Title:
Nature vs. nurture in determining athletic ability

Authors:
Xu Yan¹, Ioannis Papadimitriou¹, Ronnie Lidor², Nir Eynon¹

Affiliation:
¹ Institute of Sport, Exercise and Active Living (ISEAL), Victoria University, Australia
² The Zinman College of Physical Education and Sport, Wingate Institute, Israel

Running head:
Nature vs. nurture in athletes

Corresponding Author:
Nir Eynon, PhD
Institute of Sport, Exercise and Active Living (ISEAL),
Victoria University, Australia,
PO Box 14428, Melbourne, VIC, 8001
Phone: +61 399195615
Fax: + 61 399199480
E-Mail: Nir.Eynon@vu.edu.au
Abstract

This chapter provides a general discussion of the roles of nature and nurture in determining human athletic ability. On the nature (genetics) side, a review is provided with emphasis on the historical research, and several areas which are likely to be important for future research, including genome-wide association studies (GWAS). In addition, a number of well-designed training studies that could possibly reveal the biological mechanism ('cause') behind the association between gene variants and athletic ability are discussed. On the nurture (environment) side, we discuss common environmental variables including deliberate practice, family support, and the birth place effect, which may be important in becoming an elite athlete. Developmental effects are difficult to disassociate with genetic effects, because the early life environment may have long-lasting effects in adulthood. With this in mind, the Fetal Programming (FP) hypothesis is also briefly reviewed, as FP provides an excellent example of how the environment interacts with genetics. We conclude that the traditional argument of Nature vs. Nurture is no longer relevant, as it has been clearly established that both are important factors in the road to becoming an elite athlete. With the availability of the next generation genetics (sequencing) techniques, it is hoped that future studies will reveal the relevant genes of influencing performance, as well as the interaction between those genes and environmental (nurture) factors.
1. Introduction

1.1 Nature vs. Nurture or Nature & Nurture in sports?

Human athletic ability is influenced by a number of factors, such as environmental, physiological, psychological, and socio-cultural variables, and many others [1]. The term *nature vs. nurture*, initiated many years ago, refers to whether heredity (*nature*) or the environment (*nurture*) has a greater impact on human development (behaviour, habits, intelligence, aggressive tendencies, athletic performance, etc.). Possessing exceptional ability/abilities is, at least partially, attributed to one’s genes. Talent is passed down from parents or grandparents to the next generation, and this can include intelligence or athletic performance. In fact, many athletes from the past and today are members of the same family.

However, there is also evidence that talent is learned and earned through extended and intense practice of a skill, namely, *No pain, no gain*. This idea is encapsulated in a golden rule made popular by the writer Malcolm Gladwell in his book [2]. This “10,000 hours of practice” rule [3] is based on research by psychologist Anders Ericsson at Florida State University. The rule suggests that about 10,000 hours of dedicated practice in your particular field is sufficient to bring out the best in you. In essence, Ericsson’s theory suggests that sufficient practice in a particular skill can take anyone to a proficiency level equivalent to that of a top classical musician [3]. Ericsson and others don’t deny that genetic limitations, such as those of height and body size, can constrain expert performance in areas like athletic performance. They argue, however, that there is not enough evidence to pinpoint the genes that directly influence elite athletic performance.

Studies on twins (involving both identical and non-identical twins) and family members, which implicate genetics as a significant factor pertaining to athletic performance and adaptation to exercise training (even after adjusting for environmental factors), have shown that 30-80% of the variation in various performance traits (phenotypes) can be
attributed to genetics. An example is the well-known HEalth, RIsk, Training And Genetic (HERITAGE) family study (which is broadly discussed in the next section), in which a number of exercise-related phenotypes are to be influenced by both environmental and genetic factors. Hence, the current paradigm is that athletic ability, in particular at the elite-level, is influenced by components of both nature and nurture.

