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SHORT PAPER

Testicular hypoplasia in a horned goat with 61, XXY/60, XY karyotype

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ABSTRACT

A horned goat with testicular hypoplasia was examined cytogenetically. A total of 72 cells were analyzed, 62 of which had an extra X chromosome (2n=61, XXY) and 10 of which had a male karyotype (2n=60, XY). We propose the hypothesis that, in this case, testicular hypoplasia may have been related to mosaicism of the sex chromosomes.

In view of the present widespread interest in dairy goat breeding the world over, it is of critical importance to perform systematic studies on possible genetic factors which may negatively affect reproduction in this species. One of these factors, testicular hypoplasia, has been held responsible for at least 1/3 of all cases of goat infertility and may be associated with a condition of intersexuality.

The most common type of goat intersexuality occurs only in hornless goats and is caused by a dominant P gene which, in homozygosis, affects the embryonic sex development of XX (HY-positive) animals. On the other hand, cases of intersexuality associated with sex chromosome mosaicism have been reported for horned animals (Padeh *et al.* 1965; Bongso *et al.* 1982).

The objective of the present study was to determine the etiology of testicular hypoplasia in a horned goat in order to establish the chromosome condition of the animal.

The animal was an adult crossbred Nubian goat with horns and with the form of a male diagnosed as having testicular hypoplasia. No other physical abnormality was detected. (Fig. 1). The animal was donated to the Department of Genetics, São Paulo State University, Botucatu, and originated from the township of Botucatu. The cytogenetic technique used was based on that described by Moorhead *et al.* (1960), slightly midified. Eight drops of peripheral blood were transferred to a tube containing 4 ml RPMI or F12 medium, 1 ml bovine fetal serum and 0.1 ml phytohemagglutinin, and incubated at 37°C for 72 or 96 hours. The material was treated with 0.0004 mg colchicine/10 ml distilled water and with a hypotonic 0.075 M KCl solution for 30

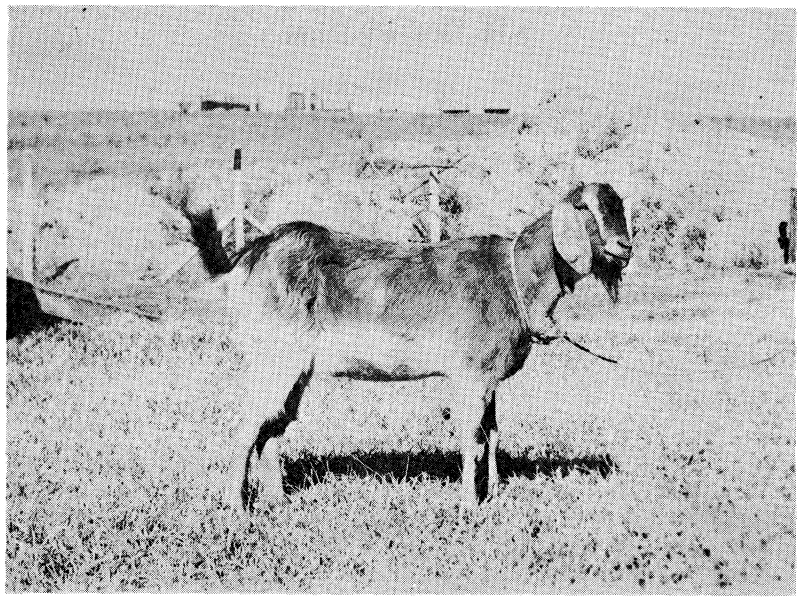

Fig. 1. Phenotype of the XXY/XY male caprine.

Table 1. *Chromosome number and sex complement detected in blood cells of a horned goat with testicular hypoplasia*

Protocols	Number of cells analyzed	Chromosome Number 60, XY	61, XXY	Aneuploid Frequency (%)
2311	72	10	62	86.11

minutes, and stained with Giemsa for 4 minutes.

The analysis of 72 metaphases obtained from lymphocyte cultures is shown in Table 1. The cells with 61 chromosomes showed an extra X chromosome. The Y chromosome was present in all of the 72 cells studied. Figs. 2 and 3 show the 2n=60, XY 2n=61, XXY constitution with conventional staining. In Fig. 4, the chromosomes were paired on the basis of the bands which appeared with light and dark regions, even though conventional staining was used.

The results permit us to raise the hypothesis that testicular hypoplasia may have been associated with an extra X chromosome. This abnormality resembles the Klinefelter syndrome occurring in humans. In man, some models of chromosome mosaicism, the most common of which is the XY/XXY group, are associated with male infertility (Ferguson-Smith, 1966).

