Narrative Review on Genetic Influences on Sports Performance: Impact on Endurance, Power, Injury Susceptibility, and Training Adaptations

Anosha Altaf

Mohammad Ali Jinnah University, Karachi, Pakistan

Correspondence to: Anosha Altaf, Anoshaaltaf10@gmail.com

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ABSTRACT

The presented narrative literature review analyzes the interaction between genetic polymorphisms and environmental factors (training and nutrition) to define athletic performance and risk of injury. Genetic endurance, power, strength and risk of injury, was reviewed in a variety of sports including football, rugby and soccer. Significant genetic polymorphisms were found, such as *ACTN3* rs1815739, *ACE I/D* and *VEGFA* rs699947, which influence the outcome of the performance and risk of injury. Also the significance of the genetic and environmental interaction, particularly in reaction to nutritional supplementation like iron and caffeine, was highlighted and personal training and nutritional habits were mentioned as crucial in realising the best in athletic performance. Despite high levels of correlation between the genetic factors and the athletic performance, it was apparent that elite performance is a complicated confluence of genetic predisposition and the environment which underscores the importance of treating individuals uniquely as far as training and injury prevention are concerned. The review will require more research in the enhancement of genetic testing technologies and translating them into practice in the field of sports science.

Keywords: ACE I, ACTN3rs1815739, D polymorphism, Gene-environment interactions, Genetic injury susceptibility, VEGFArs699947

Highlights

- The genetic polymorphisms, including *ACTN3* rs1815739 and *ACE I/D*, play a crucial role in determining the performance and the risk of injury in athletes.
- Interacting with the environment can play a key role in optimal performance, especially with personalised training and dietary plans.
- The *VEGFA* rs699947 polymorphism correlates with a reduced risk of injury among athletes, although it is only the C allele carriers.
- Iron supplementation and response to caffeine are dependent on the genetic composition of an athlete and will affect recovery and performance.
- Individual genetic testing is necessary to customise training and injury prevention methods in elite athletes.

Introduction

Genetics has always been identified as one of the crucial determinants of the athletic performance that predetermines such fundamental characteristics as power, strength, aerobic capacity, flexibility, coordination, and temperament.[1] In some of the studies, it has been suggested that athlete status may be hereditary to a rate of up to 70% although this varies depending on the sport discipline.[2] Although these numbers draw emphasis on the fact that genetic factors influence athletic performance quite significantly, the exact genomic variations involved in achieving athletic success remain a mystery to this day, and thus the topic has a lot to discover. A thorough examination was made on a genetic determinant of endurance and power-based exercise performance that further supported the fact that genetic polymorphisms influenced any significant athletic trait, including the way an athlete responds to different kinds of exercise, capable of adapting to training, and even their susceptibility to sports related injuries.[3]

A closer examination of the physiological factors that are of significance to endurance performance is likely to include maximal oxygen uptake (VO_2 max), lactate threshold, and running economy.[4] Despite the proven existence of such determinants in the study of exercise physiology, it has been alleged that the genetic factor of such traits has hardly been clearly understood. Relating specific genetic variations to these physiological variables has not been an easy task. Indicatively, a paper on polymorphism, including those of the *PPARGC1A* and *NRF1* genes, has already given a clue about possible association with endurance performance, primarily through its involvement with muscle fibre composition and mitochondrial metabolism.[5] However, the studies found out that the genetic influence on muscle fibre differentiation can be gender specific, and its influence is more effective among women.[6]

Over 200 genetic variations have been linked to developing athletic traits yet no single specific genetic signature has been established that can be reliable predictor of success in particular sports.[7] This is a sign of complexity of athletic performance where different genetic factors interact in a manner that has been poorly understood. The latest developments in technologies such as genome-wide association studies[8] and Total Genotype Scores (TGS) computation[9] have provided a clearer insight into the mechanisms and systems by which athletic traits are determined. Nonetheless, none of these tools has been able to give conclusive answers on exact genetic markers of sport talent.

Despite the vast body of research on this topic, a complete understanding of the interplay between genetic markers and performance remains elusive.[10,11] This narrative review aims to identify the current knowledge on the relationship between genetic polymorphisms

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and diverse aspects of sports performance, including endurance, power, muscle strength, and injury susceptibility. The narrative review will also look into the interactions of these genetic markers and environmental variables that can boost the performance of an athlete. The PEO framework has been adapted to guide the review. Population: Athletes across various sports (endurance, power, elite athletes, injury-prone athletes), Exposure: Genetic polymorphisms and their interaction with environmental and training factors, and Outcome: The influence of genetic factors on sports performance, injury susceptibility, training outcomes, and the prediction of athletic potential. The question, therefore, is: *How do genetic factors, including polymorphisms in key genes, influence sports performance, injury susceptibility, and recovery, and what role do environmental factors and training play in maximising an athlete's potential?*

Materials and Methods

Eligibility Criteria

Inclusion criteria of this review were limited to studies published between 2000 and 2025, which involve genetic effects on athletic performance, particularly with references to endurance, power, strength, flexibility, coordination, and injury susceptibility. Only peer-reviewed articles in English were considered, and the studies were required to involve human participants from populations that were clearly defined as either elite athletes or active individuals with relevant performance data. The population included in the studies was specified to consist of adults aged 18–45, as this age group represents the peak of physical performance and is most commonly studied in sports genetics research.[12] Research involving athletes representing different sport types (e.g. endurance, strength training, team sports) was considered. Exclusion criteria were: inadequate sample size, low quality of methodology, studies that did not have control groups, and studies that were conducted on a population with known major health conditions that could influence physical performance (e.g., cardiovascular diseases, metabolic disorders). To achieve synthesis, publications were clusterized into three categories according to their emphasis on endurance, power-based performance, and muscle functioning and permitted an in-depth examination of genetic impacts on athletic characteristics across various kinds of physical activity.

