

## WHY SEX CHROMATIN SHOULD BE ABANDONED AS A SCREENING METHOD FOR "GENDER VERIFICATION" OF FEMALE ATHLETES

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“ This paper explains how the screening of female athletes by sex chromatin, practised by most sports organizations, often does not reveal the categories of individuals who should not be allowed to compete in women's events, while it excludes other individuals who have "abnormal" sex chromatin but should be allowed to compete in these events. Since this method can be considered a near total failure, the Author sustains it should be abandoned. ”

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### Introduction

Sports organisations have not made it entirely clear what the aims of "gender verification" or "femininity control" of female athletes are. In the following I assume that the aim is to exclude two categories of individuals, namely, (1) males, (2) females whose body build and muscle strength would give them an unfair advantage over normal women in certain sports events.

A two-stage test procedure is practised by most sports organisations. First, all individuals undergo a screening test consisting of X chromatin (in some instances also Y chromatin). If the X and Y chromatin results are normal, female gender is considered established, a certificate of "femininity" is issued and participation in the women's event allowed. If the sex chromatin result is abnormal, further tests, including the determination of karyotype and clinical and gynaecological examination are carried out. The medical authorities of the sports organisations then decide in casu about

the participation of the individual. As far as I know there are no published guidelines as to what criteria are used in the decision-making.

In this paper the screening by sex chromatin is critically reviewed. I conclude that it is inaccurate in that it reveals only a fraction of those muscular females who should probably not be allowed to compete in the women's events. On the other hand, it "reveals" females with certain types of congenital chromosome abnormalities whose body build and muscle strength are female, and who should not be excluded. Thus sex chromatin screening of female athletes is a near-total failure and should be abandoned.

### **Sex chromatin**

The term sex chromatin comprises two different entities, X chromatin and Y chromatin. Both are simple in principle. Epithelial cells are scraped off from the buccal mucosa with the aid of a spatula, smeared on to a glass slide, then fixed, stained, and studied under the microscope. Separate slides are made for X and Y chromatin. For the person tested the procedure is painless and takes a few seconds.

The X chromatin is a darkly staining body at the inner surface of the nuclear membrane. The number of X chromatin bodies per cell equals the number of X chromosomes minus one. Thus the X chromatin offers an easy and relatively simple and reliable way of determining the number of X chromosomes of the person tested. Males (whose karyotype is 46,XY) have no X chromatin, while females (whose karyotype is 46,XX) have one X chromatin body. Moreover, females with the abnormal

karyotype 45,X as well as females with the karyotype 46,XY (cf below) have no X chromatin.

The Y chromatin body seen by fluorescence microscopy after quinacrine staining of cell nuclei is a brightly fluorescent spot that occurs only in cells that have a Y chromosome. Thus, in principle, Y chromatin is a relatively simple way of determining the number of Y chromosomes of the person tested. Males (and females with 46,XY) have y chromatin, while normal females (and females with 45,X) do not have Y chromatin. However, the determination of Y chromatin is notoriously unreliable for several reasons. It has been estimated that only about 85% of samples can be accurately evaluated; as a consequence the method has been largely abandoned as a screening procedure in medical practice.

### **Categories of individuals who should not be allowed to compete in the women's events**

#### *1. Males posing as females*

As long as the records of the sports organisations are not open to inspection, and as nothing has been published on this subject, it is impossible to determine how rarely or commonly males have posed as females. In private conversations with sports officials I have been told that such events have been exceedingly rare or have not occurred at all. Sex chromatin screening would reveal such individuals.

#### *2. Females who take or have taken hormones*

It is not known precisely how common hormone doping is among

athletes, but judging from the number of papers published in the lay press this is a widespread problem. There is no doubt that steroid hormones can virilize women and have a profound masculinizing effect on their muscles. In this context it may be especially important to consider the late effects on body build and muscles in females taking these drugs in e.g. childhood or adolescence. Such medication would not be revealed by hormone assays at the time of competition. Recent rumors about the use of growth hormone by athletes indicate that a previously unknown type of problem may exist. Sex chromatin screening will not reveal hormone doping of any kind in female athletes.

### 3. *Females with congenital adrenal hyperplasia*

There are several forms of this common congenital, hereditary disorder. Severely affected females are virilized at birth, have ambiguous external genitalia and masculine body proportions. In milder forms the only manifestations may be increased muscular strength and pilosity in otherwise fairly normal-appearing women. These muscular females are likely to be good athletes. The condition can usually be treated if the diagnosis is established in childhood. Successful treatment results in normal or almost normal female characteristics including fertility. Sex chromatin screening will not reveal these conditions.



