No association between tendon-related genes and performance in elite European Caucasian marathon runners.

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Tendons adapt to load under normal physiological conditions, however, under extreme loading conditions, such as those experienced by elite endurance athletes, incomplete adaptation may occur and cause injury. The prevalence of tendinopathies in elite endurance athletes is approximately 50%, thus variability exists in an athlete's tolerance to extreme loading. A number of intrinsic and extrinsic factors contribute to modulating injury risk, some of which are modifiable and others, such as genetic variants, are non-modifiable. It was hypothesized that elite marathon runners would possess a genotype associated with enhanced tendon function, and thus protective against tendinopathy. Here, we compared the genotype frequencies of six genetic variants (*COL1A1* rs1800012, *VEGFA* rs699947, *TIMP2* rs4789932, *MMP3* rs591058, *MMP3* rs650108, *MMP3* rs679620), previously associated with tendinopathy, in elite (men <2 h 30 min, *n* = 109, women <3 h 00 min, *n* = 99) and sub-elite (men 2 h 30 min-2 h 45 min, *n* = 189; women 3 h 00 min-3 h 15 min, *n* = 71) marathon runners with those of a non-athletic control group (*n* = 564). Genotype associations with marathon personal best time in the athlete group were also investigated. All participants provided either a whole blood, saliva or buccal cell sample, from which DNA was isolated, and genotyped for all six variants using real-time PCR. Genotype frequency differed between athletes and controls for *TIMP2* rs4789932 (TT = 17%, CT = 51%, CC = 32% vs. TT = 22%, CT = 42%, CC = 36%, respectively; $\chi^2 = 8.135$, $P = 0.017$) only. However, there was no clear difference in allele frequencies between groups for *TIMP2* rs4789932. *MMP3* rs650108 genotype frequency differed between female elite and sub-elite athletes ($\chi^2 = 11.913$, $P = 0.003$) only and, as hypothesized, it was the “risk” A-allele that was ~10% less frequent in the elite, than sub-elite athletes. Following combination of all genotype data into a total genotype score, no differences in score between athletes and controls were observed ($t = 2.93$, $P = 0.769$). Similarly, no associations between total genotype score and marathon personal best time in male and female runners were observed ($r \leq 0.066$, $P \geq 0.394$). The results suggest elite marathon runners do not possess a genotype protective against tendinopathy, at least for the tendon-related genetic variants we investigated.