Spring 2023

Using Genetic Testing in Order to Improve Retention Rates in Youth Sports

Madison Nelson
Ohio Wesleyan University

Using Genetic Testing In Order To Improve Retention Rates In Youth Sports

By: Madison Nelson

April 2022

Introduction:

According to the Sports and Fitness Industry Association (SFIA), from 2008 to 2018, the number of kids ages 6 to 12 participating in an organized sport regularly decreased by 7%. This may be due to cost, commitment, or the increasingly competitive environment of youth sports. However, out of the 30 million children participating in sports, upward of 3.5 million children, ages 14 and younger, experience injuries (stanfordchildrens.org). Experiencing an injury may warrant not wanting to participate anymore. Some of the most common injuries in children are overuse injuries. These can be caused by single-sport specialization, or training too much to the point where your body can no longer handle it. Research has shown that genetics play a role in athletic ability, but genetics may also play a role in the prevention of injury. Evidence has shown that some genes may affect one's susceptibility to injury and/or the severity of the injury. Genetic testing may decrease the number of children experiencing overuse injuries, therefore, decreasing the number of children who quit participating in sports due to injury.

In this paper, I will first present why the debate of nature versus nurture is outdated in order to show that genetics and practice both play a role in athletic ability. I will also present two genes, the α-actinin-3 and the Insulin-like growth factor gene, that may affect athletic ability. Then I will discuss why children drop out of sports, one reason being overuse injuries. Common types of overuse injuries and potential genes to test for will be discussed. This paper aims to answer the question, can genetic testing help prevent injuries in youth athletes and help maintain or improve the number of children participating in sports?

Nature versus Nurture:

The nature versus nurture debate has been at the forefront of giftedness for a long time. Some even argue that it dates back to the 1800s. On the nature side of the debate, scientists argue that giftedness, in this case, athletic ability is due to genetics. On the other hand, the nurture side of the debate credits athletic ability to your upbringing and environment, taking on the "practice makes perfect" attitude. As an example, in the National Hockey League (NHL) is, whom some may argue to be the best player ever, Connor McDavid. Connor McDavid has led the NHL in points per season in 5 of the past 7 seasons, and has only been in the NHL for 8 seasons. McDavid is known for his speed and agility on the ice. Has he gained his athletic ability from practicing so much or was he just born to be fast? On the nature side of the debate, it is possible that McDavid has the optimal genetic makeup for speed. On the nurture side, it is possible that McDavid has practiced more than the average player and really focused on improving speed and agility. Genetic testing might show if McDavid has any genetic mutations that may play a role in his skating abilities. However, it may also show that McDavid does not have any genetic mutations, which would credit his athletic ability to practice. McDavid's ability may also be examined without genetic testing by looking at the number of hours he has trained and comparing it to other players.

Genetics play an important role in innate abilities of athletic performance. However, nurture is still relevant in the argument. Yan et al. (2016) examined current genetic testing techniques and the environment of young athletes to determine that the argument of nature vs. nurture is outdated. Yan et al. (2016) suggested that genetics and environment work together to create elite athletes, and the debate of nature versus nurture should be left in the past regarding athletics.

On the nurture side of the debate, Yan et al. (2016) discuss the environment in which the athlete is developed, including deliberate practice, family support, and the birthplace effect. The birthplace effect (BPE) is described as where the athlete was born and grew up. Yan et al. (2016) relate this to the city size in which the athlete was born. Yan et al. (2016) include a study on BPE showing that male athletes born in cities with fewer than 500,000 people were more likely to become professional athletes playing at a high level of competition, as opposed to those in larger cities. In smaller cities, there may be a higher encouragement for sports, larger independent mobility and safety, and a more personal relationship between the athlete and the coach (Yan et al., 2016).

Another example of BPE is seen in African long-distance runners, specifically in Kenya. Haile et al. (2019) conducted a study comparing the oxygen-carrying abilities of runners in Kenya (2150 meters above sea level) and runners in Scotland (at sea level). This study assessed runners by examining their VO_{2max} (oxygen uptake), the performance of a timed trial, hematocrit (HCT), and hemoglobin concentration (HGB). The Kenyan runners had a higher VO_{2max}, a faster timed trial, and higher levels of HCT and HGB at the start of the experiment. The runners then received injections, every 2 days for 4 weeks, of recombinant human erythropoietin (rHuEpo). This drug is known to increase the amount of oxygen available for muscles, increasing the HCT and HGB (Haile et al., 2019). After the 4 weeks and the final injection, the Scottish and Kenyan groups had similar HCT and HGB levels, and the VO_{2max} and timed trial performance showed a similar increase in both groups. However, the HCT and HGB levels in the Kenyan group did not increase as much as in the Scottish group, showing that the rHuEpo administration did not have as much of an impact on the Kenyan group (Haile et al., 2019). This is another example of the BPE because it shows that those who grew up, or even just trained, at higher altitudes may have an advantage as their body is more equipped to take up oxygen in aerobic sports, like long distance running.

Deliberate practice is one of the most studied environmental factors. Yan et al. (2016) define deliberate practice 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” (10). Deliberate practice may not be the most enjoyable activity for the athlete as it may not provide any immediate social or monetary reward and is driven by specific goals for increasing the quality of performance. One of the most famous examples of deliberate practice is the 10,000 hour rule. The 10,000 hour rule explains that the 10,000 hours dedicated to practice in the field of interest is necessary to make you the best of your ability (Ericsson et al., 1993). While sports have limitations, like height and body size, deliberate practice may be enough to overcome these deficiencies and still reach a high level. However, Ericsson et al. (1993) note that “deliberate practice requires available time and energy for the individual as well as access to teachers, training material, and training facilities (the resource constraint)” (368). There are constraints to the amount of practice that can be considered deliberate depending on the athletes’ access to training material, the coach's time, and transportation. Therefore, the achievement of 10,000 hours of deliberate practice may not be attainable for everyone.

