IJSHER
Uluslararası Spor, Sağlık, Egzersiz ve Rekreasyon Araştırmaları Dergisi
International Journal of Sport, Healt, Exercise and Recreation Research

International Journal of Sport, Health, Exercise and Recreation Research, 1(2), October 2025.

The Relationship Between Athletic Performance and Genetics in Female Athletes

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Derleme Makale / Review Article Doi: https://doi.org/10.5281/zenodo.17565298

Gönderi T. / *Received* : 15.08.2025 **Kabul T. / *Accepted* :**30.09.2025 **Online Yayın T. / *Published* :** 15.10.2025

Abstract:

The aim of this review is to examine the genetic, physiological, hormonal, and environmental factors that influence athletic performance in female athletes and to explain performance differences specific to sex. This review has been prepared based on scientific sources related to the subject. A significant portion of athletic performance is associated with hereditary characteristics, and certain genetic variants play a determining role in muscle structure, power production, aerobic capacity, and recovery processes. Female athletes differ from males in terms of physiological structure, muscle-to-fat ratio, hormonal cycles, and energy metabolism. In particular, fluctuations in estrogen and progesterone levels may influence training performance and injury risk. Additionally, conditions such as low energy availability and the Female Athlete Triad pose critical risks for both performance and long-term health. Female athletes may also face psychosocial pressures related to societal expectations and body image. The increasing use of genetic testing in sports has the potential to contribute to talent identification and individualized training planning; however, ethical considerations and fair competition principles must be taken into account. In conclusion, enhancing athletic performance in female athletes requires not only physical training but also integrated approaches that consider genetic traits, hormonal balance, nutrition, injury prevention, and psychological support.

Keywords: Genetic, Female Athletes, Performance

Özet

Bu derlemenin amacı, kadın sporcularda atletik performansı etkileyen genetik, fizyolojik, hormonal ve çevresel faktörleri bütüncül bir çerçevede ele almak ve cinsiyete özgü performans farklılıklarını açıklamaktır. Bu derleme, konuya ilişkin bilimsel kaynaklar temel alınarak hazırlanmıştır. Spor performansının önemli bir kısmı kalıtsal özelliklerle ilişkilidir ve bazı genetik varyantlar kas yapısı, güç üretimi, aerobik kapasite ve toparlanma süreçleri üzerinde belirleyici rol oynar. Kadın sporcularda fizyolojik yapı, yağ-kas oranı, hormonal döngü ve enerji metabolizması erkeklerden farklıdır. Özellikle östrojen ve progesteron düzeylerindeki değişimlerin antrenman performansı ve sakatlık riskleri üzerinde etkili olabileceği düşünülmektedir. Buna ek olarak, düşük enerji mevcudiyeti ve buna bağlı gelişen Kadın Atlet Triadı gibi durumlar performans ve uzun dönem sağlık açısından kritik bir risk oluşturur. Kadın sporcular, toplumsal beklentiler ve beden algısına ilişkin psikososyal baskılarla da karşı karşıya kalabilmektedir. Genetik testlerin spor alanında kullanımının artması, spora yatkınlık ve bireysel antrenman planlaması açısından potansiyel taşımakla birlikte, etik ve adil rekabet ilkeleri gözetilmelidir. Sonuç olarak, kadın sporcularda performansın geliştirilmesi, yalnızca fiziksel antrenmanla değil; genetik özellikler, hormonal düzen, beslenme, sakatlık önleme ve psikolojik destek süreçlerinin uyumlu biçimde ele alınmasıyla mümkündür.

Anahtar Kelimeler: Genetik, Kadın Sporcular, Performance

Introduction

It is widely acknowledged that some individuals are naturally gifted in specific physical attributes, and that these attributes are closely related to sports performance (Andrijasevic et al., 2005). In some cases, the fact that talented individuals come from talented families suggests that genetics may be partly responsible for the physical, physiological, or anthropometric characteristics required to achieve athletic success. Indeed, elite athlete status is partly heritable; studies indicate that between 30% and 80% of the variance in this trait can be explained by genetic factors. Recent advances in genetic technology have enabled further exploration of the genetic foundations of elite performance, leading to

the identification of single-nucleotide polymorphisms and other genetic variants with the potential to influence sports performance either directly or indirectly (Moran & Pitsiladis, 2017).