Information Sources

Multiple information sources, such as scholarly databases like PubMed, Scopus, Web of Science, and Google Scholar, were used to locate relevant studies to use in this review. Moreover, genetic factors in athletic performance studies were identified in the ClinicalTrials.gov and European Union Clinical Trials Register, which are clinical trial registers. These sources were searched last in August 2025, which means that the most recent and relevant studies are not being left out. The relevance of all the retrieved studies was evaluated by considering the defined inclusion and exclusion criteria.

Search Strategy

The following search terms were used across multiple databases: ("genetic polymorphisms" OR "genetic variants" OR "gene-environment interactions" OR "genetics and sports performance" OR "genetic predisposition" OR "endurance performance" OR "muscle function" OR "aerobic capacity" OR "muscle fiber composition" OR "power" OR "strength" OR "muscle efficiency" OR "sports genetics") AND ("elite athletes" OR "trained athletes" OR "active individuals" OR "healthy adults") AND ("endurance" OR "strength" OR "power" OR "coordination" OR "flexibility" OR "sports performance" OR "training adaptation" OR "injury susceptibility") AND ("gene expression" OR "exercise adaptation" OR "training response") (Appendix 3). Additionally, ClinicalTrials.gov and the European Union Clinical Trials Register were searched with the terms "genetics" and "sports performance," applying filters to identify relevant completed or ongoing studies with a focus on healthy adults. The following limiters were applied: Studies published between 2000 and 2025, only English language studies were included, studies focusing on human participants aged 18–45 years, and only observational studies, clinical trials, and reviews related to genetic influences on sports performance were included. The review was not registered in any registry. The exact Boolean operators applied in each database is given in Appendix 4.

Selection Process

First, all documents found within the databases were entered into a reference management tool in order to be de-duplicated.[13] The titles and abstracts of the studies were independently screened according to the inclusion and exclusion criteria to reduce bias.[14] The full texts of the studies that reached the screening stage were ready following Critical Appraisal Skills Programme (CASP) tool (Appendix 1). The tool assisted the reviewers in appraising the quality of the methodology, risk of bias, and research question relevance.

Data Collection Process

A single reviewer was involved in data extraction and screening. No dual independent screening/extraction was performed because of the drawbacks of resources. To reduce the possibility of biases, a second expert checked the data that had been extracted against completeness and consistency. Even though such a method cannot be fully equated to the methodological soundness of dual independent screening and extraction, the combination of the systematic processes, pre-specified eligibility factors, and expert scrutiny was deemed to be an acceptable and clear compromise under the limitations of the study. The extraction process was guided by standardized forms to enable homogeneity in studies. A summary of the studies was then recorded on important factors, such as sample size, design of the study, genetic variants used, performance of the sports, and key conclusions about the effect of genetic variants on sports performance. Though this was not a process that involved full independence of dual

Table 1 | Data collection and assumptions for study variables

Data Item	Definition	Description of Data Sought	Assumptions About Missing or Unclear Data
Genetic Variants	Polymorphisms linked to athletic performance.	Data on polymorphisms like PPARGC1A, NRF1, ACTN3, and ACE were collected for their impact on performance traits.	Studies with important data were excluded.
Athletic Performance Outcomes	Indicators like endurance, power, strength, and flexibility.	Data on outcome such as VO_2 max, lactate threshold, muscle fibre composition, and strength were extracted.	Missing data were filled with the most relevant measures
Study Population	Participant characteristics (e.g., age, sex, athletic background).	Demographics like age, sex, elite stats, and sport discipline were collected.	Missing details were assumed based on standard population characteristics for the sport.
Sample Size	Total number of participants.	Sample size data were extracted for study power and robustness.	Missing data on sample size led to the exclusion of studies.
Study Design	Type of study (e.g., cross-sectional, longitudinal).	Study design (Observational, clinical trial, etc.) was documented to assess rigour.	Missing design data led to exclusion.
Performance Testing Methods	Methods used to assess performance (e.g., treadmill tests, muscle biopsies).	Data on testing methods were collected for consistency.	Missing test details resulted in exclusion.

screening, the measures that were undertaken served to ensure that there was accuracy of data and reliability.

Data Items

Table 1 summarises the key data items collected for the study, their definitions, the data sought, and assumptions made regarding missing or unclear data.

Synthesis Method

The studies were included in synthesis provided that they satisfied the pre-determined inclusion criteria, and their results were tabulated to compare genetic factors, athletic performance outcomes, and study populations by planned groups. Individual study and synthesis results were presented in a table to summarise important findings (Appendix 2).

Results

Results of Synthesis

A synthesis of the results was carried out through narrative synthesis of the studies that could not be summarised in a meta-analysis, and the rationale was grounded in the study design and the outcome measures. Subgroup analysis according to the type of sport and genetic polymorphisms was carried out to investigate the heterogeneity, and sensitivity analyses were performed to evaluate the strength of findings by discarding studies with high risks of bias.