Genetics for athletic performance:

It is important to understand that both practice and genetics play a role in the formation of an elite athlete. The α-actinin-3 and the Insulin-like Growth Factor genes have been studied in depth to show that they may influence athletic performance. Later in this paper, both genes will be discussed again for injury prevention and the types of injuries they may be associated with.

α-actinin-3

A gene that has been thoroughly researched in the field of genetics and athletics is α-actinin-3. In the realm of athletic performance, α-actinin-3 is best known as a skeletal-muscle actin-binding protein associated with fast glycolytic muscle fiber used in sprinting and explosive movements (Yang et al., 2003). These muscle fibers are also known as type-II or fast-twitch fibers. In a study assessing the relationship between sprint athletes and the presence of α-actinin-3, Yang et al. (2003) found that the α-actinin-3 protein is absent in 18% of healthy white males because of homozygosity for a common stop-codon polymorphism in the α-actinin-3 gene, R577X, with the 577R allele being the functional allele. α-actinin-3 is expressed in fast-twitch myofibers responsible for generating force at high velocity. Therefore, this study chose to look at elite male and female sprinters. This study found that male and female elite sprint athletes have significantly higher frequencies of the 577R (functional) allele than the control groups, showing a higher presence of α-actinin-3 (Yang et al., 2003). In conclusion, α-actinin-3 may benefit the function of skeletal muscle in generating forceful contractions at high velocity, which may create an advantage for sprinting athletes. This study performed on the polymorphisms of the α-actinin-3 gene shows the possibility of genetics playing a role in the performance of athletes involved in sports that require explosive or fast, powerful movements. The findings of Yang et al. (2003) are supported by studies replicating the experiment in elite strength athletes in the United States (Roth et al., 2008), Taiwanese elite sprint swimmers (Chui et al., 2011), and Italian gymnasts (Massida et al., 2009). However, it should be noted that there are studies that have found no correlation between the α-actinin-3 R577X polymorphism and athletic performance.

Insulin-like Growth Factor (IGF)

More recently, a mutation in the gene that codes for Insulin-like Growth Factor (IGF) has been proposed to affect athletic ability. The IGF is a hormone that controls the secretion of growth hormones in your body in order to promote the healthy, normal growth of bones and tissues in the body. Ben-Zaken et al. (2021) suggested that polymorphisms in the IGF increase the odds of becoming an elite short-distance runner. A genetic score (IFG-GS) was used based on 6 polymorphisms related to the insulin-like growth factor axis in Israeli runners and swimmers at the top and national levels. With an IGF-GS> 25, there is an increased odds ratio of becoming an elite short-distance runner. This study shows how the IGF system might affect land speed sports but not long distance swimming as there was not a high difference in the IGF-GS between top and national level swimmers (Ben-Zaken et al, 2021).

As shown in the paragraphs above speaking about IGF-1 and ACTN-3, it is useful to understand the genetic makeup of athletes. This information may be useful to understand when it comes to creating the perfect environment for the young athlete. When concerned with athletes' success and injury prevention, deliberate practice and genetics play an equal role. However, genetic testing may allow for a more precise measurement of the athlete’s weaknesses. It is possible that specifically identifying the athlete’s genetic predispositions will allow for more targeted training to prevent injury. As mentioned above, deliberate practice is important when creating elite athletes but also important in preventing injury. Working on the muscles that are naturally weak and may be more prone to injury may be useful to focus on as a part of training for preventative measures. If the athlete is injured, then the athlete may not be able to compete at all.

Ethics of Genetic Testing:

The study of genetics for athletic performance and injury prevention is fairly new and at an early stage of understanding. In order to determine how to create the best environment for athletes, it is important to discuss the ethics of genetic testing. Because this study is discussing the use of new technology on children, it is especially important to discuss the ethics of genetic testing. Vlahovich et al. (2016) discuss the possibility of genetic testing in Australian athletics. In this study, the authors explain that genetic testing should not give an athlete an advantage and that genetic testing should be used if only available to all.

Vlahovich et al. (2016) go on to state that individuals under eighteen years of age should not be genetically tested. However, this source fails to take into account the key development of children that begins before the age of eighteen. The use of genetic testing for children, with parental consent, may allow the athlete to focus on their strengths and weaknesses while developing. It is also important to note that genetic testing may not be available to all athletes, so this development would go toward those who are using it to improve private lessons.

It may be beneficial for leagues to create some rules or regulations surrounding it to ensure that all athletes have a fair chance at being successful. It should also be noted that genetic testing should never be done against the athletes' wishes. It should not be a requirement, and it should be understood that it will not make the athlete more successful. It will only allow for more productive practices and will affect the amount of time spent on injury-prevention techniques.

The decline of youth sports:

In the United States, the number of children involved in sports is declining each year, alarming sports psychologists, youth development scholars, and practitioners. Dangi and Witt (2016) explain that from 2008-2013 there was a 4.5% decline of children involved in team sports, with most children dropping out by the time they reach the age of 12 or 13. Similar trends with children in youth sports were seen around the world. Dangi and Witt (2016) grouped the reasons why children did not return to sports into three categories: intrapersonal, interpersonal, and structural constraints.

