Case 8

A scientific paper entitled "*ACTN3* genotype is associated with human elite athletic performance" was published in the July 2003 issue of the American Journal of Human Genetics. *ACTN3* is the gene encoding for *alpha*-actinin 3, a protein involved in the ability of skeletal muscles to contract quickly and forcefully. *ACTN3* was found in significantly higher percentages in the muscles of male and female sprinters when compared to those of endurance athletes.

A few years ago, a variation in the gene that encodes the angiotensin-converting enzyme (*ACE*), a blood pressure regulator, was linked to enhanced endurance performance and increased muscle build-up following intensive training. While *ACTN3* is only the 2nd gene to be linked to the exercising ability of skeletal muscle, new information is published with increasing frequency. Scientific data already show that, in addition to psychological make-up and muscle fiber types, body size, metabolic efficiency and lung capacity also contribute to successful athletic performance. For example, Schwarzenegger mice (a specific strain of genetically engineered transgenic mice) remain full of vitality, maintain their exercise ability and keep their muscles' athletic edge while octogenarian because their muscles have been engineered to contain additional amounts of insulin growth factor-1, a protein involved in tissue growth and development.

Coaches and athletes seem very interested in the potential of genetic engineering for improving athletic performance. The fact that genetic manipulation could change the current meaning of an athlete, i.e. a person whose physical achievements and prowess depend on such qualities as endurance, perseverance, dedication, competitiveness and patience, does not seem to raise doubts as to its future use. Former Olympic coach Hartmut Buschbacher said "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". Further, Princeton geneticist Dr. Lee Silver was quoted as saying, "By using genetic testing, you will be able to identify perhaps 80 percent of the children who have any potential to be a pro athlete."

Many consider an important accomplishment of modern society to be the transformation of sports from an elite activity of the rich to a widespread opportunity available to almost every member of society. While the possibilities of genetic profiling appear very exciting, testing could also result in loss of opportunity to participate in athletics for those not favored genetically. This could mean the loss of hope, especially for underprivileged children for whom sports might be the only way to improve their lives. Among the negative consequences of genetic profiling, feared by some, are loss of interest in exercise, health deterioration, creation of an elite athlete super race, and parents who will design babies with athletic aptitude in mind.

Based on information yielded from genetic testing, concerned voices ask for legislation to regulate the participation of athletes genetically predisposed to head injury in sports with an increased risk for repeated head trauma. The Victorian government in Australia is considering compulsory genetic testing for boxers based on the association of the *e4* variation of the apolipoprotein E (*ApoE*) gene with worse recovery after head injury. However, *ApoE4* has also been linked to an increased risk for the development of Alzheimer's disease (AD) later in life. The fact that a gene could be used both as a predictor for AD and athletic performance raises many difficult issues. For example, the need to protect an individual from possible dangers inherent in sports would have to be weighed against revealing the potentially devastating news of increased risk for AD in the future.