The presence of two cell populations (XY and XXY) in the organism of the animal under study may be explained by at least two mechanisms; a) nondis-

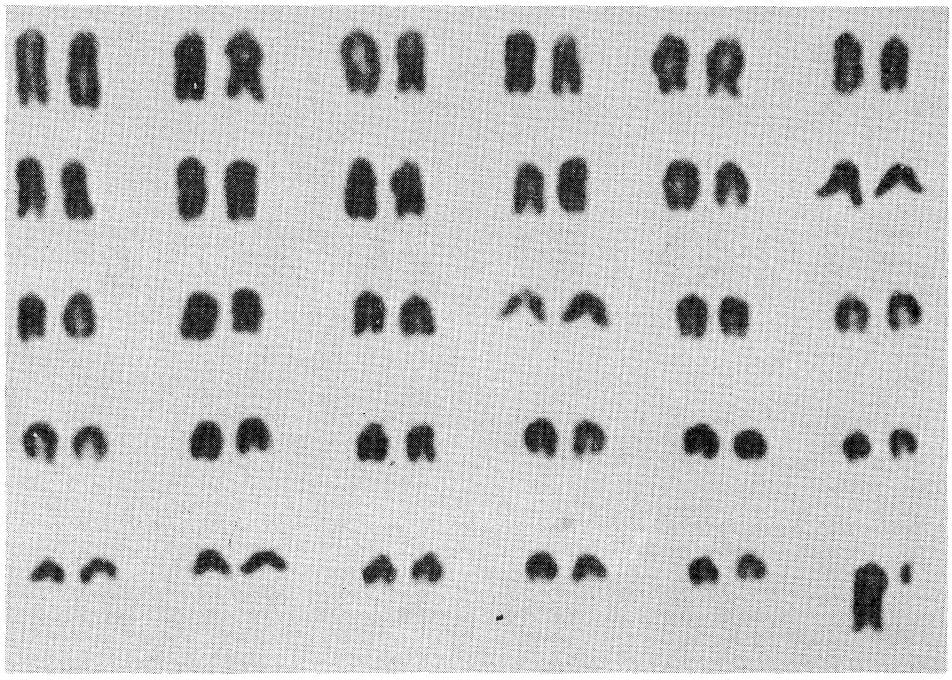

Fig. 2. Male karyotype (60, XY) of the mosaic buck.

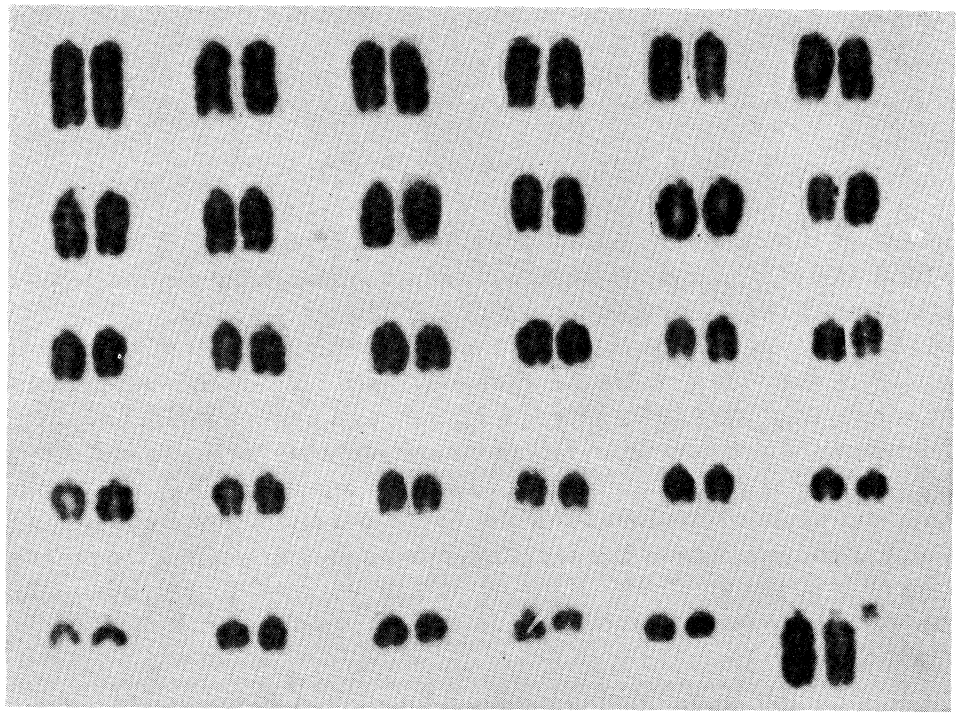

Fig. 3. The XXY karyotype in the buck with XY/XXY mosaicism. Note the typical minute short arm in the both X chromosomes.