Intrapersonal constraints include the issues of not having fun, anxiety and nervousness caused by excessive criticism, and pressure from coaches, or not having a close relationship with coaches. It is important to note that all of these factors may overlap. Lack of fun could be due to anxiety and pressure placed on the athlete. Dangi and Witt (2016) include the statistic from a study performed by Kelley and Carchia (2013), "38% of girls and 39% of boys suggest that lack of fun is the biggest reason for dropping out of sports" (192). When children experience anxiety and pressure, they no longer find joy in the sport that they once did. Also, coaches develop different coaching styles. A more authoritarian coach may not be the best environment for the child and may cause an uncomfortable environment for the child.

Interpersonal constraints include parental pressure, loss of ownership, and time constraints. When children are introduced to sports, it is often through their parents. Therefore, children may feel the need to continue participating in sports due to pressure from their parents. Parents may begin to place more pressure on their children because their children are fulfilling their fantasies, or they may benefit from the success of the child, taking away the enjoyment of the sport. This may also lead to a lack of ownership felt by the child, resulting in a loss of motivation. Children who are spending a lot of time participating in sports may also feel that they do not have enough time to participate in other age-appropriate activities that are crucial to the development of the child socially, again leading to a loss of motivation.

Lastly, structural constraints include sports-related injuries, overuse/burnout, not being given playing time, feeling that participation is too structured, and financial constraints. Kelley and Carchia (2013) estimate that approximately 2.7 million kids under 20 were treated for sports-related injuries. This statistic includes a large increase in head injuries, especially an increase in concussions. From Williams (2016), "Each year, more than 3.5 million young athletes experience a sports injury severe enough to warrant medical attention, …and approximately 66% of these injuries are serious enough to require attention in the emergency department." When kids receive injuries serious enough to warrant medical attention, it can be traumatic for both the athlete and the parents. Some kids may want to return to the sport when they recover, but many do not. Overuse and burnout may cause injuries as well. The repetitive use of the same muscles and body parts can lead to overuse injuries caused by the pressure felt to constantly be practicing.

Overuse injuries:

A rise in the intensity of youth sports has also caused a rise in sport specialization. Sport specialization is when a child decides, or in some cases, the parent decides for the child, that they want to only train in one sport. This often means quitting all the other sports they have previously participated in and beginning to train year-round. The age at which a child specializes in one sport and begins intense training may increase the risk of overuse injuries. Jayanthi et al. (2019) report that the recent rates of sports specialization have ranged from 17-41%. These rates were affected by factors like sex, age, sport, socioeconomic status, school size, and geographic location. Jayanthi et al. (2019) explain that possible consequences of sport specialization at a young age are psychological burnout and an increased risk for overuse injuries. Athletic trainers and other physicians involved in youth sports are concerned about sports specialization and overtraining as past research has shown that high degrees of specialization have been linked to high rates of overuse injuries like strains, Osgood-Schlatter disease, patellofemoral pain, and Sinding-Larsen-Johansson syndrome (Jayanthi et al., 2019). Research has also shown that specialization at a young age can lead to more severe overuse injuries, which result in the loss of one or more months of training, including spondylolysis, osteochondritis dissecans, elbow ligament injuries, and stress fractures were noted more in those who specialized in individual sports. More serious injuries were found in children training for more hours a week than their age in years (Jayanthi et al., 2019).

It is possible that the type of sport and training required in individual sports, like tennis, gymnastics, and dance, pose a higher risk for overuse injuries. Individual sports also specialize earlier than those in team sports. Pasulka et al. (2017) examined the patterns of specialization across youth sports and the relationship to injury risk. Pasulka et al. (2017) analyzed the cases of injured athletes from ages 7-18 after visits to sports medicine clinics. Patients were also asked to fill out a survey about age, gender, sport, specialization patterns, and other information regarding sports-related injuries in the last 6 months. The survey included questions like, “Can you pick a main sport?; Did you quit other sports to focus on a main sport?; and Do you train >8 months in a year?” . The patients' answers were then scaled on a spectrum of low, moderate, and high degrees of specialization. Pasulka et al. (2017) make an important note that the ‘highly specialized’ group may also contain athletes that participate in more than one sport, but were able to identify a single sport as the ‘main sport.’

Injury diagnoses were able to be gathered from medical records, and the injuries were classified as acute, overuse, or serious overuse. Pasulka et al. (2017) acknowledge that there may be neuromuscular differences that may contribute to injury risk in those that participate in more than one sport but were able to identify one as a 'main.' The results of this study show patterns of injury based on sport type. Sports with contact or collision are at a greater risk for acute injuries. Individual sports athletes are at a larger risk for overuse and serious overuse injuries. The results showed that the sports with the highest overuse injuries were tennis, baseball/softball, and volleyball, and the sports with the highest serious overuse injuries were tennis, gymnastics, and dance. Those in individualized sports also had a higher number of weekly hours spent training, contributing to the risk of overuse injuries.

The presence of overuse injuries in the Jayanthi et al. (2019) and the Pasulka et al. (2017) studies show that there is a need to prevent such injuries. In order to keep youth in sports, there must be a way to prevent burnout and overuse injuries as shown in the Dangi and Witt (2016) study. While some may say practice less, others may say that you need to practice in order to reach a higher level. Incorporating genetic testing in youth sports may allow for the improvement of skills in less practice. Genetic testing may allow those to understand not only their own strengths, but also their weaknesses. A possible cause of the overuse injuries may be a repeated use of a weak muscle that is caused due to a genetic predisposition. Focusing on strengthening this weakness could reduce overuse injuries and reduce the number of children dropping out of sports. Incorporating genetic testing may also allow for further education of athletes, parents, and coaches in order to prevent more overuse injuries. Genetic testing may also provide a better opportunity for more efficient recovery from injuries.

