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Risk factors for anterior cruciate ligament injuries: Report from a genetics study

Orthopaedics / Knee & Lower Leg / Epidemiology, Prevention & Diagnosis

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Background
Several genetic factors that could predispose to an Anterior Cruciate Ligament (ACL) injuries have recently been reported. The COL14A1 gene has been mapped to chromosome 8q23 and encodes the alpha chain of type XIV collagen, a member of the FACIT (fibril-associated collagens with interrupted triple helices) collagen family. Type XIV collagen may be involved in fibrillogenesis and some studies suggest that it may modulate the cellular response of tissue to mechanical stress. Collagen XIV is one of the main structural components of Anterior Cruciate Ligament (ACL) collagen along with collagen types I, III–VI, XII and various proteoglycans and glycoproteins.

Objectives
Therefore, we hypothesized that specific polymorphisms within the COL14A1 gene could be associated with ACL rupture in physically active population, because it is possible that greater tissue damage following exercise leads to ligament injuries such as ACL rupture. The aim of this study was therefore to test the association between the COL14A1 rs 4870723 and ACL rupture in physically active population in our Country.

Study Design & Methods
A case-control genetic association study was conducted on 69 physically active Italian participants (Levels 5-10 of the Tegner Activity Level Scale) of which 47 controls (n=21 females and n=26 males) and 20 individuals with surgically-diagnosed ACL ruptures (n=10 females and n=10 males). All the participants reported non-contact mechanism of injury. Genomic DNA was extracted from buccal swab using a standard protocol. COL14A1 rs4870723 genotype distributions were compared between cases and controls using "Statistics V. 8.0" and "GraphpadInStat 3.0" programs. The level of statistical accuracy was set up for $p<0,05$.

Results
The genotypes distribution related to the COL14A1 rs4870723 polymorphism were in Hardy-Weinberg equilibrium. Considering the whole sample, no significant differences have been found in COL14A1 rs4870723 genotypes distributions between cases and controls

(p=0.17). Conversely, gender-associated differences showed that in females the AA genotype frequency was higher in cases then in controls (cases 0.90 vs controls 0.38, p=0.006) andnone of the cases showed the CC genotype (cases=0.0 vs controls=0.19, p=0.003).

Conclusions

In the present pilot study, we found for the first time an association between COL14A1 rs4870723 polymorphism and ACL rupture in female physically active population. The carriers of the AA genotypes were higher in ACL group respect to the controls. These results could suggest a protective effect of the CC genotype in developing ACL rupture among physically active women. In conclusion, our findings suggest that the COL14A1 rs4870723 polymorphism is one of the genetic variants that could influence the susceptibility to developing ACL injury among physically active women. To confirm our findings, the same research should be replicated in a larger number of subjects and in other cohorts from different countries. The integration of the individual genetic background with training, diet and endocrinal aspects could be the basis to develop an "ad personam" multidisciplinary strategy to prevent ACL injuries.