When sport is defined as the organised playing of competitive games according to established rules, fixed regulations become essential in maintaining the boundaries of fair competition. Violations of eligibility or competition rules—such as the use of banned substances, illicit equipment, or match-fixing—create unfair competitive advantages that undermine the principles of fair play. Genetic factors are considered to play a substantial role in athletic performance and in associated phenotypes such as strength, aerobic capacity, flexibility, and coordination (De Moor et al., 2007). Despite the relatively high heritability of athlete status (up to 70%, depending on the sport discipline) and of intermediate phenotypes, the investigation of genetic variants contributing to predisposition for success in specific types of sports has attracted considerable scholarly interest.

Sports genomics is a scientific discipline that focuses on the organisation and function of the genomes of elite athletes. The field emerged in the early 2000s with the discovery of the first genetic markers associated with athletic performance (e.g., angiotensin-converting enzyme, Actin-3). With the widespread adoption of genotyping, sequencing, and DNA microarray technologies, a substantial number of genetic studies evaluating sport-related gene variants have been conducted, directly affecting all athletes who hold elite status.

Nearly all elite sports disciplines are divided into male and female categories. The primary rationale for this separation is to provide women with a fair opportunity to compete. On average, women face considerable disadvantages compared to men, who are typically taller, stronger, faster, and possess larger, more powerful muscles and bones, as well as greater endurance capacity. Consequently, elite women's competitions necessarily operate as a protected category, maintained by objective eligibility criteria linked to sex-specific physical advantages (Handelsman et al., 2018). The practical need to determine eligibility criteria for elite women's athletics led the International Association of Athletics Federations (IAAF) to establish regulations on hyperandrogenism in 2015, approved by the International Olympic Committee (IOC).

Given the extensive and potentially far-reaching impact of genetic variation on achieving elite athlete status, there is also a growing need to collect genetic information for identifying athletes with the potential to reach elite performance levels (Pickering et al., 2019). Indeed, the number of companies offering direct-to-consumer genetic testing aimed at predicting sports performance and risk of injury has increased rapidly in recent years. Contemporary research highlights the increasing use of genetic testing in talent identification processes across a wide range of countries and sporting populations (Webborn et al., 2015).

Although women's participation in the sports world has risen in recent years, the biological and environmental factors affecting athletic performance in female athletes remain a central focus of extensive research. Physiological differences between women and men have important implications for training adaptations, injury risk, and performance boundaries. In this context, understanding the factors influencing athletic performance in female athletes will contribute to the development of more effective training programs and the establishment of sex-specific performance strategies.

Physiological and Hormonal Effects

The physiological profiles of female athletes differ from those of males in terms of muscle mass, cardiovascular capacity, and energy metabolism. On average, women exhibit a higher percentage of body fat and lower muscle mass (Maughan et al., 2010). These distinctions can have a direct impact on

performance, particularly in sports that require strength and speed. Cyclical fluctuations in hormones such as estrogen and progesterone may influence both physical and psychological performance. For instance, during the luteal phase of the menstrual cycle, increases in core body temperature and perceived fatigue are commonly observed (McNulty et al., 2020). Nevertheless, recent studies suggest that in some female athletes, these hormonal variations do not significantly impact performance.

Injury Risk and the Musculoskeletal System

Female athletes are more susceptible than males to specific injuries, particularly anterior cruciate ligament (ACL) ruptures. Contributing factors include anatomical differences, hormonal influences, and variations in neuromuscular control mechanisms (Hewett et al., 2006). In this context, the implementation of strength, neuromuscular, and proprioceptive training programs tailored specifically to women is of critical importance.