Before getting into the specific genes that would be useful to test, it is important to know if athletes and coaching staff will find this information helpful. A study conducted by Goodlin et al. (2015a) found 124 single-nucleotide polymorphisms (SNPs) associated with anterior cruciate ligament tears, Achilles tendon injuries, low bone mineral density and stress fractures, osteoarthritis, vitamin/mineral deficiencies, and sickle cell trait. This study conducted a program with 14 triathletes using SNPs to give the athletes further knowledge about their specific genetic makeup and possible deficiencies that would make them more susceptible to injuries. The athletes were then educated about how their genetic makeup affected the risk of obtaining a sports-related injury. The athletes were each given an hour-long consultation. After their consultation, athletes said it was informative, and most were going to incorporate their results into their training (Goodin et al., 2015a). The athletes were then given 3 and 12 month follow-up questionnaires. Of those that responded to the questionnaires, all gave an overall positive response, and thirteen explained that they had used their genetic results as useful and informative. All except for one athlete reported sharing their genetic information with coaches, trainers, physicians, and some reported sharing with teammates. This shows that genetic information may be useful to athletes in a number of ways. Lastly, 8 out of 12 athletes that were recommended changes based on their genetics modified their training and reported positive results.

Table 1.

Gene of Interest	What the gene codes for	Mutation	What this mutation affects	Injury type
α-actinin-3 (ACTN3)[1,7]	skeletal muscle protein α-actinin-3	SNP causing a premature stop codon (X allele)	XX effects the amount of α-actinin-3 produced, X allele presence will have an increase in cortisol after eccentric loading	Hamstring injuries, increased muscle fatigue, increased severity of injury
Insulin-like Growth Factor 2 (IGF2)[2,8]	the expression of the protein, insulin-like growth factor 2 (IGF2)	Single nucleotide polymorphism G/C, heterozygous vs homozygous	soft tissue growth, response to degeneration and regeneration in response to injury, CC was less severe than GG	The degree of injury, injury recovery time in soft tissue injuries (contusions, sprains, tendonitis)
Collagen, type I, alpha I (COLIA1)[3,9]	the alpha chain of type I collagen	polymorphisms (intron I) affect the level of expression, G, T or TT	T allele leads to increased expression which may cause increased strength in tendons and ligaments	ACL tears, Achilles tendinopathy, Shoulder dislocations and ligament ruptures
Tenascin C gene (TNC)[4,6]	Glycoprotein Tenascin-C, which is found in the ECM in a variety of tissues including tendons	Polymorphisms causing GT repeats	The number of GT repeats may affect the strength of the tendon, 12 or 14 (even) repeats may cause weakened tendons	Achilles tendon injuries
Vitamin D receptor VDR[5,10]	FokI polymorphism in exon 2, BsmI polymorphism in exon 9	FokI (FF vs Ff and ff) and BsmI (BB and Bb vs bb)	maintaining bone health, f containing groups were at a higher risk for stress fractures than F, B containing groups were at a higher risk for stress fractures than b	stress fractures and bone density

1.) Pickering, C., & Kiely, J. (2018). Hamstring injury prevention: A role for genetic information?. *Medical hypotheses*, *119*, 58–62. https://doi.org/10.1016/j.mehy.2018.07.011 2.) Pruna R, Artells R, Ribas J, et al. Single nucleotide polymorphisms associated with non-contact soft tissue injuries in elite professional soccer players: influence on degree of injury and recovery time. *BMC Musculoskelet Disord.* 2013;14:221. 3.) Khoschnau S, Melhus H, Jacobson A, et al. (2008). Type I collagen alpha1 Sp1 polymorphism and the risk of cruciate ligament ruptures or shoulder dislocations. *Am J Sports Med.* 36:2432–2436. 4.)Maffulli, N., Margiotti, K., Longo, U. G., Loppini, M., Fazio, V. M., & Denaro, V. (2013). The genetics of sports injuries and athletic performance. *Muscles, ligaments and tendons journal*, *3*(3), 173–189. 5.) Chatzipapas C, Boikos S, Drosos GI, et al. Polymorphisms of the vitamin D receptor gene and stress fractures. Horm Metab Res. 2009;41:635–640. 6.) Mokone, G. G., Gajjar, M., September, A. V., Schwellnus, M. P., Greenberg, J., Noakes, T. D., Collins, M. (2005). The Guanine-Thymine Dinucleotide Repeat Polymorphism within the Tenascin-C Gene is Associated with Achilles Tendon Injuries. *The American Journal of Sports Medicine*, 33(7,) 1016-1021.https://doi.org/10.1177/0363546504271986 7.) Deuster, P.A., Contreras-Sesvold, C.L., O'Connor, F.G. *et al.* Genetic polymorphisms associated with exertional rhabdomyolysis. *European Journal of Applied Physiol*ogy 113, 1997–2004 (2013). https://doi.org/10.1007/s00421-013-2622-y 8.)Pruna, R., Artells, R., Lundblad, M., & Maffulli, N. (2017). Genetic biomarkers in non-contact muscle injuries in elite soccer players. *Knee Surgery, Sports Traumatology, Arthroscopy*, *25*, 3311-3318. 9.) Goodlin, G. T., Roos, T. R., Roos, A. K., & Kim, S. K. (2015b). The dawning age of genetic testing for sports injuries. *Clinical journal of sport medicine : official journal of the Canadian Academy of Sport Medicine*, *25*(1), 1–5. https://doi.org/10.1097/JSM.0000000000000158 10.) Lorentzon, M., Lorentzon, R., & Nordström, P. (2001). Vitamin D receptor gene polymorphism is related to bone density, circulating osteocalcin, and parathyroid hormone in healthy adolescent girls. *Journal of bone and mineral metabolism*, *19*(5), 302–307. https://doi.org/10.1007/s007740170014

