the 3′ untranslated region of the mRNA encoded by *COL5A1*, and there has been no demonstrated effect on the expression of the gene.

This single-nucleotide polymorphism (SNP) is frequent in the majority of populations that have been studied. Indeed, the T allele that results in loss of the *Bst*UI site is the more common allele in cohorts of European ancestry which are recorded in the dbSNP database. Hence, it is not surprising to find that the individual presented in this study harbours the variant. Given the variant frequency, there will be many individuals who harbour this variant and never go on to develop rupture of the quadriceps tendon.

The authors cite a previous study [2] that demonstrated an association between the presence of this variant and Achilles tendon pathology. In that study, the frequency of the variant was compared in "One-hundred and eleven Caucasian subjects diagnosed with ATP and 129 Caucasian control (CON) subjects…" The authors were cautious in their conclusions and stated: "The association of the *COL5A1* gene with Achilles tendon injuries however does not prove that type V collagen, an important component of tendons, is directly involved in the development of this pathology. Perhaps other closely linked genes are involved." A comparable study for bilateral consecutive rupture of the quadriceps tendon would require recruitment of a similar number of affected individuals and controls. Ideally, even larger numbers should be studied, but the rarity of the condition probably precludes this.

In addition, figures 2 and 3 do not include images from control individuals.

It is methodologically flawed to study a single individual, as the authors have done, and attempt to draw any conclusions with respect to the contribution of this SNP to the rupture of the quadriceps tendon.

References

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2. Mokone GG, Schwellnus MP, Noakes TD, Collins M (2006) The COL5A1 gene and Achilles tendon pathology. Scand J Med Sci Sports 16:19–26

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