Nutrition and Energy Balance Issues

Among female athletes, low energy availability (LEA) and associated conditions, such as the Female Athlete Triad—characterised by menstrual dysfunction, inadequate energy intake, and reduced bone mineral density—may severely impair performance (Nattiv et al., 2007). Preventing these issues requires careful planning of the athlete's energy intake and expenditure to ensure adequate physiological functioning and optimal performance capacity.

Psychosocial Factors

Gender roles, body-image concerns, and biases embedded within sports culture can influence female athletes' self-confidence and motivation. Several studies report that female athletes tend to exhibit higher levels of anxiety; however, this can be mitigated through strong social support mechanisms (Krane et al., 2004).

Sport-Specific Approaches

Each sport discipline imposes distinct physiological demands on female athletes. For example, in endurance sports, women's relatively greater fat stores may confer an advantage in energy utilization. Consequently, sex-specific periodized training plans may yield more effective outcomes in load management and recovery. The athletic performance of female athletes cannot be explained solely by physiological differences; hormonal, psychological, environmental, and sociocultural factors must also be taken into account. In this regard, multidisciplinary strategies that incorporate sex-specific training, nutrition, and rehabilitation programs play a critical role in maximizing the performance of female athletes.

Female athletes often aim to achieve a lean and muscular body composition to enhance competitive success. Preparing for a physique-oriented natural competition can yield numerous health benefits, including improved cardiovascular conditioning, increased muscular strength, an enhanced sense of accomplishment, and temporary elevations in self-esteem (Halliday et al., 2016). Despite these positive outcomes, a range of adverse effects may also occur, including reductions in reproductive hormone levels and symptoms associated with eating disorders. Existing research on performance-oriented athletes suggests that prolonged energy restriction and intensive training regimens are often employed to achieve and maintain a lean physique—practices that increase the risk of low energy availability and its associated consequences.

Method

Research Model

This study is based on a qualitative review (literature review) model grounded in previously published academic research. The primary aim of this study is to evaluate, from a holistic perspective, the genetic, physiological, and environmental factors that influence athletic performance in female athletes.

Research Design

The research was conducted within the framework of a descriptive systematic review model. This model is employed to collect, classify, and interpret existing scientific knowledge on a specific topic. The data obtained for the review were evaluated through the analysis of conceptual and theoretical content rather than quantitative datasets.

Data Collection Process

During the data collection phase, the following international and national academic databases were utilized:

- PubMed
- Web of Science
- Scopus
- Google Scholar
- TR Dizin

Search procedures were limited to studies published within the last five years. Keywords were selected in both Turkish and English, including combinations such as: “female athletes,” “genetics,” “athletic performance,” “sex differences,” “hormonal cycle,” “sports genomics,” “kadın sporcu,” “genetik,” “atletik performans,” “fizyolojik farklılıklar,” “spor genetiği,” “kadın atlet triadı.”

- The following inclusion criteria were considered in selecting studies:
- Research examining physiological, hormonal, or genetic factors specific to female athletes,
- Articles assessing the relationships between sports performance and genetic markers,
- Peer-reviewed original studies or systematic reviews with full-text availability.

Data Analysis and Evaluation

The collected studies were analyzed thematically. The findings were classified under the following categories: genetic factors, physiological and hormonal effects, injury risk and the musculoskeletal system, nutrition and energy balance, and psychosocial and environmental factors. Prominent results within each theme were comparatively evaluated by considering methodological distinctions across studies. Through this approach, the multidimensional nature of athletic performance in female athletes was interpreted within the context of genetic–environmental interactions.

Findings

Body Composition and Lean Mass Dynamics

The pursuit of an extremely low fat mass and a high lean mass-to-strength ratio among female athletes has been extensively examined in the literature. In one study, a female athlete was monitored over a 20-week competition preparation period followed by a 20-week recovery phase; evaluations of

energy intake, body mass/composition, and menstrual cycle fluctuations revealed that energy intake, body mass, and body fat percentage decreased during preparation, while only limited normalisation occurred during recovery (Kairaitis et al., 2025).