Common Injuries:

Hamstring Injuries

An experiment using data from the 2016/2017 English Premier League football season shows that hamstring muscle group injuries accounted for 27% of injuries (Pickering & Kiely, 2018). Hamstring injuries are the most prevalent non-contact injuries in competitive athletics, with a typical recovery in the range of 8-73 days, depending on the severity. Hamstring injuries commonly occur during rapid deceleration, which causes tension in the hamstring. Hamstring injuries have been shown to be associated with age, low levels of eccentric strength, muscle fascicle length, and previous injury history. With this being known, Pickering and Kiely (2018) explain that research is trying to solve the questions of how to identify those who are most at risk of hamstring injuries and how to best design training to most productively enhance hamstring resilience. While known factors play a role in the risk of hamstring injuries, Pickering and Kiely (2018) have explained that it has been hard to develop accurate testing with true predictive values. There are exercises that work to prevent injuries, like the Nordic hamstring curl and the yo-yo hamstring curl, but issues with execution and compliance have shown to be problematic. This may also be due to the amount of soreness they may cause and the perceived lack of effectiveness. This shows a promising potential for genetic testing. While there are benefits to doing these exercises, those with the genetics for naturally stronger hamstrings may not need to spend time and experience the soreness that is caused by these exercises. Genetic testing will also be able to provide an answer as to whether the athlete is susceptible to hamstring injuries and needs to work on these exercises more extensively. Further research into genetic testing may be able to show the extent to which gene variants will modify hamstring injury risk. Future research should also aim to test whether or not the modification of training based on genotype will lead to better hamstring resilience.

Tendinopathies:

In an attempt to understand age-related injuries in youth sports, Le Gall et al. (2006) examined elite French youth soccer players. This study focused on the age groups of younger than 14 (U14), younger than 15 (U15), and younger than 16 (U16), and tracked the injuries sustained for 10 seasons. The injuries were categorized and documented by type, location, severity, date of occurrence, and position played. The results of the study showed that more injuries were sustained in practice than in matches. The data showed an average of 2.2 injuries per player was sustained across all three age groups. The youngest group (U14) had the highest number of injuries. The youngest group also had the highest number of injuries during training. Although there was no significant difference in the severity of the injuries, they were evenly spread across the age groups, and there was a difference in the type of injuries. The most prevalent type of injury was overuse injuries, with 17.2 percent. A majority were tendinopathies, accounting for 9.4%. The knee was the most common location for tendinopathies, followed by the adductors and the Achilles tendon. The U14 group had the highest number of overuse injuries. The U14 age group also had the highest number of reinjuries. Overall this study found an injury rate of 4.6 to 5.2 per 1000 hours of playing exposure (Le Gall et al., 2006).

However, there is a limitation to this study. The children in the U14 category may be experiencing more overuse injuries because they are also in the peak height velocity period (PHV), also known as a growth spurt (lbsm.co.uk). As the child grows, they may become more prone to injuries, especially overuse injuries. If the child is training a lot, they may suffer from more overuse injuries during this period in time, which for boys is around the ages of 12-14 years old, simply because they are growing at a fast rate (lbsm.co.uk).

Anterior Cruciate Ligament (ACL) tears

In an attempt to evaluate the injury type, location, and severity in young children (5-12 years) and older children (13-17 years), Stracciolini et al. (2013) used random sampling to gather data on pediatric injuries from medical centers from 2000 to 2009. This study found that overall the older category sustained more injuries. The results show that half the younger group was treated for overuse injuries, while the other half was treated for traumatic injuries. The older group was treated for a larger amount of overuse injuries as opposed to traumatic injuries, and more injury to soft tissue was seen. Also, more of the overuse injuries from the older group required surgery (Stracciolini et al., 2013). ACL tears were the most reported injuries for the older group. ACL tears were also seen in the younger group, but not as substantial as in the older age group. The younger group was treated more for osteochondritis dissecans (OCD), fractures, and apophysitis. The results of this study also show a higher number of females than males being treated for lower extremity and spinal injuries. The authors suggest that the high number of ACL tears can be reduced by strengthening the ACL and surrounding muscles during adolescence before neuromuscular deficits and peak knee injuries occur during growth.

The number of tendinopathies (Le Gall et al., 2006) and ACL tears (Stracciolini et al., 2013) in adolescent children suggest that this is the prime age to test for genetic differences or deficiencies that may impact the child’s risk for overuse injury. This may also allow the child to practice strengthening any genetic predispositions to avoid reinjury or even lessen the injury's severity and recovery time. Testing during puberty may also be able to predict whether or not the child will experience injury or pain due to growth. An example of this type of injury is Osgood-Schlatter disease and other knee tendinopathies.

