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THE ABERRANT KARYOTYPE OF A BULL WITH CHARACTERISTICS OF KLINEFELTER'S SYNDROME (Part I)

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In this study the appearance of an aberrant karyotype with the characteristics of Klinefelter's syndrome in a Simmental crossbred bull was described. Bilateral testicular hypoplasia and depressed libido were observed in this bull. The results of cytrogenetical analyses showed that a chromosome complement of 61, XXY was found in the investigated animal. It was concluded that the aberrant karyotype originated from the time of fertilisation and resulted from union of a chromosomally abnormal gamete, with a normal gamete.

Key words: Karyotype, Klinefelter's syndrome, chromosomal aneuploidies, testicular hypoplasia.

INTRODUCTION

One widespread problem which appears in all domestic mammals is reproductive inefficiency. It is caused by many factors and different chromosomal abnormalities are only one group of them. Cytogenetical examinations carried out in cattle and other domestic species showed that structural chromosome aberrations were more frequent compared to numerical changes. The last of them are not inherited due to sterility of the carriers (Switonski et al., 1991).

Different types of sex-chromosomal aneuploidies were described by many authors in cattle with fertility problems (Rieck, 1970; Bouters et al.,1974a; Dun et al., 1977; Malek et al., 1984; Soldatović et al., 1986a; 1986b; 1986c; Soldatović et al., 1989 etc.). Cell lines with a 61, XXY chromosomal complement were detected in numerous animals. In man, such sex — chromosomal aneuploidies (e. g. 47, XXY or 46, XY / 47, XXY) are related with Klinefelter's syndrome. Bilateral testicular hypoplasia with aspermatogenesis and depressed libido are the main characteristics of this syndrome (Federman, 1967).

MATERIAL AND METHODS

The animal investigated in this study was an 18 month old Simmental crossbred bull with the diagnosis of bilateral testicular hypoplasia. The general methods of clinical inspection established that it was well fed, its weight was

488 kg but its constitution was tender. The bull showed depressed libido. Its testes were under developed and aspermia was present. The young bull directed to a slaughter house where blood and testicular tissue for a cytogenetical examination and histological investigation were sampled.

Metaphase cells were obtained throught short term cultures of lymphocytes from peripheral blood (Halnan, 1977) and monolayer cultures of fibroblasts from the testis (Ford et al., 1956; Evans et al., 1964).

RESULTS AND DISCUSSION

Based on the histological investigation it was observed that the testes were filled with fibrous and adipose tissue. Spermatogenesis was not present in testicular tissue.

In 97.01% of all cytogenetically analyzed cells (134 metaphases) a 61, XXY chromosomal complement was observed. However, in 2, 99% of all metaphases a deficiency of Y-chromosome was detected (Table 1). Those cells had a 59, XO chromosomal complement. However, the simultaneous appearance of these two different sex-chromosome aneuploidies cannot be considered as mosaicism. This presumption is based on the fact that the Y-chromosome is very small and its loss during the methodological procedure of metaphase preparation was possible. Moreover, this is very frequent in the case of other small autosomes, too.

Table 1. Karyotypic changes in the investigated Simmental crossbred bull

Type of cells	Observed metaphases		Cells with karyotypic designation 61, XXY		Cells with structural chromosome aberrations	
	No	%	No	%	No	%
Lymphocytes	56	100.00	54	96.42	16	28.57
Fibroblasts	78	100.00	76	97.43	8	10.26
Σ	134	100.00	130	97.01	24	17.91

Switonski et al., (1991) explained that aneuploidies are not inherited and cannot be spread in a certain population. They assumed that all identified cases of aneuploidy were of de novo origin. Based of these considerations it was not possible that the parents of the problematical bull were carriers of sex-chromosome aneuploidies. Therefore, it was very probable that the aneuploidy observed in this study (61, XXY) originated at the time of fertilisation. Thus, the aneuploid adult developed from an aneuploid embryo which originated from an aneuploid zygote. The aneyploid zygote resulted from the union of a karyotypic normal gamete with another that was aneuploid for the sex-chromosomes. Further, it can be considered that the aneuploid gamete resulted from the failure of the sex-chromosomes to separate during the first or second meiotic division at the time of gametogenesis.

Twenty-four cells (17.91%) of all metaphases analyzed had structural changes in the chromosomes which were classified as chromatid and isochromatid breaks. However, cells from short term cultures of lymphocytes had a higher percentage (28.57%) of structurally demaged chromosomes than cells from monolayer cultures of fibroblasts (10.26%).This can be explained by the selection effects as well as the development requirements of monolayer cultures of fibroblasts. Fibroblasts take a longer period of time to develop in cultures than lymphocytes. Therefore, many karyotypically aberrant fibroblasts are unable to survive and die.

As the investigated bull came from an area where different types of agricultural and industrial pollutants were frequently used, it was possible that the structural chromosome damage resulted from their influence (Soldatović et al., 1989).

CONCLUSION

Based on data from the literature and the results obtained in this study it can be concluded that the aberrant karyotype of the investigated bull (61, XXY) originated from the union of aneuploid gametes at the time of fertilization.

We consider that the observed structural aberrations of chromosomes may have been the consequence of the influence of various chemical pollutants on this bull.

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ABERANTNI KARIOTIP KOD BIKA SA KARAKTERISTIKAMA KLINEFELTEROVOG SINDROMA (I deo)

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SADRŽAJ

Ovaj rad predstavlja prikaz pojave aberantnog kariotipa kod bika u tipu domaćeg simentalca sa karakteristikama Klinefelterovog sindroma. Kod ispitivanog bika zapažen je smanjeni libido, kao i bilateralna hipoplazija testisa. Rezultati citogenetskih analiza metafaznih figura dobijenih iz kulture limfocita i fibroblasta testisa ispitivane životinje ukazuju na prisustvo hromozomskog seta sa karakteristikom 2n=61, XXY.

Na osnovu literaturnih podataka i naših dobijenih rezultata može se zaključiti da je aberantni kariotip 61, XXY, nastao u procesu fertilizacije spajanjem aberantnog gameta sa normalnim gametom.