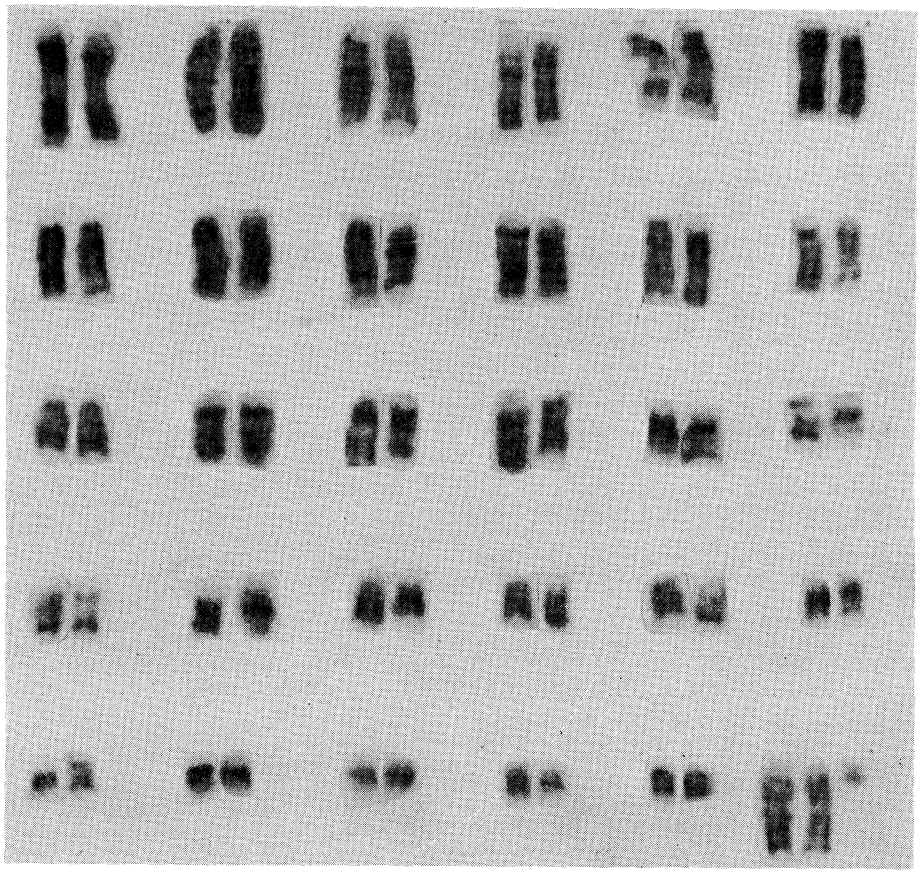

Fig. 4. Karyotype from G-banded metaphase cell.

junction of chromosome X during the first cell divisions of an XY zygote, or b) formation of XXY zygote owing to nondisjunction during meiosis, followed by the loss of one X chromosome during the first cell divisions.

The percentage of cells with 61 chromosomes was higher than that of cells with 60 chromosomes (Table 1). However, since only blood was examined, the values may have been different in the remaining tissues. The animal under study, which may be an intersex specimen with horns, should not belong to the intersex groups in which this pathology is linked to the hornless trait. Our hypothesis is that testicular hypoplasia and intersexuality may be related with mosaicism of the sex chromosomes, thus explaining the presence of horns in intersex animals. However, it is of essential importance to analyze a large number of horned animals with malformations of the reproductive system.

REFERENCES

FERGUSON-SMITH, M. A. (1966) X-Y chromosomal interchange in the aetiology of the true hermaphroditism and for XXY Klinefelter's syndrome. *Lancet* **27**, 475–476.

BONGSO, T. A., THAVALINGAM, M. and MUKHERJEE, T. K. (1982) Intersexuality associated with XX/XY mosaicism in a horned goat. *Cytogenet. Cell Genet.* **34**, 315–319.

MOORHEAD, P. S., NOWELL, P. C., MELLMAN, W. J., BATTIPS, D. M. and HUNGERFORD, D. A. (1960) Chromosome preparations of leucocytes cultured from peripheral blood. *Exp. Cell Res.* **20**, 613–616.

PADEH, B., WYSOKI, AYALON, N. and SOLLER, M. (1965) An XX/XY hermaphrodite in the goat. *J. Med. Sci.* **1**, 1008–1012.