2. Genetics and Athletic Ability

2.1 Traditional twin and family studies

The genetic basis of athletic ability has been studied for a few decades. In the 1970s and 1980s, research into the effect of genetics on exercise and athletic was mainly conducted via twin and family studies. These studies demonstrated that genetics play a significant role in athletic performance and participation in exercise, even when adjusting for environmental effects [4]. More recently, a large-scale twin study composed of 37,051 twin pairs from seven European countries suggested that the heritability of participating in leisure-time exercise was between 48% and 71% [5]. A genome-wide linkage scan for athletic status reported a heritability of roughly 66% for athletic status in 700 British female dizygotic twin pairs [6]. In the HERITAGE Family Study, 742 healthy, sedentary participants, coming from families of white or black ancestry, underwent 20 weeks of standardised exercise training, and the heritability of several exercise-related traits was measured [7]. After adjustment for age, sex, body mass index (BMI), and baseline parameters, the heritability of the training responses for maximal oxygen uptake, submaximal exercise heart rate, and submaximal exercise capacity were estimated as 47% [8], 34% [9], and 26% [10], respectively. Muscle strength and mass have also been reported to be influenced by genetic factors. The estimated heritability for muscle strength and muscle mass, assessed by twin and family studies, varies from 31% to
78\%, with large differences between muscle groups, contraction velocities, and muscle lengths [11].

The twin and family studies have been important landmarks in the field of sports genetics, however, studies using this approach did not identify individual genes that influence performance. Following the completion of the Human Genome Project (HGP) in 2003, genetic research has evolved into more precise molecular DNA testing that is able to directly analyse the association between individual genes and athletic performance, and the hunt for sport gene/s was underway.

2.2 Identification of specific gene variants associated with elite athletic ability

Genetic variants are present throughout the human genome, and reflect differences in DNA sequence among individuals, groups, or populations. Genetic variants are common (they are the opposite of ‘rare’ mutations) and may result in different phenotypes in the same population. Just as specific variants can be associated with disease conditions, they may also cause an alteration in protein function that may be beneficial or detrimental for health and athletic performance. In studies that assess the association of those variants with athletic ability, the genotype frequency of a group of elite athletes (‘cases’) is compared with the frequency in a population of sedentary non-athletes (‘controls’). If one genotype is more related to elite athletic performance than the other genotypes, it will be found at a higher or lower frequency in elite athletes than in controls [12]. Several genetic variants have been found to be associated with elite performance in the last few years, and the current paradigm is that elite performance is a polygenic trait (i.e., influenced by many gene variants), with minor contributions of each variant to the unique athletic phenotype [13, 14].

Over 200 polymorphisms associated with exercise-related traits and over 20 polymorphisms associated with elite athletic status have been reported in the literature, and
are summarized on a yearly basis [15]. These polymorphisms are extensively discussed in Chapters 3 and 4 of this book. Of the polymorphisms associated with elite performance, the \textit{ACTN3} R577X polymorphism provides the most consistent results. \textit{ACTN3} (coding the a-actinin-3 protein) is the only gene that shows a genotype and performance association across multiple cohorts of elite athletes [16, 17], and this association is strongly supported by mechanistic data from an Actn3 knockout (KO) mice model. The angiotensin-1 converting enzyme insertion/deletion polymorphism (\textit{ACE I/D}) is the next promising polymorphism to be associated with elite athletic performance, but its association with elite performance is less consistent than the \textit{ACTN3} [18], and is yet to be supported by human/animal invasive (in vitro) models.

\textbf{2.3 Biological studies can provide insights into the effect of gene variants on performance: the Actn3 KO mouse model as an example}