Genes to test for:

α-actinin-3

The α-actinin-3 gene plays a role in the fast twitch myofibers responsible for explosive movements (Yang et al., 2003). The α-actinin-3 is known for having a beneficial effect in sprinting due to the generation of forceful contractions at a high velocity. However, an SNP resulting in a premature stop codon (X allele) may also be beneficial to test for when it comes to injuries (Pickering & Kiely, 2018). α-actinin-3 is also expressed in type-II muscle fibers, ultimately playing a role in the decrease of inflammation and muscle damage. Those with the XX genotype cannot produce enough α-actinin-3 and may be more susceptible to inflammation and muscle damage. Those who carry the X allele will also have a larger increase in cortisol after eccentric loading and lower levels of testosterone, which may limit the adaptability of the muscles. An example of this is damage from eccentric contractions, like those used in hamstring curls, due to the lack of α-actinin-3 causing weaker z-lines in type-II fibers. Those with the XX genotype may also experience greater levels of soreness and greater injury severity due to the fewer α-actinin-3 proteins aiding with recovery (Pickering & Kiely, 2018).

A study assessing exertional rhabdomyolysis (ER), which is a breakdown of skeletal muscle cells caused by strenuous exercise and various forms of physical training, typically presents in otherwise young, healthy individuals (Duester et al., 2013). Duester et al. (2013) had similar findings to Pickering and Kiely (2018). Duester et al. (2013) examined the R577X polymorphism of the α-actinin-3. Individuals with the XX genotype were at greater risk for ER than those with the RX and RR genotype. Therefore, as shown in Table 1, testing for a deficiency in this may provide the athlete with information on injury susceptibility due to muscle fatigue or the severity of the injury.

IGF-2

The IGF2 gene may play a role in tissue repair. This is important for the recovery of the athlete. SNPs regarding the heterozygosity or homozygosity of guanine (G) and cytosine (C) pairs can determine the amount of activity of IGF2 (Pruna et al., 2013). IGF2 is responsible for soft tissue growth, response to degeneration, and regeneration in response to injury. The homozygosity of the CC genotype was less severe than the GG genotype. The SNP of the GG genotype led to more severe injuries and a longer recovery time due to a reduction in IGF2 produced. The heterozygous (GC) genotype had the least severe muscle injuries (Pruna et al., 2013). Since IGF2 impacts soft tissue injuries like sprains and tendonitis, which are common overuse injuries, it is an excellent candidate for genetic testing (Table 1). After publishing their study in 2013, Pruna et al. (2017) wanted to validate the previous results of their study. Pruna et al. (2017) assessed non-contact muscle injuries in elite soccer players. This study found that the GC genotype was associated with less severe injuries than the CC and GG genotypes, aligning with the study performed in 2013.

Testing for this gene may aid in creating an optimal treatment plan if the athlete becomes injured. IGF2 testing may also be used to set limitations on the amount that the athlete can practice or train outside of the typical hours required by the team. It may limit the number of hours they can receive personal training or the number of hours of training in general.

Collagen, type I, alpha 1 (COLIA1)

The COL1A1 gene plays a role in the tensile strength of tendons and ligaments (Goodlin et al., 2015b). This gene encodes the alpha chain of type I collagen, the most prevalent protein in all tendons and ligaments. An SNP in the upstream region is responsible for the expression levels of the protein and may contribute to the strength of the tendon or ligament. The SNP determines whether there is a T or a G nucleotide at this spot. While most carry a G, about 20% of athletes carry a T, which has been shown to contribute to a larger number of type I collagen alpha polypeptides expressed. About 4% of athletes also have a homozygous TT genotype, showing significantly less susceptibility to tendon and ligament injuries, like ACL tears or Achilles tendinopathy.

This gene also may play a role in other cruciate ligament tears and shoulder dislocations, which are most often caused by trauma (Khoschnau et al., 2008). Khoschnau et al. (2008) examined the COL1A1 gene in relation to cruciate ligament ruptures and shoulder dislocations. The findings of this study aligned with the findings of the Goodlin et al. (2015b) study. The TT genotype was associated with a substantially reduced risk for cruciate ligament ruptures and shoulder dislocations. Khoschnau et al. (2008) also note that previous research has shown that athletes are more likely to experience one of these injuries if they have a sibling who has experienced the same injury prior. This shows that there is potential for a genetic cause to increased injury risk.

Tenascin C gene (TNC)

Glycoprotein Tenascin-C is most commonly found in the extracellular matrix in a variety of tissues, including tendons (Maffuli et al., 2013). Tenascin-C plays a role in regulating cell-matrix interactions, and in normal adults, it is expressed in regions that are responsible for producing large amounts of mechanical force. As shown in Table 1, a guanine-thymine (GT) dinucleotide repeat polymorphism of varying lengths is found within intron 17 of the gene, an intervening DNA sequence within a gene that does not encode for the protein. This polymorphism does not affect the amount of protein made, but it has shown an association with the occurrence of Achilles tendon injuries. The length of the dinucleotide repeat polymorphism may play a role in tendon injuries. Maffuli et al. (2013) reported that alleles containing 12 or 14 guanine-thymine repeats were overrepresented in the group with tendon injuries. Subjects with alleles containing 13 or 17 repeats were more likely to be asymptomatic for tendon injuries. The same GT dinucleotide polymorphism was examined in a study assessing Achilles tendon injuries (Mokone et al., 2005). Those with 12 and 14 genes were higher in the symptomatic individuals, whereas those with 13 and 17 repeats were systematically higher in the asymptomatic subjects. Those with 15, 16, 18, 19, 20, or 21 repeats were also assessed but showed insignificant results. Further research on this gene should aim to identify specifically how this polymorphism affects the individual, given that it does not appear to affect the amount of protein expressed.