Helmar Hommel / FRG

212 - World Champion, Marita Koch (GDR) running in Helsinki (1983).

#### 4. *Pseudohermaphrodites and true hermaphrodites*

These are very rare conditions of variable etiology. Affected individuals display a range of physical signs from almost male to almost female. A few individuals are quite female in appearance yet have masculine muscles, and consequently might register as female athletes. The problem is that the majority, probably around 80%, have the karyotype 46,XX and so are not detected by sex chromatin. The proportion revealed by the present screening method is thus of the order of 20 per cent of these rare individuals.

#### **Categories of individuals who have "abnormal" sex chromatin but should be allowed to compete in the women's events**

##### 1. *Females with the karyotype 45,X who have Turner's syndrome*

Females with Turner's syndrome are female in appearance and by their psychosexual orientation. Over half of patients with Turner's syndrome have the karyotype 45,X. They are short and have either scanty menstrual bleedings or no periods at all. Childbirth is rare. They should be allowed to compete in the women's events, but are "revealed" as "male" by sex chromatin.

##### 2. *Females with the karyotype 46,XY who have either the testicular feminization syndrome or gonadal dysgenesis*

These conditions are not uncommon. The testicular feminization syndrome is a hereditary disorder characterized by the presence of intraabdom-

inal atrophic testes in association with a female habitus, including external genitalia, muscle strength and psychosexual orientation. In 46,XY gonadal dysgenesis there are no gonads at all and the appearance is female. Both types of women should be allowed to compete with other women but are "revealed" as "male" by sex chromatin.

#### **Categories of males who would pass as females by sex chromatin screening**

Klinefelter's syndrome (karyotype 47,XXY) and the XX male syndrome (karyotype 46,XX) are characterized by a male habitus including body build, muscle strength, and psychosexual orientation. The testes are small but the genitalia otherwise normal. While these individuals would seem to be as unlikely as normal men to pose as females, they would pass as females by X chromatin screening. XX males, but not XXY males, would pass as females even if Y chromatin were done in addition to X chromatin.

#### **Conclusions**

The inadequacy of sex chromatin as a screening procedure is manifested in two different ways. Firstly, it reveals only a fraction of those individuals who should not compete in the women's events. How small this fraction is cannot be determined with certainty, but there is every reason to believe that it is remarkably small. As shown above, the two major categories not revealed are women taking hormones and women with congenital adrenal

hyperplasia. The only categories revealed are males posing as females, and some 20% of hermaphrodites. Thus my estimate of the total fraction revealed is at most 10 per cent, perhaps much less. Secondly, sex chromatin screening "reveals" women who should be able to compete with other women, i.e. women with Turner's syndrome, testicular feminization, and XY gonadal dysgenesis.

In summary, sex chromatin screening fails to detect most of the potential frauds but "detects" and thus compromises and harasses women with certain congenital chromosomal abnormalities who should be allowed to compete. Thus there are two good reasons to abandon sex chromatin screening.

## Discussion

The failures of sex chromatin in detecting frauds mainly concern two conditions, namely hormone doping and congenital adrenal hyperplasia. Both are very common, but it cannot be determined how commonly they occur in the context of female sports events. Neither is detected by sex chromatin screening. Thus the present screening method must be considered a near-total failure.

If sex chromatin is as inaccurate and harmful as shown in this paper, why have sports organisations not already abandoned sex chromatin screening? It may be that the concept of an easy laboratory test that reveals "men" has appeared so attractive that sports offi-

cial have not bothered to find out what the test actually detects. There has been a need to observe strict confidentiality towards athletes in whom an "abnormal" result has been found. This in turn may have prevented sports officials from properly analysing and finding out in depth what conditions those few individuals have suffered from who have become "revealed". The profound personal tragedies inflicted on women with chromosomal abnormalities who have been prevented from participating in competitions or otherwise harassed have perhaps not been apparent because of the confidentiality and secrecy with which these cases have been (rightly) handled.

One further reason why sports officials have not abandoned sex chromatin screening may be their fear of negative reactions from female athletes. In the lay press there have appeared interviews with women saying e.g. that a physical examination is unacceptable. While there is no firm evidence one way or the other, it may be that many sports women, if asked, would indicate that they find the present test system acceptable. It would appear to be a true tragedy if sports women were to support a system that is both highly inaccurate (not detecting most frauds) and highly discriminatory ("revealing") women who should not be excluded). Such an attitude is understandable as long as it is based on ignorance about the facts. It follows that the facts about sex chromatin screening should be as widely publicized as possible.