A genetically engineered mouse, in which researchers have inactivated (knock-out), or over activated (overexpression) a certain gene, can serve as a tool for understanding the underlying mechanisms of skeletal muscle adaptations to exercise training. To gain a greater understanding of the effects of the \textit{ACTN3} R577X polymorphism on physiological and metabolic function at baseline (pre-training) and in response to exercise training, an Actn3 knockout (KO) mouse model has been developed, where KO mice that are completely deficient in the a-actinin-3 protein are compared to wild-type (WT) mice, who fully express the a-actinin-3 protein [19]. Compared with wild-type mice, Actn3 KO mice (that mimic the \textit{ACTN3} 577XX genotype in humans): 1) have reduced muscle mass due to decreased diameter of fast muscle fibres (where a-actinin-3 is primarily expressed); 2) show a significant decrease in grip strength; and, 3) have increased endurance capacity, with KO mice running 33\% further than their WT littermates at baseline [19, 20]. The absence of a-
actinin-3 protein results in a shift of muscle properties towards slower muscle fibre. Fast muscle fibres (type 2/X) from KO mice have significantly decreased anaerobic enzyme activity and increased oxidative/mitochondrial enzyme activity, without a shift in fibre-type distribution. Isolated KO muscles have longer twitch half-relaxation times and enhanced recovery from fatigue compared to the WT [19, 20]. Thus, the phenotypes of the Actn3 KO mouse mirror the gene association studies performed in humans, and provide a plausible explanation for the reduced sprint performance, and possible improved endurance performance in humans with the ACTN3 577XX genotype [21]. The shift in muscle metabolism towards oxidative metabolism could also account for the positive selection of the ACTN3 577X allele during human evolution; increased metabolic efficiency would increase tolerance to famine, and enhanced endurance capabilities may have conferred a survival advantage for hunter/gatherer societies. Recently, the calcineurin specific cell-signalling pathway was suggested for improved fatigue resistance and altered muscle metabolism. It was shown that calcineurin (a calcium-dependent protein phosphatase) activity was increased ~1.9-fold in Actn3 KO in mice, compared to their WT littermates [22]. Collectively, the data from the Actn3 KO model provide an example of how scientists can provide a biological explanation for the hypothesised effects of gene variants on athletic performance.

2.4 The future of studying genetics and athletic ability

As opposed to the hypotheses-driven approach of looking at one or a few targeted polymorphisms and assessing their association with athletic ability, an ‘unbiased’, whole-genome sequencing approach has recently come into use. In the last few years, the cost of using whole-genome sequencing methods has dramatically dropped and become more affordable, from the first complete human genome costing $3 billion to sequence in 2000 to ~$1000 per genome today. Advances in molecular technologies have enabled researchers to
apply genome-wide association (GWAS) approaches to the field. GWAS examines the association of genetic variation with outcomes or phenotypes of interest by analysing 100,000 to several millions of polymorphisms across the entire genome, without any predetermined hypotheses about potential mechanisms. GWAS has been successful in, among others, identifying novel genetic variants for age-related muscular degeneration [23] and type 2 diabetes mellitus [24]. GWAS of elite human athletic performance is ongoing, but there are no published papers to date.

Tightly-controlled exercise training studies are also required for understanding the gene x environment effect on the adaptive response to training. Perhaps the most prominent training study is the HERITAGE study, which has identified some people as ‘low-responders’ (with limited improved fitness following exercise training), and others as ‘high-responders’ to similar exercise training, in an attempt to discover the gene variants that contribute to this phenomena. Recently, based on data from the HERITAGE study, two GWAS studies were published, both using VO2max as an exercise response trait [25, 26]. In the first study, the authors identified genes associated with VO2max training response, based on global skeletal muscle gene expression profiling and DNA markers [25]. A total of 29 transcripts were strongly associated with the gains in VO2max, with 11 polymorphisms explaining about 23% of the variance in the VO2max training response. In the second study, a total of 39 different polymorphisms were associated with VO2max training response, with the strongest evidence of association observed in the first intron of the acyl-coA synthetase longchain family member 1 (ACSL1) gene [26]. Nine polymorphisms explained at least 2% of the variance, while seven contributed between 1 and 2% each. Together, sixteen polymorphisms accounted for 45% of the variance in VO2max trainability, a value comparable to the heritability estimate of 47% reported previously in the HERITAGE study [25].
The Gene SMART (Skeletal Muscle Adaptive Response to Training) study, established by our research group, is an ongoing effort to use a system biology approach to identifying the genomics (gene variants), transcriptomics (gene expression profile), metabolomics, and proteomics (proteins abundance) that predict the response to a single bout and to 4 weeks of high-intensity interval training (HIIT). We have currently recruited 40 volunteers who have completed the training phase, and the goal is to recruit a total of 250 moderately-trained, healthy participants (all males 18-45 y, BMI ≤30). Participants are undergoing exercise testing and exercise training in three different Centres (Victoria University, Australia; Bond University, Australia; and The University of Sao Paulo, Brazil), using a similar exercise program and a tightly-controlled nutrition program, with the aim of eliminating any environmental effect on the training response, and to be able to attribute the training-induced changes to the participants’ genes. Skeletal muscle biopsies and blood samples are being collected before, immediately after, and 3 hours post the first training session, as well as after the 4 weeks of training. It is anticipated that this research will contribute significant new information on the genetic basis of adaptation to exercise training, and will directly affect the elite performance environment.

