SHORT PAPER

A phenotypically normal cattle with 60, XY/61, XYY karyotype

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ABSTRACT

Cytogenetic examination was performed on a Japanese Black bull and his eight offsprings. Out of nine cattle examined, eight animals showed normal male karyotype with 60, XY. However, one offspring had a normal male cell line (60, XY) and a cell line characterized by the karyotype (61, XYY). The frequencies of cells with XY and XYY sex chromosome complements were 76.5% and 23.5%, respectively. This animal had a normal male conformation and no physical abnormalities were detected.

The presence of an XY/XYY karyotype appears to be extremely rare in any mammal. With the exception of cases in man (Melnik *et al.* 1969; Jacobs *et al.* 1974; Hamerton *et al.* 1975), only two cases have been reported in a Bulgarian Brown cattle (Dobryanov and Konstantinov 1970) and a Siamese-crossbred cat (Loughman and Frye 1974). The XY/XYY individuals had a normal male conformation and no physical abnormalities, but histopathologic examination of the testicular tissue revealed the absence of germinal cell stages in the cat. Also, the XYY condition in man (Zeuthen *et al.* 1973; Skakkebaek *et al.* 1973) and mice (Cattanach 1961; Russel and Chu 1961; Evans *et al.* 1978) is frequently associated with reduced fertility. This paper presents additional evidence on a 60, XY/61, XYY karyotype detected in a Japanese Black cattle.

Case history: To test the performance of meat production of the Japanese Black bull A, eight half-sib male offsprings were randomly selected from the progeny population sired by the bull A. They were castrated at about six months of age. After the completion of the progeny test conducted for 52 weeks, all offsprings were slaughtered to evaluate their growth rate, feed efficiency and carcass quality. A few days prior to slaughter, cytogenetic survey was performed on the bull A and his offsprings. The present case was one of these offsprings.

Cytogenetic studies: Blood samples were taken from the jugular vein in

heparinised syringe, and cultured *in vitro* for 48 h according to the method described previously (Hanada and Muramatsu 1980). Air-dried chromosome slides were prepared by the conventional methods. Thirty well-spread metaphases from each animal were microphotographed for chromosome analysis. An additional 170 metaphases were examined in the subject animal.

The present case had a normal male conformation and no decrease in the performance of meat production was noted. The results of chromosome analysis are shown in Table 1. The animal had a bimodal distribution different from the single mode with 60 chromosomes in domestic cattle (*Bos taurus*). Of 200 cells examined, 148 had a normal chromosome number, $2n=60$. However, most of the remaining cells were hyperdiploidy with 61 chromosomes, including the X, Y and small submetacentric chromosomes. As shown in Figure 1, the extra chromosome was readily identified as the Y chromosome by its size and characteristic features. These hyperdiploid cells were found to have a 61, XYY karyotype. Seven cells with 59 or fewer chromosomes showed the XY or XYY sex chromosome complement, and no XO cell line was detected in the animal. It is considered that all hypodiploid cells appeared due to random loss of chromosomes as a technical artifact at the preparation. The frequencies of cells with XY and XYY sex chromosome complements were 76.5% and 23.5%, respectively. This was an extremely rare case as both 60, XY and 61, XYY karyotypes could be found in the leucocytes.

The sire and the other offsprings had a normal male conformation and karyotype. The present case had no significant family history of defective reproductive performance or abnormal phenotypic appearance.

In cattle, only two cases of 60, XY/61, XYY karyotype have been documented: the male calf of the Bulgarian Brown cattle breed reported by Dobryanov and Konstantinov (1970) and the present case. Both animals had a normal

Table 1. *Distribution of chromosome number in the peripheral lymphocytes of the present case, the relatives and the other normal Japanese Black cattle*

	No. of cells analysed	Chromosome number					Freq. of aneuploidy (%)
		≤58	59	60	61	62≤	
Present case*	200	2	5 (1)	148 (2)	45 (45)	0	26.0
Seven half-sibs	210	1	6	202	1	0	4.0
Sire	30	0	2	28	0	0	6.7
Spontaneous incidence in the same breed**	1900	11	34	1834	17	4	3.5

* Figures in parentheses indicate the number of cells with 61, XYY sex chromosome complement.

** Hanada and Muramatsu (1980).

Fig. 1. Metaphases and karyotype from the animal studied. Arrow shows the Y chromosome.

a: Metaphase of a cell with 60, XY chromosome complement. b-d. Metaphases of a cell with 61, XYY chromosome complement. e: Karyotype of the cell (b) with an extra Y chromosome.

male configuration and no congenital malformations, as in man (Jacobs *et al.* 1974; Hamerton *et al.* 1975). Loughman and Frye (1974) detected such a karyotype in a male Siamese-crossbred cat. The cat was phenotypically normal, but germinal cell stages were absent. Some impairment of spermatogenesis

was also observed in the majority of the cases reported in man (Zeuthen *et al.* 1973; Skakkebaek *et al.* 1973) and mice (Cattanach 1961; Russel and Chu 1961; Evans *et al.* 1978) with the XYY karyotype. The extra Y chromosome which appears to have no significant effect on the phenotype, is frequently associated with a reduction in reproductive performance. Unfortunately, no information about fertility and sex chromosome behavior at meiosis was obtained in the present report because the subject animal had been castrated before slaughter.

Possible mechanisms of the occurrence of the 60, XY/61, XYY in cattle are as follows: (1) Fusion of two male zygotes early in embryogenesis, and subsequent non-disjunction in either zygote and exclusion of the XO cell line; (2) Simultaneous implantation of an XY zygote and an XYY zygote resulting from paternal non-disjunction at meiosis II, and exchange of undifferentiated cells between heterozygous twins by means of chorionic vascular anastomoses and subsequent early death of a co-twin in utero; (3) Postzygotic non-disjunction in an XY zygote, either in favor of the XYY cell line or exclusion of the XO cell line.

The first and the second hypotheses would lead to the formation of a chimera, while the third hypothesis would induce a mosaic. It seems clear that the XYY cell line must have arisen through non-disjunction of the Y chromosome during the meiosis or mitosis. However, much more evidence is needed before the choice of one of these or any of numerous other hypotheses.

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