

Genetics and Sport Injuries: ACTN3 Gene as a Possible Marker in Muscle Skeletal Injury Susceptibility

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Introduction

It is well known that the increasing overload imposed by physical training lead the athletes to improve athletic performance. On the other hand, these high-intensity training sessions, followed by factors such as high impact loads, also increase the injuries risk in different sites such as in the bones [1], muscle-tendon unit [2], and joints [3]. Previous findings have suggested that a high level of strength and/or power production ability could be considered one of the main factors related to prevention of the skeletal muscle injuries in sports [4,5]. In this way, it is believed that the enhanced muscle strength ability acts as a protector factor against sports training-induced injuries [6,7].

There is a widely number of studies reporting that muscle strength might be influenced by genetic factors [8-10]. Among the main genetic factors, a polymorphism in the ACTN3 gene (R577X) has been investigated with emphasis on its relationship with muscle strength ability [11]. It has been demonstrated that individuals with the RR genotype of the ACTN3 gene have greater muscle strength ability in different tasks [9,12]. In addition, Vincent et al. [13] and Pimenta et al. [14] observed that individuals with this genotype are less susceptible to muscle damage, which leads to an acute decrement in muscle strength [15]. Based on these evidences, it is attractive to suspect that the ACTN3 gene might be a novel marker in skeletal muscle injuries. Therefore, the objective of this short review was to provide the scientific information about the relationship between the ACTN3 gene and muscle strength and about the possible association of this gene with muscle injuries susceptibility.

ACTN3 and Muscle Strength

The ACTN3 gene encodes the α -actinin-3, a structural protein present in the Z line of the type II skeletal muscle fibers that plays an important role on the force transmission throughout the adjacent sarcomeres [16]. A single nucleotide polymorphism in the ACTN3 gene consists in a C>T nucleotide change, conversion from arginine to a premature stop codon in the 577 residue (R577X) and consequently in the expression of a non-functional form of the α -actinin-3. Thereby, the XX genotype results in a complete absence of the α -actinin-3 in the muscle tissue, while RX and RR genotypes result in a normal protein expression [17].

Previous studies demonstrated that the RR genotype is more frequent among strength/power athletes [9,18] and that this genotype can also influence the strength ability in normal individuals [12]. These authors proposed that individuals carrying the RR genotype have more pronounced skeletal muscle strength, probably due α -actinin-3 presence. Interestingly, some studies also demonstrated that RR individuals are less susceptible to exercise-induced muscle damage [13,14]. It was showed in animal model that the α -actinin-3 plays a protective effect against the muscle damage after an acute exercise bout [19]. In humans, Vincent et al. [13] observed that RR individuals presented a lower serum creatine kinase activity (a marker of muscle damage) and a lower pain score compared to XX individuals after a maximal eccentric knee extension exercise. Taken together, these

findings suggest that individuals with RR genotype of the ACTN3 gene polymorphism might have a greater ability to produce force and a lesser susceptibility to mechanical stress in the skeletal muscle than individuals with XX genotype.

Injuries and Muscle Strength

It is common to affirm that the muscle strength act as a protective mechanism against injuries [20]. For instance, Unyó et al. [6] observed that the imbalance between dorsal and palmar flexors muscles strength might be related to lateral epicondylitis. Additionally, analyzing athletes, Schnackenburg et al. [7] observed a lower (-18.3%) knee extension strength in female runners presenting tibial stress fractures. Moreover, strength gain seems to reduce the injuries incidence and to be a good way to recover injured individuals. Jayaraman et al. [21] observed that after four weeks of strength training, incomplete spinal cord injury patients improved significantly the distance walked in the 6-Minute Walk Test, as well as the isokinetic.

Aiming for Future Studies

The high incidence of the skeletal muscle injuries can be considered a motivational factor to the development of new analysis methods able to prevent injuries in athletes and physical activity practitioners. An important characteristic that should be deeper studied is the muscle strength, which seems to be influenced by genetic factors. One of the best candidate genes to influence muscle strength is the ACTN3 gene. Current new evidences suggest that RR genotype may be related to a higher strength production capacity as well as lower muscle damage susceptibility. Therefore, the ACTN3 gene seems to be a highlighted candidate for future investigations aiming to analyze genetic factors associated to the physical training-related injuries. Future studies should be performed in a multicentric design, with athletes and practitioners of different sports and competitive levels, in order to determine the impact of genetics components on the muscle skeletal injuries susceptibility, including the ACTN3 R577X genotypes.

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Received January 07, 2014; Accepted February 20, 2014; Published February 22, 2014

Citation: Pasqua LA, Lima-Silva AE, Bertuzzi R (2014) Genetics and Sport Injuries: ACTN3 Gene as a Possible Marker in Muscle Skeletal Injury Susceptibility. *J Nov Physiother* 4: 193. doi:10.4172/2165-7025.1000193

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