3. Nurture's Influence on Athletic Ability

Besides nature, nurture (environment) also plays a big role in athletic ability. Five environmental variables are discussed in this section – deliberate practice (DP), family support, the coach's influence, relative age effect (RAE), and birthplace effect (BPE). These variables have been found to be associated with achieving a level of expertise in sport.

3.1 Deliberate practice (DP)

DP is one of the most studied environmental factors determining expertise in sport (e.g., [27-29]. DP has been defined as an activity aimed to enhance specific performance
components by providing feedback and furnishing opportunities for gradual refinement of skills – or their components – through the repetitious practice most suited for attaining the desired changes in performance and its mediating mechanisms [3, 30]. DP is conceptualized as an activity performed with full concentration, and is motivated by specific goals for enhancing performance. It is not the most inherently enjoyable of the available activities, and unlike work, it does not lead to immediate social or monetary rewards. In essence, DP is considered a task-specific, structured training activity that plays a major role in capturing the processes by which a skill is acquired [3].

It has been consistently argued that individual differences in performance largely reflect the difference in the amount of DP performed (e.g., [31]); that is, those who have achieved the highest level of human performance have devoted more time to activities specifically designed to enhance their performance in a given domain (see [32-36]). In other words, extensive DP is a major determinant of attaining expertise in any given domain.

Based on a series of studies on DP in athletic performance in both individual (e.g., gymnastics, swimming, and wrestling) and team (e.g., basketball, hockey, and soccer) sports, a number of observations have been made: (a) DP features, as described below, were more evident in athletes who met their goals and became highly skilled in their specific sport domain than in those who failed to reach the expertise stage [37-41]). Among the features of DP that were found in these studies to be associated with skilled athletes were: (1) expert performers in sport accumulated more hours of DP than non-expert performers (e.g., international athletes accumulated 8,000-9,000 DP hours compared with national players who accumulated 7,500 hours, and provincial players who accumulated ~ 5,000 hours); (2) experts had more organized training and individual instruction with their coaches than non-experts; and (3) experts devoted more time to off-court/field activities, such as imagery and watching videos of their own or others' performances, than non-experts. (b) A variety of
activities related to DP were reported by the athletes participating in the reviewed studies, among them on-court/field activities, learning decision-making skills, using imagery, and recreational activities. (c) A considerable variability in accumulated practice time over the years was observed in some of the studies, and therefore this variability must be addressed when examining aspects of DP. (d) Accumulated DP time increased throughout the athlete's developmental stages. It was observed that athletes devoted less time to activities associated with DP in early stages of talent development compared with the time they devoted to DP activities in the stages of specialization and expertise.

3.2 Family support

Individuals who have achieved the highest level of proficiency in their domain (e.g., athletes, musicians, and scientists) reported that in early stages of talent development their family played a major role in encouraging and supporting their involvement in training and competition [42]. Côté emphasized the importance of the family in early stages of sport involvement, particularly in the sampling stage, where children are involved in a number of sport activities [43]. In this early stage of development, it is expected that the parents will be part of the children's sport involvement, for example by enrolling them in different sport activities, by arranging transportation and access to the sport facilities, and by providing them with some basic instruction. In a number of retrospective studies (e.g., [40, 43]), athletes who achieved a state of expertise in their selected sports emphasized that they could not have fulfilled their athletic potential and become the best in their domain without the involvement of their parents and siblings.

Fraser-Thomas et al. offered a number of recommendations for parents who aim at helping their children to achieve, by showing them how to play a positive role in the young athlete’s journey to excellence [44]. According to the authors, parents should (a) create task-
oriented climates that are autonomy-supportive, (b) be particularly cautious of pushing their children, (c) teach and model life skills and positive sport participation, (d) consider how their involvement with the child-athlete influences other siblings, and (e) be aware of gender differences in their interactions with son and daughter child-athletes.

