Description of PhD Thesis for the Tertiary Education Scholarship Scheme (TESS) of Malta

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Thesis title: The Genetic Contribution to Muscular Strength and Power in Elite Rugby Athletes

A thesis submitted in partial fulfilment of the requirements of
Manchester Metropolitan University
for the degree of Doctor of Philosophy

The research work disclosed in the thesis is funded by the Tertiary Education Scholarships Scheme of Malta

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Muscular strength and power are key determinants of physical performance in the general population and in elite athletes. Accordingly, various measures of strength/power have been used to identify the onset of sarcopenia in middle and older age populations, for example (Khanal et al., 2020). On the other hand, established measures of strength/power have been used to assess and monitor athletes, inform training interventions and training prescription, and discriminate athletes between levels of competition (Brazier et al., 2020). Muscular strength/power are multifactorial phenotypes that show a Gaussian (continuous) variation in the population, which depends on environmental and genetic factors . The main factors which contribute to strength/power (e.g. muscle mass, neuromuscular activation) and intermediate contributing factors (e.g. number of muscle fibres, fibre cross-sectional area, number of motor neurons), all demonstrate a continuous variation in the population, and are also accepted to be influenced by both the environment and genetic variation (Wackerhage, 2014). Multiple genetic variants (polygenic in nature) are thought to influence strength/power, exerting a combined effect that in addition to environmental factors (mainly physical activity and nutrition) produce the inter-individual variation in strength/power observed in diverse populations (Hughes et al., 2011). To date, ~62 genetic variants have been found to influence strength/power phenotypes however, how these genetic variants combine and/or interact with the environment to affect strength/power in elite athletic populations is unclear. Furthermore, there is a scarcity of research that have considered the exploration of genotype-phenotype associations in elite athletes – in particular, whether specific genetic variants exert an influence on the athletes' strength/power, or on their performance during competition.

As part of the ongoing RugbyGene project (Athlome project Consortium: www.athlomeprojectconsortium.org) the overall aim of the thesis was to characterise muscular strength/power and the genetic characteristics of elite rugby athletes using established measures of strength/power and 10 genetic single nucleotide polymorphisms (SNPs); and investigate whether the 10 variants, individually and collectively, were associated with measures of strength/power and/or in-game performance variables. 567 elite rugby athletes and 1138 non-athletes were genotyped for 10 SNPs found in nine genes (e.g. angiotensin-converting enzyme, adenine monophosphate deaminase-1, thyrotropin releasing hormone receptor) and established measures of strength/power data (using the isometric mid-thigh pull (IMTP) and countermovement jump (CMJ), and the 'gold standard' method of data collection that involved a sophisticated force-platform (Owen et al., 2014)) were collected for 263 athletes and 14 non-athletes. Differences in CMJ and IMTP variables were observed between athletes and non-athletes and between the two major playing positions (forwards and backs), whilst SNPs within some of the genes investigated were found to be associated with athlete status and playing position. In addition, SNPs within some of the genes were associated with either the IMTP or CMJ, or with both. In-game performance data (e.g. involvement in tackling, frequency of carries, ability to gain territory) were acquired for 291 athletes during 8 seasons (2012–13 to 2019–20) of rugby union competition in the highest professional competitive leagues in the UK, and associations were observed between some of the investigated SNPs and in-game variables. Interestingly, the polygenic influence of seven of the SNPs expressed as a Total Genotype Score (Williams and Folland, 2008) was associated with some of the investigated in-

game performance variables. Furthermore, forwards and backs possessing the most favourable alleles were involved more in some of the in-game performance variables that reflect superior strength, compared to those athletes possessing less favourable alleles.

Most of the results in this thesis identify novel genetic associations with strength/power in an elite rugby context. In addition, there appears to be a genotype-dependent influence on strength/power phenotypes and competitive performance within elite rugby athletes that varies with positional roles. Further research is needed to replicate the associations observed in comparable and larger athletic cohorts. Nonetheless, the work in this thesis has added to our understanding of the genetic contribution to strength/power and competitive performance of elite rugby athletes, which when combined with physiological data, may have implications for management and performance enhancement of elite athletes in future

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