

## REVIEW

A MINI REVIEW ON SPORTS GENETICS RESEARCHES IN INDIA:  
WHERE WE STAND NOW

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## ABSTRACT



Human genome consists of more than 20,000 genes. The genetic variants of each individual's genes propose characteristic sport capabilities. Genetic polymorphisms are strongly linked to physical performance in humans. Studies related to analysing the association between genetic variations and good athletic capability is of great interest. Athletic performance is highly related to sports training, anthropometric measures, and so on which are influenced by various relevant genes. In this review, we have focused on research papers based on genetic studies related to sports in order to get insight into the factors and underlying genes responsible for the best performance in Indian athletes.

## INTRODUCTION

**KEY WORDS**  
Genetics, Sports,  
Athletes, India,  
polymorphisms, ACL

Both genetic and environmental factors play an important role to be a good performer in athletes which is considered as a complex multifactorial trait [1]. The idea that genetics are strongly linked to human physical performance is believed among various researchers. If a player has participated in sports at a national or international level, he/she is considered an athlete [2]. De Moor et al (2007) [3] proposed that the athletic trait was inherited for approximately 66%. Researchers have identified certain genes related to sports performance, especially power, endurance and speed and are consistently increasing for in the past 2 decades [4, 5]. So far, greater than 200 gene polymorphisms have been discovered responsible for good performance in athletes [1, 6]. India is the second most highly populated country in the world and is generally diverse in nature. It is indeed important to perform genetic related studies in Indian athletes to better understand and recognize talented players to represent the country in games and make champions.

## CRITERIA ADOPTED FOR REVIEW OF LITERATURE

The literature search was performed in PubMed database by using the words "SPORTS" AND "GENETICS" AND "INDIA". The search ultimately resulted in 140 articles. However, the final relevant research papers included 7 records. The search also included the analysis of sub references to include any missed articles. Combining this, it totally resulted in 9 research papers. These articles focused on ACE gene polymorphisms, VEGFA polymorphisms, COL1A1 gene polymorphisms, and oxidative stress and genetic stability.

## GENETIC STUDIES RELATED TO SPORTS ON INDIAN PLAYERS

Sharma et al. (2012) [7] reported the occurrence of genomic instability in professional sports persons due to the comprehensive exercises and intense training that are carried out during practices resulting in disruption of the intracellular oxidant-antioxidant balance and destruction of macromolecules. Thus, the players are imposed with high risk related to cancer and various other diseases. Gandhi et al. 2019 [18] also found the elevated levels of genetic damage and oxidative stress in Kho-Kho players which might be due to the continuous physical exercises for sports events. Sharma et al (2012) [7] investigated the extent of genomic damage in judo players and revealed that the DNA and chromosomal damage were more prevalent in judo players when compared to controls. Gandhi and Kumar (2007) [8] analysed whether chromosomal damage occur in wrestlers due to extreme exercises and found statistically significant numbers of micronuclei

Das et al. (2019) [9] studied the genetic variants with respect to dopaminergic pathways and various other genes in persons involved in gambling sport (teer). They demonstrated the contribution of GDNF gene in the development and survival of dopaminergic neurons and also CNTNAP2 in psychological disorders. Shukla et al. (2020) [10] studied the relationship between the polymorphism specified as COL1A1 Sp1 + 1245 G > T of Sp1-transcription factor binding site and ACL (anterior cruciate ligament) injury risk in North-Indian athletes. However, they found no significant differences in the studied genotypic distribution of GT/TT and T-allele frequency distribution. Kothari et al. (2012) [11] found no association between ACE gene and athletic performance. However, they found over representation of I allele in the studied athletes.

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Prabhakar et al. (2018) [12] investigated the genetic association of *COL1A1* gene polymorphisms such as rs1800012 and rs1107946 with ACL injuries in which they found no significant differences in the genotypes or allele distribution between the ACL defective patients and control groups for both the polymorphisms rs1800012 ( $p=0.516$ ) and rs 1107946 ( $p=0.971$  for GT and  $p=0.823$  for TT). In another study, Shukla et al. (2020) [13] analyzed the link between *VEGFA* (Vascular endothelial growth factor) promoter polymorphisms and ACL tears. They identified the elevated frequencies of the A allele (rs699947) and I allele (rs35569394) in ACL group. **Table 1** shows the genetic studies related to sports in Indian sports players.