3.3 The coach's influence

While family members are expected to take the leading role in the young athlete's early stages of sport development, the coach takes over the role of the leading figure in advanced stages of talent development, serving as the professional authority for any aspect of the young athlete's training program. The ability of the coach to create an environment that fosters optimal learning is one of the most significant keys to athlete development [45]. Coaches play an important role in optimizing the athletes' practice time, in some cases having optimal control over the entire training program [46]. According to leading experts in the theory of training (e.g., [47], the structure and content of practice of those athletes who achieved the highest level of proficiency in their sport is superior to that of the athletes who did not reach the same level of performance.

While working with athletes in their early stages of talent development, coaches should provide them with high levels of instruction, support and encouragement, and management behaviors with an overall positive focus [48]. Based on the recommendations, coaches should create a learning climate where the young athlete will enjoy the sport activities, be motivated to become part of the sport program, and perform without fear of making errors. Coaches should also praise the athlete for making the required effort, and provide him or her with the relevant guidance if he or she has failed to appropriately perform the learned task.
As athletes advance in their skill development, coaches can focus on the technical-tactical aspect of training by using the current knowledge from different areas of sport and exercise sciences, among them biomechanics, motor learning, exercise physiology, sport and exercise psychology, and measurement and evaluation [47, 49]. During their journey to reaching the highest level of sport performance, athletes will benefit most from training programs in which knowledge from the above-mentioned sport sciences is synthesized. The better the synthesis among these bodies of knowledge, the higher the chances for the athlete to be prepared for practice and competition.

3.4 Relative age effect (RAE)

RAE implies that individuals who are relatively older than their peers in a given cohort/year (typically those who are born in the first quartile), or whose birthdate is closer to the cutoff date for their age group classification, are more likely to reach higher athletic achievements (see [50, 51]. In a series of studies on elite male individual (e.g., swimming and tennis) and team (e.g., baseball, basketball, and soccer) sports, it was consistently found that those athletes who were born early in the competition year had a higher representation than those who were born late in that year [52, 53]. Among the reasons suggested by researchers to explain the existence of the RAE in male elite performers in sport are: (a) the older athletes are probably more experienced than the younger athletes in various motor-physical abilities, such as balance, coordination, speed, and strength, and therefore they perform these sport skills better, and (b) the older athletes in their cohort/year are more likely to be selected to better teams, and therefore are provided with guidance and training superior to that given to the younger athletes, who practice with lower-skill level athletes and may be exposed to lower-quality training conditions [50].
Concerning RAE in female athletes, studies on basketball [54] and ice hockey [55] players showed that the RAE did exist; however, this effect reflected an over-representation of players in the second birth quartile compared to the over-representation in the first quartile found in male athletes. It was assumed by the authors of these studies that the lower rate of participation of females in sports and the lack of competition among them, compared with the higher rate of participation of males in sports and the emphasis of competition in male sports, were the reasons for the results obtained on RAE in female athletes.

3.5 Birthplace effect

Another environmental factor associated with achieving a high level of proficiency in sport is the place of birth – the birthplace effect (BPE), where he or she was born, and/or where he or she grew up during the developmental years, namely up to about 14 years of age [56]. Mixed results have been reported for the BPE. For example, early studies on BPE [52, 56] showed that male athletes who were born in cities of less than 500,000 people were more likely to play for professional leagues and attain a high level of sport competition than athletes born in larger cities. More recent studies showed different results: larger cities were just as effective in producing elite athletes as were smaller cities [53, 57].

Researchers reported that certain developmental opportunities exist that may be superior in smaller cities, such as (a) a greater amount of independent mobility and physical safety, (b) an integrative approach to sport participation involving schools, families, and the community at large, and (c) a more personal relationship between athletes and coaches [46, 56]. These conditions may provide young athletes with the most favourable conditions for developing their abilities and skills. However, it has also been argued that large cities can provide children with enhanced conditions, such as better-designed and better-equipped sporting facilities (e.g., arenas, fields, and swimming pools) and more experienced coaches.
3.6 The missing link – The need for a theory