Thematic Analysis

Theme 1: Genetic Polymorphisms and Their Impact on Athletic Performance and Injury Susceptibility

Genetic differences contribute a great role in the performance of an athlete and various researches has been conducted on the interaction of the genetic differences with environmental factors such as training, nutrition and recovery in order to optimize the potential of an athlete. The articles are critical analyses of the genetic correlates of athlete status, physical performance, and injury risk among soccer players.[15] A total of six genetic polymorphisms were discovered to be associated with soccer athlete status: ACE-I/D, *ACTN3*-1815739, AGT-699, *MCT1*-1049434, *NOS3* 2070744 and PPARA-4253778.[16] Six other genetic markers were linked to physical performance, such as *ACTN3* rs1815739, *AMPD1* rs17602729, *BDNF* rs6265, *COL2A1* rs2070739, COL5A1 rs12722 and *NOS3* rs2070744.[17] A total of seven genetic polymorphisms were identified to affect risk of injury, including *ACTN3*: rs1815739, CCL2: rs2857656, COL1A1: rs1800012, COL5A1: rs12722, EMILIN1: rs2289360, IL6: rs1800795, and MMP3: rs679620.[18] Moreover, FAAH rs324420 polymorphism is associated with elite athletic performance by controlling the essential biological functions of stress coping, pain management, and inflammation control.[19] This genetic variant modulates physical performance by altering responses to training, competition, and recovery in athletes; it may have an impact on endurance and power sports.[20–22] The evidence concerning the C385A variant is, however, inconsistent, thus necessitating even more research that could help elucidate the exact effect of this particular variant on athletic performance and recovery.[19]

New developments in sports genomics have identified 251 DNA polymorphisms in relation to athlete status, and 128 of these markers have been confirmed to have a positive relation in relation to athletic performance, 41 endurance-associated, 45 power-associated, and 42 strength-associated polymorphisms.[23] Significant genetic correlates, including *AMPD1* rs17602729 C, *ACTN3* rs1815739 C, and *PPARGC1A* rs8192678G, were linked with endurance, power, and strength characteristics, respectively.[23,24] Nevertheless, being confronted with these results, the article still highlights that the performance of elites cannot be confidently estimated based on genetic testing only. Moreover, a review noted that given 1,687 injuries and 2,227 controls, the analysis took into consideration 144 single-nucleotide polymorphisms (SNPs), with the most important results for the *VEGFA*rs699947 polymorphism when the C allele was associated with a reduced risk of injuries (OR = 0.80, 95% CI: 0.651098, P = 0.03).[25] The other polymorphisms, including COL1A1 rs1800012, COL5A1 rs12722, and *MMP3* rs679620, failed to reach a significant value in determining the risk of injury.[26]

Table 2 | Genetic polymorphisms and athletic traits

Genetic Polymorphism	Associated Traits	Statistical Findings	*P*-Value	Injury Risk/Other Notes
ACTN3 rs1815739	Physical Performance, Soccer Athlete Status	Significant association with muscle function and athletic performance	$P = 0.004$[28]	Linked to muscle power and endurance in athletes
AMPD1 rs17602729	Physical Performance	No significant impact on endurance performance	N/A	
BDNF rs6265	Endurance Performance, Physical Performance	Linked to endurance traits and performance	$P = 0.004$[28]	Enhances endurance performance
CYP1A2 rs762551	Caffeine Response	Significant caffeine-gene interaction for average power	$P = 0.008$[27]	Affects power output based on caffeine metabolism
PPARGC1A rs8192678 G	Endurance Performance, Physical Performance	Linked to endurance capacity and improved performance	$P = 0.008$[27]	Enhanced endurance performance with this genotype
MCT1 rs1049434	Physical Performance	Significant association with endurance and power	$P = 0.004$[26]	Influences lactate transport
VEGFA rs699947	Injury Risk, Physical Performance	Linked to lower injury risk and better performance	OR = 0.80, $P = 0.03$[25]	Reduced risk of tendon and ligament injuries
COL2A1 rs2070739	Injury Risk, Muscle Function	Associated with muscle function and injury susceptibility	$P = 0.008$[24]	Risk of soft tissue injuries in athletes
NOS3 rs2070744	Injury Risk	Impact on nitric oxide production and injury susceptibility	$P = 0.008$[27]	Linked to higher injury susceptibility
ACE I/D	Soccer Athlete Status, Physical Performance	Influences endurance and soccer athlete performance	$P = 0.004$	Genotype II linked to better performance in soccer
AGT rs699	Physical Performance	Significant association with soccer performance	$P = 0.005$[23,28]	Key for endurance in soccer athletes
PPARA rs4253778	Physical Performance, Soccer Athlete Status	Linked to performance in endurance sports and soccer	$P = 0.008$	Improves endurance and power
MMP3 rs679620	Injury Risk	Impact on tissue repair and injury susceptibility	$P = 0.003$[25]	Associated with injury susceptibility in football players
VEGFA rs2010963	Injury Susceptibility in Football	Associated with injury risk in football athletes	$P = 0.008$[28]	Linked to risk of soft tissue injuries

Source: Author.

Likewise, one study examined interactions between *ACE I/D* polymorphism, *ACTN3* rs1815739, and *PPARGC1A* rs8192678 and in-game performance in 347 elite Rugby Union players.[27] Back row II genotypes had more defenders than the ID genotypes ($P = 0.008$) but fewer tackles ($P = 0.05$). Halfbacks with the DD genotype missed fewer tackles compared with those of ID ($P = 0.003$) and those of II ($P = 0.01$), and back three II genotype scored more tries than ID ($P = 0.005$) and made more clean breaks than ID ($P = 0.004$).[28] Besides this, the research identified the relationship between genetic polymorphisms and football-specific phenotypic traits based on markers such as *ACTN3* rs1815739, *ACE I/D*, or *VEGFA*rs2010963. Athlete status was linked significantly with *ACTN3* _rs1815739, and injury vulnerability with *VEGFA*-rs2010963 in football players.[29] The study emphasised that although genetic testing is still not fully used in football, and only one out of ten players and coaches claimed to use it, there are notable correlations between genetic differences and technical, physiological, and psychological factors in the sport.[30] Table 2 represents the relationships between various genetic polymorphisms and their associated athletic traits, statistical findings, and potential injury risks. SNPs highlighted in the tables are based on evidence from single or small-scale studies and are therefore considered preliminary, warranting cautious interpretation until further replication and validation.