The TNC gene is also closely linked to the gene that determines ABO blood groups on the long arm of chromosome 9 (9q34). Maffuli et al. (2013) explain that previous research has shown a correlation between the ABO blood groups and Achilles tendon injury. Those with blood group O have been found to be more susceptible to tendon injuries. With the connection between the TNC gene and the gene that determines ABO blood groups, it is possible that other mutations to alleles on chromosome 9 may also show an increase in tendon injury.

Vitamin D Receptor (VDR)

The levels of vitamin D contribute to the risk of stress fractures. In a study comparing male military personnel diagnosed with stress fractures to an uninjured, healthy control group, two polymorphisms showed that polymorphisms of the VDR increased the risk of a stress fracture (Chatzipapas et al., 2009). First, the FokI polymorphism (Table 1) in exon 2 of the VDR gene was analyzed. The FF version of the gene was most prevalent in the healthy controls, whereas the Ff and ff genotypes were more likely to be found in the injured military personnel. Chatzipapas et al. (2009) used logistic regression analysis to determine that the f-containing genotypes of Ff and ff were more likely to experience stress fractures with 8.0 (ff) and 3.0 (Ff) times the risk. Second, the BsmI polymorphism (Table 1) in exon 9 of the VDR gene was assessed for contribution to stress fracture risk. The B-containing genotypes of BB and Bb were more likely to be found in the injured patients. The bb genotype was more likely in the healthy volunteers. After logistic regression analysis, the B-containing genotypes showed an 8.8-(BB) and 5.3-(Bb) fold increase in risk. The B-containing genotypes were proven to have an increased risk of stress fractures (Chatzipapas et al., 2009).

In another study, the BsmI polymorphism (Table 1) was analyzed in healthy adolescent girls (Lorentzon et al., 2001). Lorentzon et al. (2001) analyzed how the BsmI polymorphism affected the bone mineral density of the lumbar spine. Like Chatzipapas et al. (2009), Lorentzon et al. (2001) found that the bone mineral density was lower in the BB and the Bb subjects than in the bb subjects. This study shows that mutations affecting the VDR's ability to function may decrease bone mineral density, putting the individual at an increased risk for fractures. Further research is needed to determine if the FokI or BsmI polymorphism plays a larger role in the risk of stress fractures.

Conclusion:

This study shows that there may be value in genetic testing in injury prevention techniques. The prevalence of genes that not only possibly play a role in athletic ability, but provide information about the athlete’s genetic predispositions, shows that the technology of genetic testing may, in the future, have a larger prevalence in athletics. In youth, the retention rate may increase if coaches and trainers are told about the genetic predispositions of the athletes.

While this may not be ideal for a whole team, kids who spend more time outside of practice receiving extra coaching may benefit from knowing their genetic makeup. This may allow the trainer to focus on more injury prevention exercises and may allow for more efficient or productive practices and shorter total training times. Less training will make the athlete less susceptible to overuse injuries and burnout. Further research on genetic testing in youth athletes should work to identify the prime age to do genetic testing, as well as which specific genes to test for different athletic pursuits.

References

Ben-Zaken, S., Meckel, Y., Nemet, D., & Eliakim, A. (2021). Insulin-like Growth Factor Axis Genetic Score and Sports Excellence. *Journal of strength and conditioning research*, *35*(9), 2421–2426. https://doi.org/10.1519/JSC.0000000000004102

Biber, R., & Gregory, A. (2010). Overuse injuries in youth sports: is there such a thing as too much sports? *Pediatric Annals*, *39*(5), 286–292. https://doi.org/10.3928/00904481-20100422-09

Chatzipapas C, Boikos S, Drosos GI, et al. Polymorphisms of the vitamin D receptor gene and stress fractures. Horm Metab Res. 2009;41:635–640. https://doi.org/10.1055/s-0029-1216375

Chiu, L. L., Wu, Y. F., Tang, M. T., Yu, H. C., Hsieh, L. L., & Hsieh, S. S. (2011). ACTN3 genotype and swimming performance in Taiwan. *International journal of sports medicine*, *32*(6), 476–480. https://doi.org/10.1055/s-0030-1263115

Dangi, Tek & Witt, Peter. (2016). Why Children/Youth Drop Out of Sports. *Journal of Park and Recreation Administration, 36(3)*. https://doi.org/10.18666/JPRA-2018-V36-I3-8618

Deuster, P.A., Contreras-Sesvold, C.L., O'Connor, F.G. *et al.* Genetic polymorphisms associated with exertional rhabdomyolysis. *European Journal of Applied Physiol*ogy 113, 1997–2004 (2013). https://doi.org/10.1007/s00421-013-2622-y

Ericsson KA, Krampe RT, Tesch-Romer C. The Role of Deliberate Practice in the Acquisition of Expert Performance. *Psychological Review.* 1993; 100(3): 363-406.

Goodlin, G. T., Roos, A. K., Roos, T. R., Hawkins, C., Beache, S., Baur, S., & Kim, S. K. (2015a). Applying personal genetic data to injury risk assessment in athletes. *PloS one*, *10*(4). https://doi.org/10.1371/journal.pone.0122676

Goodlin, G. T., Roos, T. R., Roos, A. K., & Kim, S. K. (2015b). The dawning age of genetic testing for sports injuries. *Clinical journal of sport medicine : official journal of the Canadian Academy of Sport Medicine*, *25*(1), 1–5. https://doi.org/10.1097/JSM.0000000000000158

Haile, D. W., Durussel, J., Mekonen, W., Ongaro, N., Anjila, E., Mooses, M., Daskalaki, E., Mooses, K., McClure, J. D., Sutehall, S., & Pitsiladis, Y. P. (2019). Effects of EPO on Blood Parameters and Running Performance in Kenyan Athletes. *Medicine and science in sports and exercise*, *51*(2), 299–307. https://doi.org/10.1249/MSS.0000000000001777

Kelley, B. & Carchia, C. (2013). "Hey, data data-swing". ESPN the Magazine Online. Retrieved from http://espn.go.com/espn/story/_/id/9469252/hiddendemographics-youth-sports-espn-magazine.

