



# Biotechnology

*Biotechnology, as applied to sports, refers to working with the human genome to influence sport performance. A genetic predisposition for sickle cell anemia, for example, can be life-threatening in combination with certain sports. Genetic testing is also being used to select individuals for particular sports, which creates ethical issues, particularly when children are involved. Gene doping—inserting genetic material into an athlete—has the potential to increase athletic performance, although most sports decision-makers consider this a form of cheating: and one which cannot, at present, be tested for.*

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In 1953 Francis Crick and James Watson published their famous article in which they presented the structure of deoxyribonucleic acid (DNA). Half a century later the HUGO-project (Human Genome Organisation) completed the mapping of the human genome. Expectations of biotechnology have grown enormously ever since. These expectations not only concern the mapping and sequencing of many organisms, but also the modification of genes to cure diseases or to enhance human features. The world of sport is now confronting a variety of—more or less realistic—promises, varying from preventive screening to the genetic selection of talent and the enhancement of athletic performance with gene doping.

## Short Introduction to Biology

The human body is made up of about  $6 \times 10^{13}$  cells and 320 different types of cells (muscle cells or blood cells, for example). The nucleus of each human cell contains 23 pairs of chromosomes. These chromosomes carry most of our genetic material. This material, DNA, is made up of a chain of

nucleotide bases: adenine (A), guanine (G), thymine (T), and cytosine (C). These bases are grouped into two pairs: A binds to T and G binds to C.

In 1953 Watson and Crick discovered the double helix, the structure of DNA. DNA carries our genes and almost all hereditary information. DNA passes information to proteins via RNA (ribonucleic acid). RNA and proteins play a crucial role in reproducing DNA and in life processes in general (instead of thymine, RNA has uracil, and it is not structured as a double helix, but is instead single stranded). DNA is passed on from one generation to the next.

The human body is made up of around 3 billion base pairs and probably around 30,000 genes. A gene may be understood as a distinct unit of DNA, a particular string of A, T, G, and C. Each particular string of bases may stand for a piece of information that is used to produce amino acids and proteins (proteins are chains of twenty different types of amino acids).

Molecular biologists are not just getting insight into the genetic building blocks of life. Genetic engineering (or genetic transfer technology) of DNA is developing rapidly, and the applications of gene therapy seem promising. Gene technology has also resulted in controversial applications. After scientists started cloning animals (such as the famous sheep Dolly) many people were concerned about what would follow. Even greater concern followed when the theme of gene doping entered the media. Big muscled, genetically modified mice, named after the former bodybuilding champion (and later movie star and governor of California) Arnold Schwarzenegger, created many worries about the future of elite sport.

## Biotechnology and Sports

Genes are crucial for sport talent. Research on similar performances of identical (monozygotic) twin athletes already provided some clues to the heritability and the dominant role of genes that are related to sport performance. Empirical

evidence shows that the genetic component for human performance has grown quickly over the last few decades. Genes determine to a large extent anthropometrical features like height and length of arms and legs. Features like muscle size, muscle fiber composition (fast- and slow-twitch), heart size, lung size and volume, resting heart rate, muscular strength, flexibility of joints, and aerobic endurance are all in some respect trainable, but the influence of genes dominate.

Several large research programs all over the world are searching for the location of crucial genes that are related to (elite) sport performance. Several candidate genes are under scrutiny that might have a critical influence on specific sport talents. Genes that regulate muscle tissue and blood flow, for example, are associated with elite endurance athletes. More complex than establishing the relative contribution of genes (as opposed to non-genetic factors) is the search for specific genes, their location, and their specific influence on athletic performance.