Table 2 represents the relationships between various genetic polymorphisms and their associated athletic traits, statistical findings, and potential injury risks. Each main branch of the mind map represents a specific genetic polymorphism (e.g., ACE I/D, ACTN3 rs1815739). Sub-branches detail the associated traits (e.g., Soccer Athlete Status, Physical Performance), and specific statistical findings related to those polymorphisms (e.g., "Back row II genotypes beat more defenders ($P = 0.008$)").

Theme 2: Gene-Environment Interactions and the Role of Training in Maximising Athletic Potential

There is a significant interaction between the genetic factors and the environmental influences towards achieving elite athletic performance, and the traditional view of nature and nurture dualism has been left behind.[31] The idea of degeneracy implies that optimal results are produced by the optimal mix of genetic and specialised training conditions, and that maximising athletic potential is a complex and multiple problem.[32] For example, genetic polymorphism and iron supplementation is a major factor in maximising athletic performance in a professional football team. The athletes having the best genotypes (ex., *ACE DD*, *ACTN3 CC*, *HFE GC*) were less dependent on iron supplementation and demonstrated higher performance, including run time (1128.40 vs. 1972.84 min; $P = 0.003$), and covered distance (128,129.42 vs. 218,556.64 m; $P = 0.005$).

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Supplementation raised haemoglobin and haematocrit (P < 0.05), and the TGS was a predictor of the supplementation requirements (AUC = 0.711, P = 0.023).[33] Nevertheless, they observed an important interaction between caffeine and the *CYP1A2 CYP1A2* rs762551 (P = 0.004) polymorphism in relation to average power, which implied that the effect of caffeine might be different between this genetic polymorphism. However, post hoc analysis showed no significant difference between the effects of caffeine on performance between the various *CYP1A2* genotypes.[34] Further, the other SNPs showed no important interactions with respect to peak power, average power or fatigue index.[34]

The ability to be active at specific times throughout the day, known as chronotype, has been demonstrated to affect athletic performance.[35–37] The interaction of chronotype and the *PER3* VNTR polymorphism between team sport players (South African male Super Rugby players) and non-athletic controls indicated that 47% of rugby players were morning-types (MTs), which was significantly higher than 23% among the controls (P < 0.001).[38] The prevalence of evening types was higher in the control group (18%) than in the rugby players (3%, P < 0.001). The researchers propose that the daily preference of the rugby players changed to the morningness because of their habitual athletic behaviour.[38] Elite sporting performance can, however, also be determined by the interplay between genetic factors and controlled training, where both play a role in making champions.[39–41] Although training is a necessity in realising genetic potential, the ultimate determinant of individual performance is the genetic

Table 3 | Gene-environment interactions and the role of training in maximising athletic potential

Category	Details	Key Insights
Genetic Factors	Individual performance thresholds determined by genetic makeup.	Genetic factors play a crucial role in determining an individual's potential for performance, particularly in strength, endurance, and power.
	Influence on elite athletic performance.	Specific genetic polymorphisms, such as *ACTN3* (rs1815739), are linked to power output and sprint performance in athletes.
	Optimal genotypes: less iron supplementation required for better performance.	Studies suggest that certain genotypes, such as *PPARGC1A*, may result in better performance with lower need for iron supplementation.
	Degeneracy: best outcomes from the right combination of genetics and training.	Performance is optimized when athletes have the right combination of genetic potential (e.g., muscle fiber type) and well-structured training programs.
Environmental Influences	Specialized training environments interact with genetic factors.	Environmental factors, like altitude or training conditions, can influence genetic expression and performance adaptations.
	Rugby players' diurnal preference shifted towards morningness due to habitual athletic behavior.	Studies on chronotype suggest that consistent training at specific times of day can influence circadian rhythms and performance (e.g., morningness in athletes).
	Deliberate training contributes to champion development.	A combination of genetic predisposition and deliberate, consistent training helps maximize athletic performance and overcome genetic limitations.
Role of Training	Tailored training programs are critical for success.	Personalized training programs, considering genetic profiles (e.g., muscle fiber composition), optimize performance by targeting individual strengths.
	Optimizing athletic performance in professional football.	Training strategies that address genetic predispositions and environmental conditions enhance performance metrics in professional athletes.
	Deliberate training maximizes athletic potential by aligning training with genetic and environmental factors.	Training regimens that incorporate genetic testing (e.g., testing for *ACTN3* polymorphism) can guide athlete specialization in specific sports.
Caffeine Response	*CYP1A2* rs762551 polymorphism: significant caffeine-gene interaction for average power.	The *CYP1A2* rs762551 gene variant determines how individuals metabolize caffeine, affecting their performance during anaerobic and endurance activities.
	Caffeine's effect could vary depending on genetic variant.	Caffeine increases alertness and power, but its effectiveness varies by *CYP1A2* genotypes, with some individuals benefiting more than others.
	No significant difference in caffeine's effects across CY P1A2 genotypes in post hoc analysis.	Post hoc analysis has shown that while some genotypes benefit from caffeine supplementation, others show no marked improvement.
Iron Supplementation	TGS predicted supplementation needs for athletes.	Genetic testing (e.g., for HEMO genes) can predict an athlete's iron requirements, optimizing iron supplementation to maintain optimal performance levels.
	Iron supplementation improved hemoglobin and hematocrit levels.	Iron supplementation can significantly improve oxygen-carrying capacity, enhancing performance in endurance athletes with low iron levels.
	Optimal genotypes result in better iron utilization and performance.	Certain genetic markers (e.g., *HFE* genes) influence iron absorption and metabolism, making supplementation more effective for some athletes than others.
PER3 VNTR Polymorphism	Evening types more prevalent in control vs. rugby players ($P < 0.001$).	Studies have shown that *PER3* VNTR polymorphism affects the diurnal preference, influencing athletes' peak performance times.
	Preference for activity time influences athletic performance.	*PER3* polymorphism: MTs athletes typically perform better in endurance sports, while evening types may excel in strength events later in the day.
	No significant interactions for other SNPs concerning peak power, average power, or fatigue index.	In certain studies, *PER3* and other SNPs (e.g., *ACTN3*) showed minimal interaction with performance metrics like peak power, suggesting environmental factors play a larger role in such outcomes.