In order to further increase our understanding of the contribution of task-enhancement environmental variables to athletic ability, additional research is needed on each of the discussed variables, as well as on the interaction between these variables. For example, while reviewing the literature on the contribution of the coach to the young athlete's development, it can be observed that the amount of available research that includes children under the age of 14 appears to be relatively limited compared to the body of literature using samples consisting entirely of athletes aged 14 years and above [48]. Therefore, additional studies are needed to explore the contribution of the coach to the young athlete's ability at the initial phases of sport involvement (e.g., ages eight to 14). Considering the influence of the family on children's involvement in sport, future work should acknowledge and prioritize the dynamic and changing roles of parental involvement throughout the child-athletes' development, as the enormity of the changes that occur from the child's sport initiation through adolescent levels of expertise cannot be underestimated [44].

While investigating each of the discussed variables, researchers should also examine the interaction that occurs between these variables. For example, the relationships between the parents of the young athlete and his or her coach should be studied throughout the different stages of talent development. In order to provide the young athlete with the optimal preparation – physical, psychological, and emotional, both parents and coaches should be aware of the needs of the athletes and cooperate with each other in helping them cope with the multifaceted challenges they may face during their long-term athletic journey. Therefore, more information is needed on how to establish effective cooperation between parents and coaches, which ultimately will help the athletes advance from one stage of athletic development to another. In essence, the conduction of additional quantitative and qualitative
inquiring will result in a better understanding of the contribution of environmental variables to athletic ability.

4. Fetal Programming and Epigenetics: An Example of How our Environment can Influence our Genes

Fetal programming, also known as developmental programming or the Barker hypothesis, pertains to the fetal origins of adult diseases. This hypothesis is based on epidemiological data showing that low birth weight due to maternal malnutrition has long-term effects on adult health [58]. The fetal programming hypothesis suggests that the alteration in the uterine environment as a result of nutritional stress at certain stages of conceptus growth and development might permanently change tissue structure and function [59]. The offspring of malnourished mothers have a higher chance of developing coronary heart disease, stroke, diabetes, and hypertension [60]. The prevalence of type-II diabetes (T2D) increases 3-fold for men who weighed 5.5 lb or under at birth when compared to those who had a birth weight of at least 9.5 lb [61]. The correlation between human maternal nutrition and offspring birth weight, and the subsequent development of hypertension, insulin resistance, T2D, and cardiovascular diseases, has been well studied by many researchers [61-63].

Animal studies have shown that both maternal under-nutrition [64] and over-nutrition [65, 66] during gestation affect skeletal muscle development. Low birth weight is associated with decreased voluntary physical activity in adult rats [67, 68]. In humans, low birthweight is associated with higher adult adiposity and decreased lean body mass [69]. Overall, these animals and humans studies suggest that the environment can affect our genes, and traits such as athletic ability not only depend on the genetic code, but also on epigenetics [70, 71].
Epigenetics describes heritable changes in gene function without changes in gene sequence [72]. Epigenetic changes can pass from one cell generation to the next (mitotic inheritance), as well as from one generation of a species to the next (meiotic inheritance) [73]. Epigenetic modifications include DNA methylation, histone modification, and microRNAs, which explain the cell differentiation into different cell types with various phenotypes despite their having the same DNA sequence [73]. Interestingly, epigenetic status can be influenced by environmental factors, among them nutrition and exercise training [70]. A recent study showed that maternal overnutrition during gestation led to different epigenetic profiles in fetal skeletal muscle in animals [71]. The influence of exercise training on epigenetics signals will be discussed more fully in Chapter 10 (Perikless).

4.1 Conclusions

We conclude that athletic ability is influenced by both nature and nurture, and with recent progress in sports genomics research into what makes an elite athlete, using the Nature vs. Nurture argument is no longer applicable. We note that the identification of novel genetic variants associated with performance is only the “tip of the iceberg” in this field, and that the implications of training and environment must also be considered. While research in this area has progressed in the last few years, the greatest challenge would be to combine these two fields together and create models that will take into account both genetics and environmental factors. Achieving this would potentially assist in the development of a meaningful tool for coaches and exercise scientists to use in order to personalise exercise training at the highest standard.
References


47. Bompa TO, Buzzichelli C: Periodization training for sports: Champaign : Human Kinetics, [2014]


