

# Possibilities of gene assisted regulations in elite athlete's landscape of injury prevention and performances

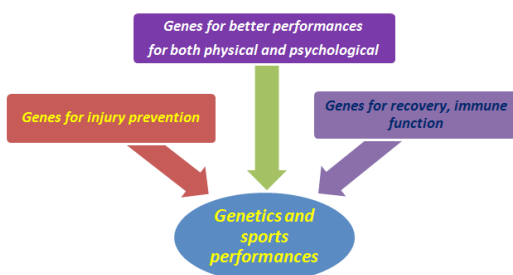
## Commentary

The frontiers of Sports science with respect to the elite sports performances, training and injury management are experiencing unprecedented growth leading to unforeseen interactive discussions and consequently to the best possible practices in sports training for elite performances. Exercise and sports genetics is fast developing along with the science of exercise endocrinology and other frontiers. Emergence of novel ideas with interactive and interdisciplinary approaches in sports sciences are bringing out hitherto unseen scientific sports training concepts which are helping for surging in elite sports performances. It is unequivocal that the elite performances are the result of genetics, epigenetics and appropriate scientific nurture. Many research studies of sports genetics emphatically suggest that genetics of individuals may show significant impact on the performance levels of sportspersons. This renaissance in sports sciences with interdisciplinary approach has led to deeper understanding of various scientific concepts which help to identify the potential top level sportspersons, training them and nurturing them into world class athletes.<sup>1-5</sup>

## Domains of genetic predisposition for sports performances

Among the important possibilities in which sports genetics and sports endocrinology might contribute, identifying the suitability of an individual to a particular sporting event, appropriate training possibilities, recovery from training rigors, injury prevention are some of the quintessential areas of sports training. As already suggested, plethora of research studies have already been rhetoric in identifying several genes that are identified to better physical performances.<sup>6,7</sup> Apart from being genetic predisposition to better physical performances, another significant essentiality is being genetically supported less injury proneness. Some of the sports genetic studies have identified such genetic predisposition of being better muscular, tendinous, bone and cartilaginous status thereby less susceptibility to injury, even when involved in high intensity sports trainings. There are several studies which revealed that even the post training recovery differences were also significantly ascribed to genetic variations among individuals. Hence, it would be ideal to examine the genetic predisposition of individuals for elite sports performances, in three broad areas essentially.<sup>8</sup>

## Genetic disposition for elite sports performances



Volume 3 Issue 1 - 2019

Rajasekhar Kali Venkata

Wellness and sports scientist, Director of Physical Education and sports, University of Hyderabad, India

**Correspondence:** Rajasekhar Kali Venkata, Wellness and sports scientist, Director of Physical Education and sports, University of Hyderabad, India, Tel +91 9949513504, Email jatiraj@yahoo.co.in

**Received:** December 27, 2018 | **Published:** January 07, 2019

However, the above three domains of genetics for sports performances shall be restricted to the physiological connotations of the individuals and shall not be related or extended to the other areas like anthropological, biomechanical connotations of individuals.<sup>9,10</sup> For example, morphological advantages such as slender foreleg could be an advantageous factor for distance running as seen in several Kenyan and Ethiopian elite runners. It may be untimely and inexpedient to ascribe this morphological character either to genetics or to epigenetics. Another challenging and difficult to establish domain is psychological aspects of sports performances like extreme pain tolerance, achievement motivation, controlled aggression etc for elite sports performances, though some sports genetic studies have established remote influences of genes in these areas also.<sup>11</sup>

## Genes for better performances

Sports genetic studies have established abundantly the relevance of certain genes and the higher order sports performances.<sup>12-14</sup> It is now indubitable that certain genetic predisposition makes certain individuals prone for higher order sports performances. Many candidate genes have been identified in this endeavour by the sports genetics experts. ACTN3 gene is one such very important gene that has been identified to link to the speed and power muscular performances.<sup>15</sup> Though this gene was initially identified as 'speed gene', the latest studies on sports genetics have confirmed that this gene has remarkable positive potential in the areas of post exercise recovery, positive modulation in terms of injury prevention etc also. A very common polymorphism of this gene appears to be R577X. R allele of the ACTN3 gene strongly associated with the speed and power properties of the muscle where as the XX genotype may be more favourable for the endurance type performances. Some studies indicated that the RR genotype of ACTN3 are more responsive to the resistance and power training when compared to the XX genotype of the ACTN3 gene,<sup>16</sup> reason may be the higher levels of bioavailability of testosterone among RR genotype. These variations were also observed in different ethnic groups, but there were no unequivocal evidences to support that XX genotype could be more favourable for endurance type performances. Many exercise genetics studies also confirmed that RR genotype of ACTN3 are more favourable in terms of injury prevention and post recovery of protein synthesis in muscle. It may be true that the RR genotype of this gene may be responsible

for less aggressive exercise wear and tear of muscle fibres causing less muscle damage during the high intensity exercise training.<sup>17</sup>