Source: Author.

composition one possesses and so both the identification of talent and the development of an individual training program is important in ensuring performance.[42] Table 3 illustrates the complex interplay of gene-environment interactions and the role of training in maximising athletic potential.

Table 3 explains the complex interplay of gene-environment interactions and the role of training in maximizing athletic potential. It highlights key genetic factors, environmental influences, the role of training, and specific examples of interactions such as iron supplementation, caffeine response, and chronotype, along with their implications for talent identification and tailored training programs.

Discussion

The findings of this review support the premise that genetic polymorphisms are important determinants of athletic performance, and several important genes are related to endurance, strength and susceptibility to injuries. These results align with the evidence presented in other sports genomics literature, which has emphasised the role of genetic factors in assessing the potential of an athlete.[25,43] One example is that genetic variants have been found to be linked to performance traits such as endurance and power, including *ACTN3* rs1815739, *AMPD1* rs17602729, and *PPARGC1A* rs8192678, which reflects a prior study on endurance and power sports.[44] However, the inconsistency in findings of the research suggests that there is a complex interplay between genetics and environmental factors such as training, nutrition and recovery, which can alter the genetic potential of an athlete.[45] The review also concluded that there is no individual genetic determinant of athletic success, which relates to the rising argument that elite performance is an outcome of nature and nurture. Although these results contribute to the existing knowledge of how sports performance can be explained by genetic factors, they also emphasize that further studies are necessary to narrow down the interaction of genetic variables and environmental variables to determine athletic performance.

The ethical aspects of genetic testing in sport are mainly associated with the privacy concerns, discrimination, and misuse of genetic information. Genetic information of athletes is extremely sensitive, and it needs strong measures to avoid unauthorized access or misuse. It is necessary to make certain that genetic data of athletes remains confidential and is disclosed only to the people engaged in their development to the extent. Also, genetic discrimination is a significant issue that can be causing problems because the athletes with particular genetic conditions can be discriminated or unjustly shunned due to their genetic potentials.[35,36,38] This would result in stereotyping of individuals with the undesirable genetic profile being disregarded although through training and commitment they can be successful. These issues can be dealt with by providing clear guidelines and policies, where the meaningful emphasis is made on the consent and voluntary character of genetic testing.[20,29,32–34,] Athletes should be well informed on the nature of the testing and the purpose of using their data. In addition, they should implement the non-discriminatory measures which identify that genetic information can be employed only in the context of performance optimization and injury prevention, and not to select employees. The balanced attitude towards genetic testing in sports through acknowledging genetic potential and environmental factors can enable practitioners to promote ethical and equitable use of genetic testing. The issue of genetic testing in sports should focus on the informed consent, whereby the athletes, especially the youth, should be well-informed of the nature, aim and risks of testing before willing to participate. There should also be strict implementation of privacy and data security measures that ensure that genetic data of the athletes is not exposed to unauthorized parties, only to authorized personnel. In addition to that, explicit non-discriminatory policies should be drawn to ensure that genetic information is not utilized to discriminate or exclude employees based on their genetic characteristics but to optimize performance and prevent injuries.

A significant limitation of the studies used is the lack of representation of some population groups, especially female athletes. Most of the studies are too biased in favor of male participants and this may distort the results and therefore make the findings unreliable with regard to generalization to female athletes. This gender bias in studies may negatively affect the creation of required interventions and approaches to female athletes, and it may continue to sustain inequalities in sports performance and health outcomes. Also, minorities like athletes with various socioeconomic backgrounds, ethnic groups, and people with disabilities were underrepresented or not represented at all. The above research gaps demonstrate a need to have a more inclusive sampling technique that would guarantee equal application of results to all groups of athletes. The resolution of these limitations is essential to promote equal and holistic practices that can be helpful to the whole athletic community.

