Sex and gender issues in competitive sports: investigation of a historical case leads to a new viewpoint

Kaye N Ballantyne, Manfred Kayser, J Anton Grootegoed

ABSTRACT

Based on DNA analysis of a historical case, the authors describe how a female athlete can be unknowingly confronted with the consequences of a disorder of sex development resulting in hyperandrogenism emerging early in her sports career. In such a situation, it is harmful and confusing to question sex and gender. Exposure to either a low or high level of endogenous testosterone from puberty is a decisive factor with respect to sexual dimorphism of physical performance. Yet, measurement of testosterone is not the means by which questions of an athlete’s eligibility to compete with either women or men are resolved. The authors discuss that it might be justifiable to use the circulating testosterone level as an endocrinological parameter, to try to arrive at an objective criterion in evaluating what separates women and men in sports competitions, which could prevent the initiation of complicated, lengthy and damaging sex and gender verification procedures.

INTRODUCTION

In 1949, the Dutch track athlete Foekje Dillema (1926–2007) came to prominence on the world athletic stage. She started to rival Fanny Blankers-Koen, the world-famous Dutch track athlete who won four gold medals during the 1948 Summer Olympics in London and was elected Female Athlete of the Century by the International Association of Athletics Federations (IAAF) in 1999. In contrast, Dillema’s career was of short duration, with a dramatic ending. In 1950, she was expelled for life by the Royal Dutch Athletics Federation, due to the results of a ‘sex test’, for which details or results were never revealed and no records are available. Her 1950 national record of 24.1 s for the 200 m, which she took from Fanny Blankers-Koen, was erased, and only after her death 57 years later was she reinstated by the Royal Dutch Athletics Federation (figure 1).1

The verification of the sex of athletes has been an issue for many decades. It should be noted that reports and reviews on this topic refer to gender verification, rather than sex verification.2–4 However, what counts in competitive sports is a person’s sex characteristics. Trying to avoid the word sex, given its charged nature, can only cause confusion.5 6 Herein, we will use the term sex for the biological and physiological characteristics that define men and women, as compared to gender and gender identity in reference to the socially and individually perceived sexual identity of an individual from birth to puberty and adulthood.5 7 8

Sex verification in 1950 was based solely on physical examination, predating hormone assays or sex chromosome analysis. Following discovery of the Barr body in female cells in 1949, it took some 12 years before it was known that this body represents an inactivated X chromosome,2 from the late 1960s its detection was used in sex verification tests in the context of sports competitions.2–4 Subsequent tests focused on the male-specific region of the Y chromosome, particularly the male sex determining SRY gene.10

To broaden the perspective on sportswomen confronted with questioned sex characteristics, we have investigated the case of Foekje Dillema, with informed consent from her heirs, by means of DNA analysis of samples from worn clothing. Appreciating the nature of the samples tested, we applied DNA methodology and lab quality standards used in human forensics. Our DNA analysis indicates that Foekje Dillema had a 46,XX/46,XY mosaic condition with a rare origin, which we interpret as leading to hyperandrogenism from her puberty. Based on this historical case we discuss that, if a sportswoman is confronted with signs of a DSD early in her sports career, it is harmful and confusing to question such a person’s sex and gender. Rather, we suggest that it is necessary to try to arrive at an objective criterion in evaluating what separates women and men in sports competitions.

RECONSTRUCTION OF A HISTORICAL CASE

From the combined genotyping and DNA quantification results, we conclude that Foekje Dillema was a 46,XX/46,XY mosaic, with equal numbers of both genetic cell types at least in her skin (online supplementary data). In the fetal gonads of a 46,XX/46,XY mosaic, the tissue ratio of XX:XY cells will push the bipotential gonads to become either ovaries or testes, or both. A preponderance of 46,XX cells in fetal gonads can

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lead to the development of ovaries, but with some 46,XY testis tissue present in one or both of these ovaries. Such an ovotestis condition, which can also occur in the form of a complete ovary and a complete testis on either side, has been referred to with the term true hermaphroditism. The most common karyotype found in true hermaphroditism is 46,XX, followed by 46,XX/46,XY chimerism and mosaicism. We would like to emphasise that the term hermaphrodite, as well as other terms such as intersex, need to be replaced by these regulations, hyperandrogenism caused by ovotesticular DSD in Dillema’s case of ovotesticular DSD. From photographs, it is evident that from puberty she had breast development, but personal accounts also indicate that she showed some facial hair growth. Hyperandrogenism from puberty may have contributed to Dillema’s athletic performance.

**SEX CHARACTERISTICS, GENDER IDENTITY AND TESTOSTERONE**

To have, or not to have, a Y chromosome is the primary decisive factor in human sexual differentiation, but there are exceptions. A prominent example is offered by 46,XY females who have the complete form of androgen insensitivity syndrome (cAIS), when the testes produce testosterone but the body is not able to respond to androgens (testosterone and its more powerful metabolite dihydrotestosterone) due to mutation of the X-encoded androgen receptor. Consequently, these individuals are born and raised as girls, and have a female gender identity. Action of testosterone through binding to the androgen receptor in the developing fetal brain is the predominant factor in programming human male gender identity, and the female gender identity of 46,XY cAIS women is explained by loss of this androgenic effect. In sports, 46,XY cAIS women can be expected to have a disadvantage compared to 46,XX women with a functional androgen receptor, the latter profiting from stimulation of muscle strength by a low level of circulating testosterone.