### Genes for injury prevention of muscles, tendons, ligaments etc

Several candidate genes are associated with the synthesis and regulation of collagen the main bio factor and component for the making of tendons, ligaments, cartilages etc. Even the organic part of the bone consists significant amounts of Type I collagen. Type I collagen is abundantly present in most of the elastic but very hard tissues like tendons,<sup>18</sup> ligaments and cartilages. COL1A gene with its several polymorphic presence in collagen fibres shows significant and differential effect on the strength and resilience aspects of the tendons, ligaments etc. COL1A1 and COL1A2 are mostly widely studied gene polymorphisms of type I collagen synthesis. Gene variants within COL1A1 shows differential impact on the structure and the strength of the collagen fibres. Studies identified that the COL1A1 G-T haplotype abundance in collagen fibres increase the tensile strength and resilience of the collagen thereby reducing the risk of injury in Ligaments and tendons. Though no much research is available with respect to the gene variants of COL2A1,<sup>19,20</sup> it is established clearly that this gene is responsible for the collagen synthesis in cartilages of joints and abundance and the substance quality could bring differential effects on the quality and strength of cartilages leading to differences in the injury proneness to the cartilages.<sup>21</sup>

### Genetic influence on immune variations and sports performances

Several genetic studies on human immune system clearly indicated about the differences in immune system through gene variation consequences.<sup>22</sup> Exercise immunologists identified several immune systems operate to control the infection levels of sports persons. Among many mechanisms of immune system, mucosal immunoglobulin system is one primary immune system that protects the sports persons from the infection during the exercise and post exercise.<sup>23-25</sup> Immunoglobulin A subclasses (IgA1 and IgA2) are two primary immunoglobulins that monitors and controls the immune function of mucosal membranes. Suppression of these immunoglobulins may cause for the reduction in the immune status of the mucosal membranes of lungs and could cause for reduction in lung capacities leading to disturbance in performances of elite sports persons. Oxidative stress genetics, redox reaction genetics are still underway in sports genetics perspective and needs further extensive research. Also, the research basing on the circadian rhythms and genetics need to be studied extensively.<sup>26,27</sup>

### Conclusion and recommendation

Plethora of studies on sports genetics and sports immunology indicate that several candidate genes are significantly responsible for higher order performances among sports persons and also consequent variability in terms of the injury prevention levels and immune protection levels. These three factors show emphatic effect on the elite sports performances by showing impact on the bio environment of metabolism of physical activity, tissue differentiation and endocrinal reactions. It is highly essential to further deepen the research perspectives of sports performance genetics taking into consideration of the interactive issues like tissue genetics, immune genetics.

### Acknowledgments

None.

### Conflicts of interest

The author declares there is no conflict of interest.

