An Investigation into Genetic Associations with Musculoskeletal Soft Tissue Injuries in Elite Rugby – Preliminary Findings

Brazier, J.J.,[1] Antrobus, M.R., [1] Heffernan, S.M, [1] Stebbings, G.K, [1] Day, S.H., [1] Hennis, P.J., [1] Erskine, R.M., [2] Kilduff, L.P. [3] and Williams, A.G. [1]

[1]Sports Genomics Laboratory, Manchester Metropolitan University, Crewe, UK. [2] Research Institute for Sport & Exercise Sciences, Liverpool John Moores University, Liverpool, UK. [3] College of Engineering, Swansea University, Swansea, UK.

INTRODUCTION

Rugby union has been reported to have one of the highest incidences of match injuries within professional sport (Brooks et al., 2005). Muscle/tendon and joint/ligament injuries are the two most frequent injury groups, with the lower limb having the highest incidence of injury (Williams et al., 2013).

One of the most severe joint/ligament injuries in rugby Union is Anterior Cruciate Ligament (ACL) ruptures with 110 days absence per 1000 player hours among forward players and 104 days absence among the backs (Brooks et al., 2005). The most common muscle/tendon injury amongst the forward players is Achilles Tendon Injury (AT), causing 81 days absence per 1000 player hours (Brooks et al., 2005). Several risk factors have been implicated in the causation of ligament and tendon injuries with genetic polymorphisms proposed as a non-modifiable risk factor (Riley, 2004; September et al., 2012).

Several genes have been associated with ligament and tendon injuries, such as *MMP3, COL1A1, COL5A1, TNC, GDF5, TIMP2,* and *VEGFA. MMP3* is one of the Matrix Metalloproteinases family of genes and its protein product can catalytically degrade a number of components of the extracellular matrix including type II, IV, V, IX and X collagens (Raleigh et al., 2009). Sequence variation in *MMP3* has been associated with AT in a Caucasian cohort and ACL ruptures in contact sports in an Asian cohort (Collins et al., 2015).). Type I collagen makes up ~80% of the dry mass of ligaments and tendons and the α1 chain is encoded by *COL1A1*. The TT genotype has been significantly underrepresented in ACL injury groups (Collins et al., 2015).

To date, there have been several studies into genetic associations with musculoskeletal soft tissue injuries. However, there has been no research into gene associations with ligament and tendon injuries within elite rugby. Thus, the purpose of these studies is to establish whether elite rugby players are more or less susceptible to ligament and tendon injuries than the general population and whether that susceptibility to injury differs amongst playing positions.

METHODS

Participants

- Elite Caucasian male rugby athletes (*n*=334; 1.85 ± 0.07 m, mass 101 ± 14.1 kg, age 27.5 ± 7 yr).
- 74% British, 12% South African, 9% Irish, and 5% of other nationalities.

Procedures

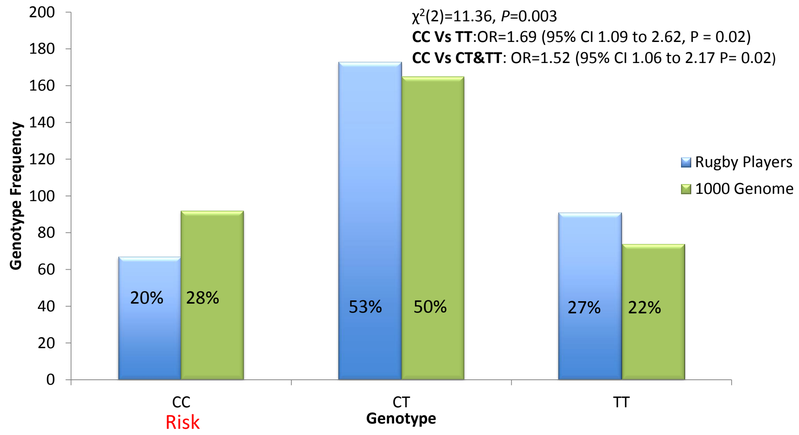
- DNA isolated from blood for all samples.
- Genotyping was performed using real-time PCR and allele-specific fluorescent probes.
- 1000 Genome participants (European Caucasians) used as controls for genetic variation.

Statistical Analysis

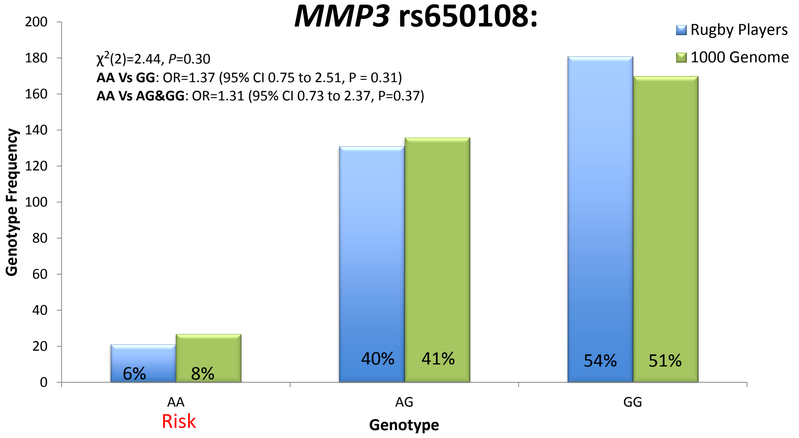
- Genotype *f* were compared between groups using χ2 and odds ratio (OR) statistics.
- All genotyped data were in Hardy-Weinberg equilibrium.

PRELIMINARY RESULTS

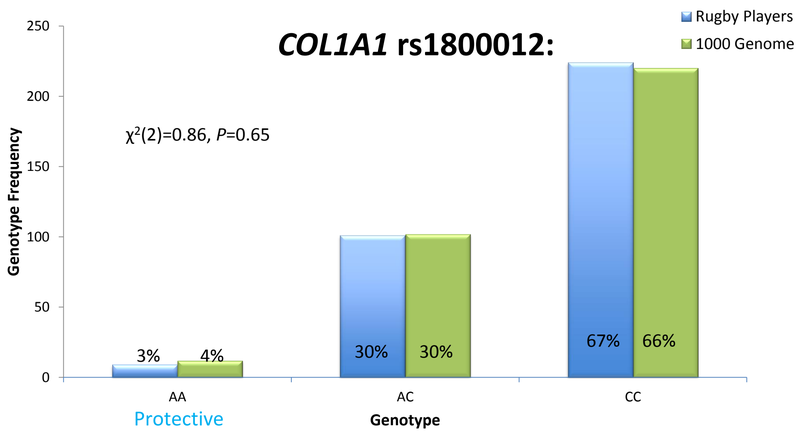
MMP3 rs679620:



MMP3 rs650108:



COL1A1 rs1800012:



CONCLUSION/ FUTURE DIRECTION OF STUDY

The preliminary results suggest that for the MMP3 rs679620 polymorphism, elite rugby players have a lower frequency of alleles previously associated with increased risk of ligament and tendon injuries compared to controls. Players carrying the risk alleles might have been more susceptible to such injuries and thus failed to reach elite level. As yet no significant associations have been found for MMP rs650108 and COL1A1 rs1800082 polymorphisms, however this may change with the increasing of the sample size to the target of ~800 players, as well as analysing the control samples rather than utilising 1000 Genome samples.

Given these encouraging initial findings, the MMP3 rs679620 genetic variant together with additional polymorphisms yet to be identified, may provide further information on the aetiology of ligament and tendon injuries and could potentially aid in a more personalised prehabilitation and injury management protocol of players. Thus, creating more robust strength and conditioning practises within elite rugby.

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