The biological basis for sex segregation in sports is the consequence of long-term endogenous androgen exposure of men after puberty. It cannot be excluded that proteins encoded by genes in the male-specific region of the Y chromosome (MSY) might act together with androgens, widening the physiological gap between women and men. However, such a role for MSY genes will be minor, compared to the predominant role of androgen action. In men, the postpubertal testosterone level is a proven dose-dependent factor when muscle strength and other physiological factors such as the blood haemoglobin level come into play. A moderate pubertal and postpubertal excess of testosterone in a young woman can give extra muscle development and other signs of hyperandrogenism, but it would be a rude error to even suggest that this would affect her female gender identity.

Competitive athletes exploit fortunate combinations of natural differences in physical and mental personal characteristics, including individual variation of the endogenous testosterone level. The World Anti-Doping Agency states that an athlete’s sample will be found positive if the concentration of an endogenous androgenic steroid hormone is above the range normally found in humans, and is not likely consistent with normal endogenous production, unless the elevated concentration of the steroid hormone (or metabolites or markers) is attributable to a physiological or pathologic condition. Strictly speaking, a female athlete is free to benefit from any endogenous source of androgen production. Some female athletes may benefit, probably to a small extent, from increased androgen production originating from a polycystic ovary. This is viewed as acceptable by the IAAF, who stated that conditions that may provide some advantages but nevertheless are acceptable include congenital adrenal hyperplasia, androgen-producing tumours and an ovulatory androgen excess associated with a polycystic ovary. According to these regulations, hyperandrogenism caused by ovotesticular DSD would be unacceptable only if sex and gender
men,27 and with a reference range of 11–35 nmol/l for men,31 there is a substantial and significant gap in the testosterone level between women and men.

It has been argued that sex is not a binary quantity, with the far-reaching implication that sex segregation in competitive sports is an inconsistent and unjust policy.32 This argument was substantiated by pointing out that an individual's genetic background may cause a differential sensitivity to testosterone. Indeed, a genetic polymorphism such as the CAG repeat polymorphism in exon 1 of the gene encoding the androgen receptor affects the sensitivity of cells and tissues to androgen.33 However, this effect is likely far too small34 to provide any female athlete with an advantage bringing her on par with male athletes. Such a common genetic variation should not be taken into account and does not obstruct the prevailing thought that women and men are to compete separately, meaning that there is a need for a dividing line.35 Thinking about a dividing line, there is much agreement that current principles and procedures need to be revisited.35–37

The historical case described herein concerns ovotesticular DSD, where the amount of steroidogenic testicular tissue will determine if the affected person develops as a woman or as a man, regarding both gender and sexual characteristics. As such, this type of DSD can be viewed as a paradigm, demonstrating that the testosterone level might offer an objective parameter to separate the sexes, if required. In fact, this parameter is already implemented, in the context of sports. Athletes (46,XY and androgen sensitive) who have undergone male-to-female sex reassignment are welcome to engage in sports competitions from 2 years after the sex change, as of the Olympic Games 2004 in Athens, according to fortunate emancipative regulations by the International Olympic Committee.38 Similarly, athletes (46,XX and androgen sensitive) who have undergone female-to-male sex reassignment can compete, but they will receive exogenous testosterone. For female-to-male sex-reassigned individuals receiving testosterone supplementation, a total testosterone level of around 30 nmol/l has been reported.39 Perhaps, one day we may witness a talented 46,XX sex-reassigned male who is able to successfully compete with 46,XY males, thanks also to approved testosterone supplementation. Men exposed to stress and exhaustion face difficulty in maintaining their endogenous testosterone level,40–42 which might imply an advantage for 46,XX sex-reassigned males, particularly in endurance sports. Similarly, a therapeutic use exemption for long-term testosterone administration in 46,XY men, to compensate for a secondary loss of gonadal testosterone production, might provide an advantage. The above serves to illustrate the point that current concepts and regulations regarding the relationship between sex and testosterone in sports offer room for consideration. We feel that this should be taken as a starting point to discuss the circulating testosterone level as a relevant criterion in evaluating what separates women and men in sports competitions.

The present report is not meant to provide a guideline, which would require detailed analysis of total testosterone levels in large numbers of female and male athletes in relation to possible confounding factors, and consensus meetings. Rather, we aim to contribute to an open discussion involving experts from the fields of biology, medicine, genetics, psychology, sports and ethics, to accomplish a procedure which would respect the authenticity of an adult individual's sex and gender identity.
Note: On 12 April 2011, the International Association of Athletics Federations (IAAF) announced the adoption of new rules and regulations governing the eligibility of females with hyperandrogenism to participate in women’s competition, which will come into force from 1 May 2011 (http://www.iaaf.org/aboutiaaf/news/newsid=59874.html).

It appears that these new IAAF rules, as announced, are in full agreement with the viewpoint expressed in our article, which at the time of the IAAF announcement was already in press with the British Journal of Sports Medicine. We would like to emphasise that our viewpoint was composed independently from IAAF, and that none of the authors has been in contact with the respective IAAF expert working group.

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