Gender verification in sport

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Sir

I was interested in the controversial paper on gender verification in sport by Professor Ferguson-Smith and Dr E. A. Ferris in the March issue of the Journal (Br J Sp Med 1991; 25: 17-20). They produced evidence that 1 in 500-600 sportswomen were ineligible to compete in various prestigious competitive events because testing showed them to be chromatin-negative. They pointed out the difficulty in obtaining diagnoses but they know of no case where the test revealed a man masquerading as a woman and presumably none had Turner's syndrome with associated dwarfism. It was therefore thought that those ineligible were XY females and, because the majority would have the androgen insensitivity syndrome, most would be disqualified unfairly. My main concern is whether this assumption is correct.

It is certainly stated that the XY female phenotype (male pseudohermaphroditism) is most commonly caused by androgen insensitivity due to lack of androgen receptors. Its incidence has been quoted as 1 in 20 000 and tentatively concluded as 1 in 62 400 male births1. There is thus a huge discrepancy between its frequency in the population and the 1 in 500-600 ineligible sportswomen quoted in the paper. If the XY status conferred no benefit in sporting performance, then the incidence of XY females in sport and in the population should be the same. One wonders whether the ineligible group had achieved sporting excellence by having an unfair advantage in terms of their biologically active androgen status. If so, they were correctly disqualified. However, without clinical and laboratory data this remains unproven.

Although it is accepted that androgen insensitivity accounts for the biggest proportion of XY females, the claim that only 10% have the partial or incomplete syndrome with virilization at puberty2 seems open to question.

Of the seven XY female patients studied by this laboratory and for whom records are still available, only one was thought to have the complete androgen insensitivity syndrome (testicular feminization), albeit in a variant form, while three had pure gonadal dysgenesis. I agree with Professor Ferguson-Smith and Dr Ferris that, in spite of being sex chromatin-negative, these patients would not have an unfair advantage in competitive sports and should be eligible. On the other hand, three presented with ambiguous genitalia and other signs of virilization; two had incomplete androgen insensitivity and one had deficiency of 17β-hydroxysteroid dehydrogenase. Curiously, although this enzyme defect produces a fall in gonadal testosterone production, plasma levels are in the male range from peripheral conversion of precursors which are produced in excessive amounts.

Another group which virilizes at puberty is that with 5α-reductase deficiency, but none have presented at our clinic. Incidentally, these male pseudohermaphrodites sometimes undergo gender reassignment after puberty and can produce normal semen3. They may account for some of those documented in the historical background section of the paper.

In the light of statistics about XY females, it is surprising that almost half our small series had evidence of excess biologically active androgen. Furthermore, in a much larger series of intersex patients with a Y chromosome from the Johns Hopkins Hospital, 11 had gonadal dysgenesis, two had asymmetrical gonadal differentiation, 23 had testicular feminization and 62 had virilizing male hermaphroditism4. Thus well over half had higher androgen activity than that found in normal women. It could be argued that hospital figures are distorted because virilization rather than primary amenorrhoea may be the reason that XY females seek referral. This is unlikely considering the number of other patients with primary amenorrhoea seen in hospital practice.

Although the argument is that perhaps fewer sportswomen were unjustly ineligible than proposed by Professor Ferguson-Smith and Dr Ferris, it is still unsatisfactory for anyone to be disqualified unfairly. The current sex chromatin testing programme therefore needs to change. A move to have gender verification undertaken before sportswomen register with national associations or governing bodies would be welcome. The sex chromatin pattern is immutable, and so it should be possible to avoid putting competitors through the indignity of recurrent buccal smear tests. Perhaps a few further comments can be made.

I certainly agree that those found to be sex chromatin-negative should be able, indeed encouraged, to consult a clinician. Apart from the need to establish a diagnosis and determine eligibility to compete in sport, XY females should be advised to undergo gonadectomy to avoid the high risk of tumour development4. Indeed, from the figures quoted in the paper, the sex chromatin testing of sportswomen is an efficient screening procedure for this potential malignancy. Whether virilized pseudohermaphrodites can be deemed eligible after gonadectomy is open to debate, especially as this may lead to similar demands from male to female trans-sexuals who have undergone gender reassignment surgery.

Finally, it has to be mentioned that, apart from those with pure gonadal dysgenesis, all the XY females discussed above have normal male blood testosterone levels. In fact, in complete androgen insensitivity the levels are even higher because of increased secretion of gonadotrophins in response to activation of the feedback at the pituitary. These testosterone levels would be reflected in urine concentrations well above the female range, leading to possible detection in the drug testing programme. After gonadectomy this would not be a problem, but otherwise the Doping Control Laboratories would need to know the sex chromatin status of sportswomen with high urinary testosterone secretion.

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