By 2003 about 99 percent of the human genome was successfully mapped, although some researchers think that the human gene map is still in its infancy and that there is much more to accomplish. Research continues in the belief that in the long run, sport-relevant features may be localized in our genes. In 2003 a link was established between the ACTN3 gene and speed and endurance, and by 2008 companies in the United States were marketing a simple genetic test for sports aptitude, which they claimed would enable parents to steer their children toward or away from sports at which they would be more or less likely to succeed. More recent research, however, has disproven the likelihood that a single “performance gene” exists (Scott et al. 2010; Puthcheary et al. 2011). Genetic research instead focuses on identifying and using genes to improve specific physical attributes (such as muscle types and lung function mentioned earlier) that provide advantages in sports. After localizing these genes, the next step beyond testing and selection is modification, using the knowledge for enhancements of some kind.

Developments within molecular biology raise several promises that might have spectacular influence on modern sport, ranging from realistic possibilities to wild speculation. Three of these promises include: 1) the prevention of risks that are related to sports, 2) genetic screening to maximize

selection processes in elite sport, and 3) genetically modifying athletes (or gene doping).

## Genetic Screening and Prevention

Knowledge about our genetic makeup and its relation with health risks raises various possibilities for prevention. It is conceivable that the emergence of “sport genetic passports” has consequences for the ideology of “sports for all.” Insight into genetic makeup can contribute to a restriction of choices. A genetic predisposition for sickle cell anemia can be life threatening, for example, in combination with certain types of sports.

There is quite a body of evidence by now on the potential risks of brain damage when engaging in contact sports like boxing or even football (both American-style and soccer). Information on genetic makeup can radically change the discussion on compulsory testing and preventive measurements in boxing. Besides factors such as the length of the boxer’s career, and the number of knockouts and punches taken to the head, there are clear indications that genetic predisposition plays an important role in the prevalence of Parkinson’s disease, Alzheimer’s disease, and “dementia pugilistica” in boxing. Professional boxers who are homozygous for the Apo-E4-gene (which is seven percent) are at great risk for Parkinson’s or Alzheimer’s disease at a very young age. One out of six boxers will have Parkinson’s or Alzheimer’s disease by his fiftieth birthday. While no curative therapy is available, decisions have to be made with respect to preventive options like medical examination, waiting periods between matches, or restriction of a boxing career (temporarily or definitively). Also the question arises about what preventive options are available for young children who are practicing contact sports, either on a recreational basis or with the intention to become an elite athlete.

With specific preventive screening and advice, there are clear health benefits to be gained. Negative advice, such as “it is better not to box” are not without controversy. They do seem less complex, however, than the situation in which genetic testing is translated into more positive advice, for example when an extremely favorable genetic blueprint with respect to athletic features leads to a forced predestination to become an elite athlete.

## Using Genetic Information to Maximize Athletic Potential

Genetic screening has been used since about 2008 for the purpose of selecting children whose genetic makeup is particularly fitting for performing in a certain sport. This is a new step within the existing logic of “talent-scouting.” The application of gene technology as a selective tool would be one tool among others, but it does allow the opportunity to advance the moment of selection and to advance the efficiency of scouting.

Top coaches have expressed their interest in the possibilities of gene technology. Hartmut Buschbacher, a native of East Germany, has been coach of the US national women’s rowing team for nine years. He said the following about genetic screening: “As a coach, I’m interested in performance, and if this information would give me a better opportunity to select the athletes for my team, I would like to use that. That way you’re not going to waste so much time and energy on athletes who may not be as successful” (Farrey 2002).

This new and augmented level of knowledge, however, might decrease sports participation in general, especially among those who find that they have a less favorable genetic profile for high performance. Early insight into “objective” standards of talent can have a discouraging effect on young children with not enough talent for elite sports. Genetic screening may also contribute to a widening gap between sport as an educational and cultural phenomenon and sport as a merely selective and performance-driven practice in which the matching of the “right child to the right sport” becomes a central objective.

There is, on the other hand, a right to get an honest insight into the feasibility of high ambitions of parents and their children that are willing to offer their time and money in the prospects of sport success. In any case, the interests of the child need to be carefully weighed against those of ambitious parents, coaches, and countries.