**Table 1:** Summary on Indian sports genetics studies carried out between 2011 and 2021

Sports	Study centre	Cases/Control	Findings	Ref
Hockey, Baseball	Punjab	36/20	Athletes are at high risk for various age-related diseases and cancer due to genomic instability	[7]
Kho-Kho	Punjab	18/10	Oxidative stress and genomic instability	[18]
Athletes ACL vs Non-ACL	Gwalior	90/76	A allele (rs699947), I allele (rs35569394) - significantly overrepresented in ACL cases rs1800012- Insignificant with ACL	[13]
Various sports	Chandigarh	52/52	Insignificant association of <i>COL1A1</i> with ACL	[12]
Athletes	Mumbai	147/131	No significant association; Overrepresentation of I allele of ACE	[11]
Judo	Punjab	17/10	Increased chromosomal and DNA damage	[7]
Wrestlers		15/10	Cytogenetic damage	[8]

## DISCUSSION

Genetics helps to determine a person's athletic abilities. A wide range of studies have shown varied connections between particular gene variants and athletic abilities in different sports and ethnic groups and genders [5]. *ACE* is one of the two genes that have been widely studied in combination with athletic ability [14]. An excess of the I allele is shown to be linked with some characteristics of endurance performance [15, 16].

Many researchers have demonstrated the oxidative stress and oxidation of cellular biomolecules by ROS affecting genomic stability in various professional sports players [17]. Time, intensity, frequency and features of exercises are known to show impact on oxidative stress and the oxidation of cellular macromolecules (including DNA) and cellular dysfunction, and can acquire with age, physical aspects and enhance disease risk [18]. Even though oxygen uptake during intense physical activity and during aerobic and anaerobic training is necessary in order to sustain stamina and endurance, it can, probably, lead to oxidative stress due to a disruption in intracellular homeostasis of the pro-oxidant system, as well as an increase in the formation of reactive oxygen/nitrogen species (ROS/RNS). Oxidative stress causes oxidation of cellular components viz. lipids, proteins and nucleic acids due to which continuous exercising leads to DNA damage [19]. Knowledge about genetic variants and their influence on many areas of training, competitive performance and injury prevention may surely enrich the periodization scheme, shorten injury time and successfully rehabilitate sports among athletes [20]. The associations of *COL1A1* gene polymorphisms with ACL tear were carried out on several populations namely Poland, South Africa, and Sweden by various researchers. Posthumas and his co-researchers observed under-representation of TT-genotype in their analysis of *COL1A1* gene in ACL injured sportspersons in South African population [21]. Similarly, Stepien Slodkowska et al. (2013) [22] identified 11245G/T polymorphisms in rs1800012 SNP in *COL1A1* gene in Poland population. Ficek et al. (2013) [23] found no significant association of *COL1A1* gene polymorphisms but observed over-representation of G-T haplotypes (-1997G+1245T) in Poland population. Similarly, no significant association of *COL1A1* gene polymorphisms with ACL tear was seen in Indian athletes. SNPs within genes coding for collagen matrix remodelling in singularity, linkage analysis, and gene-gene interactions are intriguing targets for further research in sports injuries in general and ACL injuries specifically. It has been found that the rate of ACL injury related to multifactorial aetiology to be 86% among Indian athletes [24].

Shukla et al. (2020) [10] proposed that the genetic link to ACL injuries and subsequent healing capacity might be helpful in identifying 'high risk' athletes related to ACL tears and configuring their trainings sessions for the purpose of continuing their "sporting career". *ACE* gene polymorphism is identified to be related to athletic abilities [25]. The functional polymorphism rs.5186 exists with absence (deletion; D allele; corresponding to good performance in sprint of power based sports), rather than the presence (insertion, I allele; related to excellence in endurance sports) of 287bp Alu repeat element in intron 16 (Thompson and Macleod, 2006). Researchers found that it is difficult to predict the connection between *ACE* genotype and sporting supremacy among sportspersons who take part in multiple sports rather than a single discipline [11].

## CONCLUSION

Very few genetic studies related to sports have been conducted in India. It is clear that among Indian athletes, the polymorphism COL1A1 Sp1 +1245 G> T SNP is not related to ACL. Studies should be focused on Indian athletes who are involved in single sport to get insight into the association between ACE and sporting capacity. Since oxidative stress due to continuous exercises can lead to severe diseases, conducting such studies in Indian athletes will allow preventing those diseases. Also, the studies are based on few sports only which should be extended on various other sports. Identification of polymorphisms in various other genes such as ACTN3, NOS3, UCP2, UCP3, and many is much more important for correlating different physiological parameters with sport performance. Also, majority of the studies were conducted in Northern region of India and is also gender biased. Thus, studies should combine sport players from all the parts of India including women to conclude the association of genes and its polymorphisms which would certainly help identification of talented players to give the best performance in sports.

### CONFLICT OF INTEREST

Author declares no conflict of interest.

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### FINANCIAL DISCLOSURE

None.

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