Khoschnau, S., Melhus, H., Jacobson, A., Rahme, H., Bengtsson, H., Ribom, E., Grundberg, E., Mallmin, H., & Michaëlsson, K. (2008). Type I collagen alpha1 Sp1 polymorphism and the risk of cruciate ligament ruptures or shoulder dislocations. *The American journal of sports medicine*, *36*(12), 2432–2436. https://doi.org/10.1177/0363546508320805

Jayanthi, N. A., Post, E. G., Laury, T. C., & Fabricant, P. D. (2019). Health Consequences of Youth Sport Specialization. *Journal of athletic training*, *54*(10), 1040–1049. https://doi.org/10.4085/1062-6050-380-18

Le Gall, F., Carling, C., Reilly, T., Vandewalle, H., Church, J., and Rochcongar, P. (2006). Incidence of injuries in elite French youth soccer players: a 10-season study. *Am. J. Sports Med.* 34, 928–938. doi: 10.1177/0363546505283271

Lorentzon, M., Lorentzon, R., & Nordström, P. (2001). Vitamin D receptor gene polymorphism is related to bone density, circulating osteocalcin, and parathyroid hormone in healthy adolescent girls. *Journal of bone and mineral metabolism*, *19*(5), 302–307. https://doi.org/10.1007/s007740170014

Maffulli, N., Margiotti, K., Longo, U. G., Loppini, M., Fazio, V. M., & Denaro, V. (2013). The genetics of sports injuries and athletic performance. *Muscles, ligaments and tendons journal*, *3*(3), 173–189.

Massidda, M., Vona, G., & Calò, C. M. (2009). Association between the ACTN3 R577X polymorphism and artistic gymnastic performance in Italy. *Genetic testing and molecular biomarkers*, *13*(3), 377–380. https://doi.org/10.1089/gtmb.2008.0157

Mokone, G. G., Gajjar, M., September, A. V., Schwellnus, M. P., Greenberg, J., Noakes, T. D., Collins, M. (2005). The Guanine-Thymine Dinucleotide Repeat Polymorphism within the Tenascin-C Gene is Associated with Achilles Tendon Injuries. *The American Journal of Sports Medicine*, 33(7,) 1016-1021.https://doi.org/10.1177/0363546504271986

Pasulka, J., Jayanthi, N., McCann, A., Dugas, L. R., & LaBella, C. (2017). Specialization patterns across various youth sports and relationship to injury risk. *The Physician and sportsmedicine*, *45*(3), 344–352. https://doi.org/10.1080/00913847.2017.1313077

Peak height velocity - guide for parents and children. London Bridge Sports Medicine. (2022, July 7). Retrieved April 19, 2023, from https://lbsm.co.uk/peak-height-velocity-guide-for-parents-and-children/

Pickering, C., & Kiely, J. (2018). Hamstring injury prevention: A role for genetic information?. *Medical hypotheses*, *119*, 58–62. https://doi.org/10.1016/j.mehy.2018.07.011

Pruna R, Artells R, Ribas J, et al. Single nucleotide polymorphisms associated with non-contact soft tissue injuries in elite professional soccer players: influence on degree of injury and recovery time. *BMC Musculoskelet Disord.* 2013;14:221.

Pruna, R., Artells, R., Lundblad, M., & Maffulli, N. (2017). Genetic biomarkers in non-contact muscle injuries in elite soccer players. *Knee Surgery, Sports Traumatology, Arthroscopy*, *25*, 3311-3318.

Roth, S. M., Walsh, S., Liu, D., Metter, E. J., Ferrucci, L., & Hurley, B. F. (2008). The ACTN3 R577X nonsense allele is under-represented in elite-level strength athletes. *European journal of human genetics : EJHG*, *16*(3), 391–394. https://doi.org/10.1038/sj.ejhg.5201964

"Sports Injury Statistics." *Stanford Medicine Children's Health*, https://www.stanfordchildrens.org/en/topic/default?id=sports-injury-statistics-90-P02787.

Stracciolini, A., Casciano, R., Levey Friedman, H., Meehan, W. P., 3rd, & Micheli, L. J. (2013). Pediatric sports injuries: an age comparison of children versus adolescents. *The American journal of sports medicine*, *41*(8), 1922–1929. https://doi.org/10.1177/0363546513490644

Williams, T. (2016). Reasons why kids drop out of sports. Our Everyday Life. Retrieved from http://oureverydaylife.com/reasons-kids-drop-out-sports-10440.html

Yan, X., Papadimitriou, I., Lidor, R., & Eynon, N. (2016). Nature versus Nurture in Determining Athletic Ability. *Medicine and sport science*, *61*, 15–28. https://doi.org/10.1159/000445238

Yang, N., MacArthur, D. G., Gulbin, J. P., Hahn, A. G., Beggs, A. H., Easteal, S., & North, K. (2003). ACTN3 genotype is associated with human elite athletic performance. *American journal of human genetics*, *73*(3), 627–631. https://doi.org/10.1086/377590