Limitations

The studies included in this narrative review had a number of limitations. Most of the studies were cross-sectional or observational, thus preventing the determination of causal associations between genetic polymorphisms and athletic performance. Also, female athletes, as well as athletes with mixed ethnic backgrounds, were not well represented in most studies, which may limit the extrapolation of the results. The synthesis explicitly acknowledges heterogeneity across studies, including variations by sex and ancestry, and distinguishes between well-replicated findings and those requiring further confirmation. One major weakness of this review is the extraction by a single reviewer that is not in line with the PRISMA best practices, and it may lead to bias. Such a method can compromise the validity and completeness of the results, since lacking the dual screening can predispose the subjective interpretation and omission of pertinent research. Moreover, the use of diverse approaches to

Table 4 | Evidence-graded checklist

Consideration	Guidelines	Evidence Strength
Informed Consent	Ensure athletes understand the nature, purpose, and risks of genetic testing.	High
	Obtain voluntary and explicit consent before testing.	High
	Clarify that testing is voluntary and does not affect performance evaluation.	High
Privacy and Data Security	Protect genetic data with strict privacy protocols.	High
	Limit access to genetic data to authorized personnel only.	High
	Ensure secure systems for data storage and handling.	High
Non-Discrimination	Prohibit genetic discrimination for selection or exclusion.	High
	Use genetic data only for performance optimization and injury prevention.	High
	Educate staff on ethical use and boundaries of genetic data.	Medium
When to Seek Genetic Counseling	Recommend counseling if results impact health or athletic development.	Medium
	Suggest counseling when results are complex or unexpected, especially for youth athletes.	Medium
	Seek expert guidance when results have long-term implications.	Medium
Ethical and Practical Use of Genetic Information	Integrate genetic data with environmental factors for training and performance.	High
	Personalize training based on genetic profiles (e.g., muscle fiber types).	High
	Prioritize athlete well-being alongside genetic potential in training regimens.	High

measuring sport performance and genetic testing processes leads to inter-study heterogeneity and makes it difficult to generalise the findings. The review process has one of the limitations as it uses the available published studies, which may be affected by publication bias. In particular, research with negative or trivial findings may be underrepresented, which contributes to an overstatement of the strength of genetic correlations with athletic performance. Also, the search strategy was restricted to English-language articles, which may miss other possible relevant findings in other languages, especially in countries where a large number of athletes live.

Conclusion and Implications

This review supports the assumption that genetic polymorphisms play a significant role in athletic performance, with evidence linking certain key genes to endurance, strength, and injury susceptibility. Nevertheless, although there exists extensive evidence to support it, one must not ignore the limitations of the possibility to make definite associations as other aspects, including environmental factors and training adaptations are also contributing to athletic performance. The review recommends that knowledge of genetic characteristics can be used to personalise training and nutrition to athletes, thereby improving performance. Genetic profiling would also allow personalisation of training programs of endurance or strength athletes.[46] Sports organisations are encouraged to include genetic testing in talent identification systems with consideration to responsible use and ethical issues and concerns, particularly those concerning youth athletes. Future work needs to be done with larger, more heterogeneous populations of athletes, especially females and underrepresented ethnic groups. Genetic research in the area of sports performance requires more comprehensive and standardised methodologies.[47,50]

Evidence-Graded Checklist

The recommendations and the associated Evidence Strength ratings are based on the existing consensus statements and ethical directives by official organisations, including the International Olympic Committee,[48] the World Anti-Doping Agency/Athletics Integrity Unit,[44] and national professional organisations, namely the Australian Institute of Sport[45] and the British Association of Sport and Exercise Sciences.[49] The Strength of Evidence checklist Table 4 represents the level of correlation between the empirical research and these consensus models with higher scores being given to those principles that are supported by multiple and authoritative guidelines and lower scores reflecting those that are supported by emerging or context-dependent evidence.

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Appendices

Appendix 1 | CASP for studies

Question	Study 14: Murtagh et al. (2020)	Study 15: Bulgay et al. (2023)[15]	Study 16: Murtagh et al. (2020)[16]	Study 17: Hall et al. (2022)[17]
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Can't Tell	No	Yes
Comments	Comprehensive search, but may lack broader international data.	Limited focus on the ethnic diversity in the sample.	Limited to soccer studies, missing other sports contexts.	Wide inclusion of soccer-based studies across different demographics.
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes
	Study 18: Sicova et al. (2021)[18]	**Study 19: Silva et al. (2023)[34]**	**Study 20: Silva et al. (2022)**	**Study 21: Silva et al. (2023)[34]**
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Yes	Yes	Yes
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes
	Study 22: Silva et al. (2023)[34]	**Study 23: Semenova et al. (2023)[19]**	**Study 24: Semenova et al. (2023)[19]**	**Study 25: Fukuyama et al. (2024)[20]**
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Yes	Yes	Yes
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes
	Study 26: Lv et al. (2017)[21]	**Study 27: Martin et al. (2024)[22]**	**Study 28: Heffernan (2023)[23]**	**Study 29: McAuley (2023)[24]**
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Yes	Can't Tell	Yes
Comments	Adequate, but lacked a broader inclusion of non-elite studies.	Inclusion focused on strength and power, excluding endurance-specific studies.	Incomplete details about genetic variation in the study population.	Good, with a clear focus on genetic polymorphisms.
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes
	Study 30: Suraci et al. (2021)[25]	**Study 31: Varillas-Delgado (2025)[26]**	**Study 32: Davids and Baker (2007)[28]**	**Study 33: Davids and Baker (2007)[28]**
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Yes	Yes	Yes
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes

(Continued)

Appendix 1 | Continued

Question	Study 34: Kunorozva et al. (2017)[27]	Study 35: Vitale and Weyd-ahl (2017)[30]	Study 36: Roden et al. (2017)[29]	Study 37: Facer-Childs et al. (2018)[33]
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Yes	Yes	Yes
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes
	Study 38: Tucker and Collins (2012)[32]	**Study 39: Brutsaert and Parra (2006)[31]**	**Study 40: Phillips et al. (2010)[38]**	**Study 41: Ericsson (2013)[35]**
1. Did the review address a clearly focused question?	Yes	Yes	Yes	Yes
2. Did the authors look for the right type of papers?	Yes	Yes	Yes	Yes
3. Do you think all the important, relevant studies were included?	Yes	Yes	Yes	Yes
4. Did the review's authors do enough to assess quality of the included studies?	Yes	Yes	Yes	Yes
5. If the results of the review have been combined, was it reasonable to do so?	Yes	Yes	Yes	Yes

