

## Genetic differences in elite rugby players and the general population

We have known for many years that genetic variation is important in understanding any physiological trait and contributes substantially to the way people develop. Height for example is estimated as being ~80% determined by genetic variation with the other ~20% determined by environmental influences, such as infant nutritional status. Most traits however, are not so highly, but still considerably, genetically determined – body mass ~60%,  $VO_{2peak}$  ~50%, leg muscle strength ~60% and the likelihood of being an elite athlete (athlete status) ~70%. Nonetheless understanding this substantial component of physical performance, in addition to environmental factors, is crucial. As such, elite athletes represent the highest achievable level of physical performance and looking at the genetic influences on elite athletes can give us a better understanding of the highest limits of human physical performance.

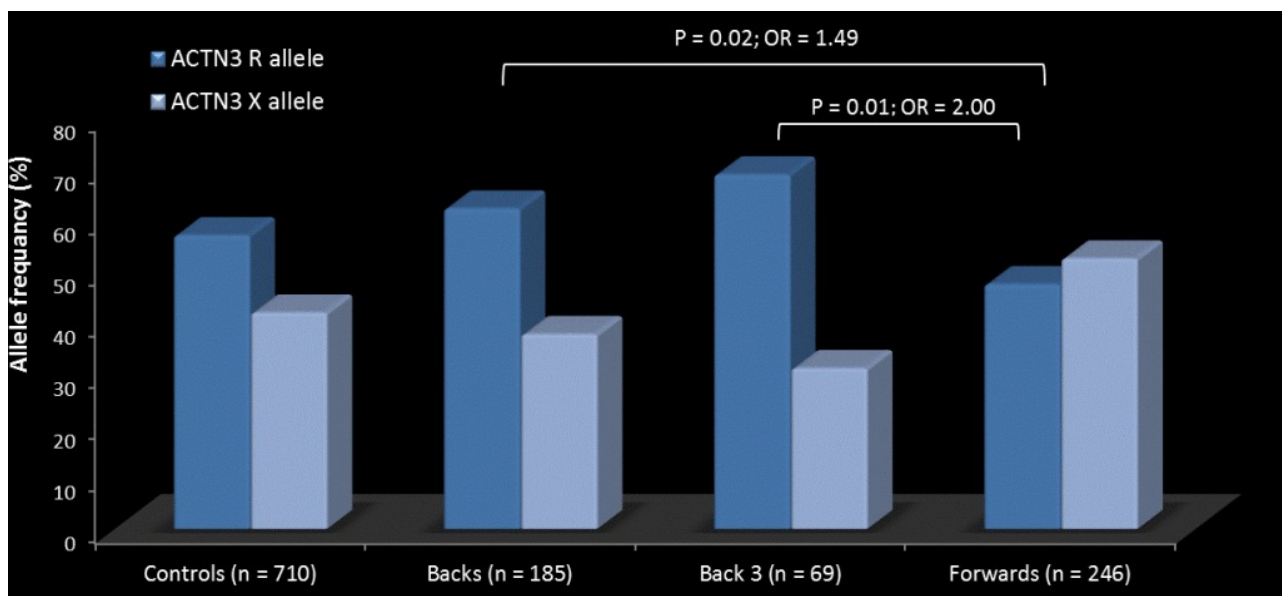


Fig. 1. ACTN3 allele frequencies. Allele frequencies of controls, rugby union athletes divided into playing sub-group (forwards and backs), back 3 group (wingers and fullbacks).

Elite rugby athletes are an ideal group of individuals to study the genetic variation of various performance traits (athlete status, muscle strength,  $VO_{2peak}$ , etc.), because of the positional diversity across the athletes. There are 15 playing positions that can be grouped according the physical demands when playing; Forwards and Backs, which can be further sub-divided; Front 5, Back row, Halves, Centres and Back 3. For example, in rugby union, the mass of athletes playing on the same field at any one time can vary from 140 kg (Front 5) to 75 kg (Halves) individuals, with similar variation evident for other physiological variables (muscle mass, power, speed, etc.).

Because of these positional differences, associated physiological traits are likely to be evidenced by genetic variation - in addition to differences from the general population. As such, the first steps when exploring the genetic variation in elite rugby athletes is to investigate which gene variants are different between athletes and the general population and then, specifically in the case of team sports, examine the genetic differences across playing position. These first steps are what we have done here.

We analysed DNA from 507 elite rugby athletes (~45% international representatives) and 710 individuals from the general population and looked at the differences in genetic variation of the *ACE* I/D and *ACTN3* R577X gene variants. Briefly, when some DNA letters (ATGC...) of the *ACE* gene are deleted (D allele) it increases the activity of its protein product, compared to when those letters are inserted (I allele). The 'I allele' version of the gene has been convincingly associated with certain aspects of endurance physiology. For *ACTN3* R577X, when the gene sequence is read in full (R allele), the  $\alpha$ -actinin-3 protein that is present only in fast-twitch muscle fibres is produced and functions normally. When the reading of the gene sequence is stopped prematurely (X allele) because of a mis-spelling in the DNA code, the protein is not produced. The *ACTN3* 'R allele' has been associated with speed/power physical traits and athletes, with the X allele associated with fatigue resistance, endurance traits and endurance athletes.

We found that elite rugby union players selected as backs possessed significantly more *ACTN3* R alleles than players selected as forwards and compared to the general population (Fig. 1.). Furthermore, players that possessed the R allele had 1.49 greater odds of becoming backs than forwards and more specifically, R allele players had twice the odds of becoming a Back 3 player (generally the fastest players) than a forward (OR = 2.00; Fig. 1.). Additionally, the forwards possessed a greater amount of individuals carrying the X allele than backs. We found no differences regarding the *ACE* gene. The *ACTN3* gene variant results suggest that elite rugby union forwards possess an inherited fatigue resistance (X allele), while inherited sprint ability is more prevalent in backs, especially wings and full backs (Back 3 group). This study was the first step towards understanding the genetic building blocks associated with physiological traits underpinning elite rugby union athlete success.

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## Publication

[Association of \*ACTN3\* R577X but not \*ACE\* I/D gene variants with elite rugby union player status and playing position.](#)

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*Physiol Genomics. 2016 Mar*