### References

- Ahmetov I, Donnikov AE, Trofimov DY. ACTN3 genotype is associated with testosterone levels of athletes. *Biol Sport*. 2014;31(1): 105–108.
- Belli T, Crisp AH, Verlengia R. Greater muscle damage in athletes with ACTN3 R577X (RS1815739) gene polymorphism after an ultra-endurance race: a pilot study. *Biol Sport*. 2017;34(2):105–110.
- Bouchard C, SARzynski MA, Rice T, et al. Genomic predictors of trainability. *Exp Physiol*. 2012;97(3):347–352.
- Craig Pickering, John Kiely. ACTN3: More than just a gene for speed, *Frontiers in Physiology*, 18<sup>th</sup> December. 2017.
- Del Coso j, Salinero JJ, Lara B, et al. ACTN3 X-allele carriers had greater levels of muscle damage during the half ironman. *Eur J Appl Physiol*. 2017;117(1):151–158.
- Ildus I, Ahmetov, Emiliya S Egorova, et al. Genes and athletic performance: an update. *Med Sport Sci*. 2016;61:41–54.
- Kim JH, Jung ES, Kim CH, et al. Genetic associations of body composition, flexibility and injury risk with ACE, ACTN3 and COL5A1 polymorphisms in Korean ballerinos. *J Exerc Nutrition Biochem*. 2014;18(2):205–214.
- Krzysztof Ficek, Pawel Cieszczyk, Mariusz Kaczmarczyk, et al. Gene Variants within the COL1A1 gene are associated with reduced anterior cruciate ligament injury in professional soccer players. *Journal of science and Medicine in sport*. 2013;16(5):396–400.
- Rosetti L, Kuntz A, Kunold E. The microstructure and micromechanics of the tendon-bone insertion, *Nature materials*. 2017.
- Shang X, Li Z, Cao X, et al. The Association between the ACTN3 R577X polymorphism and noncontact acute ankle sprains. *J Sports Sci*. 2015;33(17):1775–1779.
- Vancini R, Pesquero JB, Fachina RJ, et al. Genetic aspects of athletic performance of African runners phenomenon. *Open access J Sports Med*. 2014;5:123–127.
- Wessner B, Stuparits P, Fail C, et al. Genetic polymorphisms in alpha-actinin 3 and adrenoceptor beta genes in Austrian elite athletes and healthy controls. *Swiss sports & exercise medicine*. 2016;64(4):13–19.
- Yang R, Shen X, Wang Y, et al. ACTN3 R577X gene variant is associated with muscle-related phenotypes in Elite Chinese sprint/power athletes. *J Strength cond Res*. 2017;31(4):1107–1115.
- Gyrd O Gjevestad, Kirsten B Holven, Stine Mulven. Effects of exercise on Gene expression of inflammatory markers in Human peripheral blood cells: a systematic review. *Curr Cardiovasc Risk Rep*. 2015;9(7):34.
- Maqueda M, Roca E, Brotons D, et al. Affected pathways and transcriptional regulators in gene expression response to an ultra-marathon trail: Global and independent activity approaches. *Plos One*. 2017;12(10):e0180322.
- Candice Colbey, Amanda J Cox, David B Pyne, et al. Upper respiratory symptoms, gut health and mucosal immunity in athletes. *Sports Med*. 2018;48(1):65–77.
- Walsh NP. Recommendations to maintain immune health in athletes. *Eur J Sport Sci*. 2018;18(6):820–831.
- Orysiak J, Malczewska Lenczowska J, Bik-Multanowski M. Expression of SCGB1C1 gene as a potential marker of susceptibility to upper respiratory tract infections in elite athletes – a pilot study. *Biol Sport*. 2016;33(2):107–110.

19. Zehsaz F, Farhangi N, Monfaredan A, et al. IL-10 G-1082A gene polymorphism and susceptibility to upper respiratory tract infection among endurance athletes. *J Sports Med Phys Fitness*. 2015;55(1-2):128–134.
20. Cox AJ, Gleeson M, Pyne DB, et al. Cytokine gene polymorphisms and risk for upper respiratory symptoms in highly trained athletes. *Exerc Immunol Rev*. 2010;16:8–21.
21. Baumert P, Lake MJ, Stewart CE, et al. Genetic variation and exercise induced muscle damage: implications for athletic performance, injury and ageing. *Eur J Appl Physiol*. 2016;116(9):1595–1625.
22. Venckunas T, Skurvydas A, Brazaitis M, et al. Human alpha-actinin-3 genotype association with exercise induced muscle damage and the repeated bout effect. *Appl Physiol Nutr Metab*. 2012;37(6):1038–1046.
23. Del Coso J, Hiam D, Houweling P, et al. More than a 'speed gene': ACTN3 R577X genotype, trainability, muscle damage, and the risk of injuries. *Eur J Appl Physiol*. 2018;16.
24. Kikuchi N, Tsuchiya Y, Nakazato K, et al. Effects of the ACTN3 R577X genotype on the muscular strength and range of motion before and after eccentric contractions of the elbow flexors. *Int J Sports Med*. 2018;39(2):148–153.
25. McCulloch RS, Mente PL, O'Nan AT, et al. Articular cartilage gene expression patterns in the tissue surrounding the impact site following applications of shear and axial loads. *BMC Musculoskelet Disord*. 2018;19(1):449.
26. Ashwell MS, O'Nan AT, MG Gonda, et al. Gene expression profiling of chondrocytes from a porcine impact injury model. *Osteoarthritis Cartilage*. 2008;16(8):936–946.
27. Ashwell MS, Gonda MG, Gray K, et al. Changes in chondrocyte gene expression following in vitro impaction of porcine articular cartilage in an impact injury model. *J Ortho Res*. 2013;31(3):385–391.