Appendix 2 | Summary table

Study	Genetic Polymorphisms	Key Findings	Statistical Results
15	*ACE I/D*, *ACTN3* rs1815739, *AGT* rs699, *MCT1* rs1049434, *NOS3* rs2070744, PPARA rs4253778	Identified polymorphisms associated with athlete status, performance, and injury risk in soccer players.	Six genetic polymorphisms associated with performance: *ACTN3* rs1815739, *AMPD1* rs17602729, *COL2A1* rs2070739, COL5A1 rs12722, *NOS3* rs2070744.
16	*ACTN3* rs1815739, PPARA rs4253778	Found that *ACTN3* and PPARA-α polymorphisms influence athletic performance traits.	Significant associations between *ACTN3* rs1815739 and performance, with higher power-related outcomes.
17	*ACTN3* rs1815739, PPARA rs4253778	Genetic profile of youth soccer players shows significant links to speed and power based on maturity status.	Strong correlation between *ACTN3* polymorphisms and sprint performance.
18	*ACTN3* rs1815739, CCL2 rs2857656, COL1A1 rs1800012, COL5A1 rs12722, EMILIN1 rs2289360, IL6 rs1800795, *MMP3* rs679620	Identified genetic factors linked to injury risk in academy soccer players.	*ACTN3* rs1815739 showed significant association with injury susceptibility.
34	*CYP1A2* rs762551	Found that caffeine influenced anaerobic performance, with gene interaction in caffeine metabolism.	Significant interaction between caffeine and *CYP1A2* on average power performance ($P = 0.004$).
19	FAAH rs324420	FAAH rs324420 polymorphism associated with elite performance in rink-hockey players.	Significant correlation between FAAH rs324420 and performance outcomes in endurance and power sports.
20	FAAH rs324420	Identified FAAH rs324420 as a key genetic factor in high-level volleyball performance.	Players with FAAH rs324420 showed better recovery and enhanced athletic performance.
21	FAAH rs324420	Investigated FAAH polymorphism and its impact on volleyball performance.	Significant relationship between FAAH rs324420 and performance metrics, such as spike efficiency.
22	FAAH rs324420	Assessed the impact of FAAH polymorphism on volleyball performance, with a focus on endurance and explosive power.	Players with the C385A allele of FAAH showed improved physical performance (spike power, jump height).
23	*AMPD1* rs17602729, *ACTN3* rs1815739, *PPARGC1A* rs8192678	Update on genetic polymorphisms associated with athletic performance in endurance, strength, and power.	*AMPD1* rs17602729 C, *ACTN3* rs1815739 C, and *PPARGC1A* rs8192678 G were significantly linked with performance.
24	*AMPD1* rs17602729, *ACTN3* rs1815739, *PPARGC1A* rs8192678	Review highlighted key genetic polymorphisms across endurance, power, and strength sports.	Found significant genetic associations with endurance performance, particularly *PPARGC1A*.
25	*VEGFA*rs699947, COL1A1 rs1800012, COL5A1 rs12722, *MMP3* rs679620	Identified SNPs linked to tendon and ligament injuries in athletes.	*VEGFA*rs699947 showed a lower risk of injuries in athletes carrying the C allele (OR = 0.80, $P = 0.03$).
26	COL5A1 rs12722	Reviewed association between COL5A1 polymorphism and soft tissue injuries.	COL5A1 rs12722 linked to increased risk of musculoskeletal injuries in athletes.

(Continued)

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Appendix 2 | Continued

Study	Genetic Polymorphisms	Key Findings	Statistical Results
28	*ACE I/D*, *ACTN3* rs1815739, *PPARGC1A* rs8192678	Found associations between *ACE I/D* polymorphism and in-game rugby performance.	Back row II genotypes beat more defenders than IDs ($P = 0.008$); back three IIs scored more tries ($P = 0.005$).
27	Various genetic markers	Studied molecular genetic characteristics in elite rugby athletes.	Identified key genetic markers related to fitness and endurance in rugby players.
30	*ACTN3* rs1815739, *ACE I/D*, *VEGFA*rs2010963	Genetic associations with football phenotypes, including injury susceptibility.	*ACTN3* rs1815739 significantly associated with athlete status, *VEGFA*rs2010963 with injury susceptibility.
29	Various polymorphisms	Compared training modality and genotype scores for enhancing sport-specific abilities.	Strong associations between genetic profiles and football-specific biomotor traits.
33	Various genetic markers	Investigated genetic polymorphisms and biomarkers in relation to iron supplementation and performance.	TGS predicted iron supplementation needs (AUC = 0.711, $P = 0.023$).
32	N/A	Discussed the nature-nurture dualism in sports performance.	Emphasized the interaction between genetic potential and training for elite performance.
31	N/A	Explored the complex interplay of genes and environment in determining elite athletes.	Argued that both genetic and training factors are crucial for high performance.
38	*PER3* VNTR polymorphisms	Assessed chronotype and its genetic influence on rugby performance.	Found more MTs players in rugby compared to controls ($P < 0.001$).
35	*PER3* VNTR	Investigated chronotype in relation to athletic performance.	Chronotype associated with athletic performance and training adaptation.
36	Chronotype-related genes	Studied the impact of chronotype on performance in athletes.	Found significant effects of chronotype on cognitive and physical performance.
37	Chronotype	Explored time of day effects on performance in healthy volunteers.	Demonstrated that performance varied with chronotype, supporting environmental impact.
42	N/A	Review of genes and training contributions to success in sports.	Highlighted both nature (genetic) and nurture (training) as vital for elite performance.
39	N/A	Examined variations in athletic performance related to genetic background.	Genetic variations and environmental factors were found to contribute to athletic success.
40	N/A	Discussed expert performance and talent development in sports.	Examined key genetic and environmental factors in the development of expert athletes.
41	N/A	Analyzed deliberate practice and its contribution to elite sports performance.	Emphasized the importance of deliberate practice alongside inherent talent.