Knowledge about genetic makeup may even enhance autonomy and contribute to the prevention of harm. A parent’s decision to exploit the athletic talent of a child is potentially a more restricting choice (given the relative uncertainty about the talents) than a choice that is based upon a genetic profile. This practice of early selection does,

however, raise moral and pedagogical questions on the threat of the autonomy of the selected children. The impact of knowing as a child that one is a carrier of genetic variants associated with a higher probability of success in high-performance sport may be problematic. The right to know may have to be balanced with the right not to know.

On the other hand one can imagine that potential grief will be spared for the non-selected, since they will not frustrate the high ambitions of their environment. In any case (with or without genetic screening) it remains of utmost importance to take care of alternative scenarios when the talent cannot live up to high expectations. That means, if necessary, a flexible readjustment of the athletic blueprint. It also means considering the (social, ethical, pedagogical and commercial) limits of “athletic predestination.”

## Genetically Modifying Athletes to Enhance Performance

One can think of several biotechnological applications to enhance athletic performance. Genetic information might be used, for example, to fine-tune training and nutrition in relation to the individual athlete’s genetic makeup (accepted as a legal application). One of the issues that appeal most to the imagination is the prospect of gene doping (or somatic genetic modification) in elite sport. Gene or cell doping is defined by the World Anti-Doping Agency (WADA) as “the non-therapeutic use of genes, genetic elements, and/or cells that have the capacity to enhance athletic performance” (WADA 2008).

The insertion of artificial genes is already possible, although there still remains the unsolved problem of controlling the activity of the artificial genes so they don’t produce too little or too much of the required substance. There are three possible ways to insert the artificial gene into the patient. The simplest way is to inject the DNA directly into the muscles. Some of the muscle fibers will then take up the DNA. Alternatively, one could introduce the DNA into cells in the laboratory and then inject these modified cells back into the body. Finally, one can utilize viruses for introducing foreign DNA into human cells.

Two possible applications of enhancing performance in sport could be the increase of red blood cells by inserting an EPO gene (erythropoietin is a synthetic hormone that

stimulates production of oxygen-carrying red blood cells) or the building of muscle mass by inactivating the myostatin gene (successfully applied to mice). After being injected with a synthetic gene into the muscles (to produce more insulin-like growth factor-1 [IGF-1]), the muscle force of the so-called Schwarzenegger mice increased by 60 percent after a month. The injection of EPO-genes in monkeys made the level of red blood cells rise from 40 percent red blood cells by volume to 70 percent (50 percent is a normal limit within elite sports).

Research on these possibilities for gene doping are part of the preparatory stage for the first genetically modified athlete. In the opening decade of the twenty-first century there was much talk and concern about the future possibility of gene doping, with some researchers, coaches, and athletes believing that as soon as gene therapy becomes a well-established technique, gene doping or genetic engineering of elite athletes will also become a routine practice. At the 2012 Olympics in London some speculated that gene doping had become reality. The case that caught people's attention was that of 16-year-old Chinese swimmer Ye Shiwen. Relatively unknown in international swimming before the Olympics, she won two gold medals, set a new world record, and significantly improved over her previous best times. But what especially caught judges' and officials' attention was the fact that she beat the male world champion's time. That fact, combined with the officials' experience with similar remarkable performances being the result of doping, brought Ye under suspicion. The officials described her performance as literally unbelievable because it shouldn't have been possible. She passed the drug tests, however, which led some officials to claim that genetic modification was one plausible explanation for her startling performance (Bull 2012; Naish 2012). The Chinese denied the charge, and as gene doping cannot yet be tested for, the dispute could not be resolved by scientific evidence. Nonetheless, many in the sports world believe that some athletes are also trying or about to try doping. Scientists feel certain that even if gene doping did not occur at the 2012 Olympics, its emergence is nevertheless inevitable.