Similarly, another study tracked energy and macronutrient intake, body composition, and plasma leptin levels of female athletes during a 7-week preparation period. Findings showed a reduction in energy intake from 1700 kcal to 1520 kcal, a 4.2-kg loss in total body mass, and a slight decline in lean mass. As with many factors influencing physical performance, ankle mobility appears frequently in the literature. Damage to neural, ligamentous, or contractile tissues across the multisegmental ankle–foot complex can impair joint mobility and stability (Baydemir & Dilican, 2025). Ankle flexors are one of the muscle groups that require optimal mobility. Collectively, these data indicate that the "lean and muscular" objective among female athletes is primarily driven by strategies aimed at substantially reducing fat mass. In contrast, lean mass tends to be maintained or only minimally altered throughout the process.

Energy Status, Resting Metabolic Rate, and Low Energy Availability (LEA)

Intensive training regimens and caloric restriction during preparation phases substantially increase the risk of low energy availability in female athletes. For instance, one study reported that the prevalence of low energy availability among athletic women ranged from 18% to 58%, with functional hypothalamic amenorrhea observed at exceptionally high rates among middle- and long-distance runners (e.g., 60% among elite athletes) (Melin et al., 2019). Another study demonstrated significant reductions in resting metabolic rate during the pre-competition period (e.g., −191 kcal/day) and found that the prevalence of amenorrhea increased to 24% prior to competition (Mathisen et al., 2020). More specifically, a study involving 104 female physique athletes reported that 58.65% exhibited either amenorrhea or the absence of menstruation for at least three consecutive months, confirming a high risk of low energy availability in this population (Witkoś et al., 2024).

Hormonal and Reproductive Effects

Menstrual irregularities such as amenorrhea and oligomenorrhea—commonly observed in female athletes exposed to energy restriction and intensive training regimens—represent the most concrete indicators of hormonal imbalances and impaired reproductive function. One study reported that female athletes striving for high leanness and low body fat frequently undergo prolonged caloric restriction combined with high training loads. This situation contributes to chronic low energy availability, which in turn heightens the risk of decreased reproductive hormone levels, reduced bone mineral density, weakened immune function, and adverse metabolic outcomes. Additionally, reviews have documented that low estrogen levels are associated with reductions in bone mineral density, highlighting that low energy availability and amenorrhea pose substantial risks to skeletal health in female athletes (Alwan et al., 2019).

Performance, Strength, and Recovery Parameters

Data on variables such as muscular strength, neuromuscular performance, and recovery are available for female athletes striving to achieve a lean body composition. In one study, a female athlete undergoing competition preparation and post-competition monitoring demonstrated a marked reduction in fat mass. In contrast, lean mass was maintained and even increased during the recovery phase. However, concomitant declines in resting metabolic rate and neuromuscular strength were also reported.

These findings suggest that achieving the "ideal" body composition comes at a risk to strength and physiological capacity (Tinsley et al., 2019).

Similarly, research examining the accuracy of body composition assessments found significant discrepancies between measurement methods in lean female athletes (Houska et al., 2018), serving as an important caution for both scientific monitoring and practical application. From a performance standpoint, the literature emphasises that low energy availability can impair recovery, muscle repair, immune function, and overall training capacity.

Findings Related to Psychological Factors and Eating Behaviours

The stringent energy restrictions and fat-reduction strategies commonly utilised by female athletes impose psychological as well as physiological burdens. Studies have noted that during the post-competition period, athletes may experience reductions in their resting metabolic rate, increased hyperphagic tendencies (heightened hunger), weight cycling, distorted body image, and an elevated risk of disordered eating or maladaptive eating behaviours (Luther & West, 2025).