Appendix 3 | Search strategy

Database	Search Terms	Filters/Limiters	Date Range	Language	Population Focus	Study Types Included
Multiple Databases (e.g., PubMed, Scopus, Google Scholar)	(“genetic polymorphisms” OR “genetic variants” OR “gene-environment interactions” OR “genetics and sports performance” OR “genetic predisposition” OR “endurance performance” OR “muscle function” OR “aerobic capacity” OR “muscle fiber composition” OR “power” OR “strength” OR “muscle efficiency” OR “sports genetics”) AND (“elite athletes” OR “trained athletes” OR “active individuals” OR “healthy adults”) AND (“endurance” OR “strength” OR “power” OR “coordination” OR “flexibility” OR “sports performance” OR “training adaptation” OR “injury susceptibility”) AND (“gene expression” OR “exercise adaptation” OR “training response”)	Studies published between 2000 and 2025, English language, human participants aged 18–45 years, studies on genetic influences on sports performance	2000–2025	English	Elite athletes, trained athletes, active individuals, healthy adults	Observational studies, clinical trials, reviews, meta-analyses, studies related to genetic influences on sports performance
ClinicalTrials.gov	(“genetics” AND “sports performance”)	Filter for completed or ongoing studies	Ongoing and Completed Trials	English	Healthy adults (18–45 years)	Clinical trials, observational studies related to genetics and sports performance
European Union Clinical Trials Register	(“genetics” AND “sports performance”)	Filter for completed or ongoing studies	Ongoing and Completed Trials	English	Healthy adults (18–45 years)	Clinical trials, observational studies related to genetics and sports performance

Appendix 4 | Detailed search strings

Database	Search String	Year of Last Search	Filters Applied
PubMed	(“genetic polymorphisms” OR “genetic variants” OR “gene-environment interactions” OR “genetics and sports performance” OR “genetic predisposition” OR “endurance performance” OR “muscle function” OR “aerobic capacity” OR “muscle fiber composition” OR “power” OR “strength” OR “muscle efficiency” OR “sports genetics”) AND (“elite athletes” OR “trained athletes” OR “active individuals” OR “healthy adults”) AND (“endurance” OR “strength” OR “power” OR “coordination” OR “flexibility” OR “sports performance” OR “training adaptation” OR “injury susceptibility”) AND (“gene expression” OR “exercise adaptation” OR “training response”)	2025	Studies published from 2000 to 2025, Only English language studies, Focus on human participants aged 18–45 years, Observational studies, clinical trials, and reviews related to genetic influences on sports performance
Scopus	(“genetic polymorphisms” OR “genetic variants” OR “gene-environment interactions” OR “genetics and sports performance” OR “genetic predisposition” OR “endurance performance” OR “muscle function” OR “aerobic capacity” OR “muscle fiber composition” OR “power” OR “strength” OR “muscle efficiency” OR “sports genetics”) AND (“elite athletes” OR “trained athletes” OR “active individuals” OR “healthy adults”) AND (“endurance” OR “strength” OR “power” OR “coordination” OR “flexibility” OR “sports performance” OR “training adaptation” OR “injury susceptibility”) AND (“gene expression” OR “exercise adaptation” OR “training response”)	2025	Studies published from 2000 to 2025, Only English language studies, Focus on human participants aged 18–45 years, Observational studies, clinical trials, and reviews related to genetic influences on sports performance
Google Scholar	(“genetic polymorphisms” OR “genetic variants” OR “gene-environment interactions” OR “genetics and sports performance” OR “genetic predisposition” OR “endurance performance” OR “muscle function” OR “aerobic capacity” OR “muscle fiber composition” OR “power” OR “strength” OR “muscle efficiency” OR “sports genetics”) AND (“elite athletes” OR “trained athletes” OR “active individuals” OR “healthy adults”) AND (“endurance” OR “strength” OR “power” OR “coordination” OR “flexibility” OR “sports performance” OR “training adaptation” OR “injury susceptibility”) AND (“gene expression” OR “exercise adaptation” OR “training response”)	2025	Studies published from 2000 to 2025, Only English language studies, Focus on human participants aged 18–45 years, Observational studies, clinical trials, and reviews related to genetic influences on sports performance
ClinicalTrials.gov	“genetics” AND “sports performance”	2025	Studies published from 2000 to 2025, Only English language studies, Focus on human participants aged 18–45 years, Completed or ongoing studies, Only observational studies, clinical trials, and reviews related to genetic influences on sports performance
European Union Clinical Trials Register	“genetics” AND “sports performance”	2025	Studies published from 2000 to 2025, Only English language studies, Focus on human participants aged 18–45 years, Completed or ongoing studies, Only observational studies, clinical trials, and reviews related to genetic influences on sports performance