On the other hand, biogenetical scientists temper high expectations like these, and think it is rather unlikely that in the near future the fiction of the genetically modified athlete will become reality. Many genes are involved in

athletic performance, especially in those sports that do not just measure force or speed. There are complex interactions among different genes, and there are complex interactions among genes and the environment. There is not one crucial gene for sport talent, which can be identified, inserted, or modified at will.

Notwithstanding these diverging interpretations of future scenarios, there is no doubt that biotechnological developments will raise and stimulate a broad spectrum of ethical and socio-political question.

## Sport, Biotechnology, and Ethics

General basic principles, like autonomy, privacy, justice, equity, and human dignity have been applied to reach an international consensus and to harmonize national regulations on biotechnology. It is, however, questionable if conventional human rights meet all the dilemmas that will arise from modern biotechnology. Some of the following human rights come into play with the engineering of human genes: the Kantian maxim of not treating an individual as a means to an end, a right to a unique genotype, no harm done to unborn life, no discrimination, the right to privacy, and an equal availability to new technology. People who oppose the use of molecular biology to enhance the human being think this new technology will endanger the "intrinsic value of diversity" or stress that it is a reprehensible way of "playing God."

The ethical discussions on genetic enhancement in sport are more specific. The purpose of the World Anti-Doping Program as enforced by WADA is to protect the athletes' fundamental right to participate in doping-free sport and thus to promote health, fairness, and equality for athletes worldwide. WADA has tried to keep up with all the latest developments within gene technology. International regulation and harmonization of the anti-doping policy remains the primary aim. The representation of a "pure and honest sport" is also related to the possibilities of public identification and with the protection of commercial interests.

Although there is a long history of the use and detection of performance enhancing substances in sport, it seems that the use of biotechnology raises some new and rather complex issues. A major problem concerns the (moral and practical) implications of control and detection. Although WADA initiates genetic research with the aim of staying ahead of the

athlete, anti-doping authorities are facing serious problems in their search for a useful test to find gene-doped athletes. The protein produced by the artificial gene will be identical to the endogenous protein since the human body itself produces it. If pure DNA or cells are used, this DNA will be present only at the site of injection. That means that a muscle biopsy of the injected site would be required. But what if athletes do not consent to a biopsy of the muscle? What is considered an autonomous decision within a clinical context means fraud in the context of elite sport. The implications of control for “pure genes” ask for a careful consideration of the principle of informed consent in elite sport. Controlling practices also raise questions on the privacy of the screened athlete. WADA aims at an insight into medical passports of all athletes to be able to screen the relevant medical parameters for a longer period of time. The question is, who will “own” and protect the genetic information of the athletes? Besides moral and practical questions concerning detection, there is also a concern how these developments will influence the sport and athlete themselves.

Another complex issue concerns the distinction between therapy and non-therapy. It is very hard to draw a clear line between a therapeutic and enhancing use of genetics. Why is it that, contrary to the ill patient who wants to become better, the healthy athlete who wants to get “better” is the target of condemnation? Is there really a principle difference between gene-therapy to replace defective genes with healthy genes versus the enhancement of initially healthy genes? Although this boundary is often hard to draw, it is still one of the most important fundamentals of the international policy on sport and doping.

It is clear that the discussion on sport and genetics is much broader than just about protecting the health of the athlete or protecting the idea of a fair competition. What happens when “talent” is not something “just given” but a feature we can modify or choose for? It might become necessary that high-performance sport will have to adopt new ethics that are adapted to changing times and technologies. For the time being, good genes do not suffice for success in sport. “Dedication” remains an important, “humanlike” factor that composes one of the relevant inequalities we want to measure for in sport. Sport is about measuring performances with the aim of comparing differences in talent and dedication. To be competitive within modern elite sports takes both talent

and dedication in extreme quantities. This will not change with genetic technology. Biotechnology cannot replace training effort. Gene technology can however change our perception of a fair and human way of practicing sport.

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See also Doping; Elite Sport Parents; Olympics 2012; Sports Science; Technology; Training, Athletic

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