DISCUSSION

The current literature reviewed in this study demonstrates that female athletes' pursuit of a lean and muscular physique for competitive success generates multidimensional physiological and psychological effects. Findings particularly emphasise that over the past five years, issues such as low energy availability, menstrual disturbances, metabolic adaptations, and disordered eating behaviours have been widely reported (Logue et al., 2020; Fagerberg, 2018).

Energy Imbalance and Metabolic Adaptation

Female athletes commonly reduce their energy intake while simultaneously increasing training loads during competition preparation periods. This combination exacerbates energy imbalance, leading the body to develop metabolic adaptations. Reductions in resting metabolic rate, declines in leptin concentrations, and diminished thyroid function are among the physiological responses indicative of these adaptations (Robinson et al., 2015). Although this process may support the short-term achievement of desired body composition, it substantially increases the long-term risk of developing Relative Energy Deficiency in Sport (RED-S). This syndrome affects not only the reproductive system but also immune function, cardiovascular health, bone mineral density, and psychological well-being (Mountjoy et al., 2018).

Effects on Hormonal and Reproductive Health

The literature consistently highlights that low energy availability in female athletes can lead to reductions in estrogen and progesterone concentrations and may induce hypothalamic amenorrhea (Loucks et al., 2013; Craighead et al., 2019). Such hormonal disruptions affect not only reproductive health but also the integrity of the musculoskeletal system and bone mineral density. Therefore, female athletes who engage in prolonged caloric restriction should be monitored through specialised programs that address bone health and injury risk. Findings further indicate that hormonal fluctuations should be taken into account when planning performance-related strategies, such as periodised training.

Psychological and Behavioural Dimensions

Eating behaviours and body image concerns constitute another critical dimension for female athletes. Research indicates that strict dieting practices and aesthetic pressures during competition phases elevate the risk of eating disorders and body dissatisfaction (Shriver et al., 2013). Moreover, continued metabolic adaptation following competitions can lead to weight regain, guilt, and

psychological fluctuations. This highlights the importance of providing psychological support in conjunction with performance-oriented training. Appropriate psychological counselling and nutritional guidance may mitigate these risks during post-competition recovery (Hulmi et al., 2016).

Balancing Performance and Health

Female athletes must navigate a delicate balance between enhancing performance and maintaining health. While increasing lean mass contributes positively to performance, excessive caloric restriction and very low body-fat levels may diminish performance capacity over time (Tarnopolsky, 2008). Consequently, training and nutrition programs should prioritise not only short-term aesthetic goals but also long-term sustainable performance and overall health. Recent literature demonstrates that maintaining adequate energy availability and adopting individualised training and nutrition strategies can optimise performance while reducing health risks in female athletes (Escalante et al., 2023).

Genetics and Individual Variability

Genetic research has shown that some female athletes exhibit individual differences in energy metabolism, muscle fibre type distribution, and hormonal responsiveness (Ahmetov & Fedotovskaya, 2015). These variations may contribute to differential responses to identical training or dietary regimens. Therefore, future studies are encouraged to examine the relationship between genetic markers and performance within a female-specific physiological context in greater depth.

CONCLUSION

The pursuit of a lean and muscular body composition for competitive success among female athletes may yield short-term improvements in aesthetic appearance and performance; however, it must be considered alongside significant long-term risks, including low energy availability, hormonal dysfunction, reductions in bone mineral density, weakened immune function, and psychological disturbances. Therefore, a comprehensive evaluation of energy balance, hormonal health, psychological resilience, and individual genetic variability is essential within the performance planning of female athletes.

A multidisciplinary structure—integrating coaches, sports physicians, nutritionists, and psychologists—will support both sustainable performance and overall athlete health. Individualised strategies that account for sex-specific physiological, hormonal, and psychosocial characteristics contribute to optimal outcomes not only during competition periods but throughout the entirety of an athlete’s career.

In conclusion, achieving long-term success and maintaining healthy performance levels in female athletes requires a scientifically grounded, gender-sensitive, holistic, and multidisciplinary approach.

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