

## ROBERTSONIAN FUSION IN A SIMMENTAL COW-BULL MOTHER (Part II)

B. SOLDATOVIĆ\*, Z. STANIMIROVIĆ\*, MARIJANA VUČINIĆ\*\*, D. ĐOKIĆ\*, and MARIJANA VUČIĆEVIĆ\*

*\*Department of Biology, Faculty of Veterinary Medicine, Bulevar JNA 18, 11000 Belgrade, Yugoslavia*

*\*\*Institute for Animal Production, Faculty of Agriculture, Nemanjina 6, 11080 Zemun, Yugoslavia*

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The aim of this work is to present one of the current problems which frequently accompanies high production in modern cattle management. It is the appearance of Robertsonian translocations in the cattle karyotype.

Cytogenetical examination was performed on a 4 year old Simmental cow which was phenotypically normal and had normal fertility and high milk production. On the basis of these characteristics this cow had been chosen as a bull mother. However, all of its three pregnancies ended by the birth of an avital calf. They died 30 minutes after births. This was the reason for a karyotypic analysis of the problematic cow. A heterologous Robertsonian fusion of two acrocentric autosomes was found. The conclusion is that the Simmental Cow which attracted our attention was a carrier of balanced karyotypic changes that could be transmitted to the offspring and eventually spread all over the population.

Key words: Robertsonian fusion, Chromosomes aberrations, Cytogenetics, Simmental cow-bull, reproduction.

### INTRODUCTION

The results of intensive cytogenetical investigations suggest that different types of structural and numerical aberrations of cattle chromosomes frequently result in alterations of reproductive functions (Gustavsson, 1969; Balzak and Eldridge, 1977; Gustavsson, 1977; Popescu et al. 1984; Swartz i Vogt, 1983; Soldatović et al., 1986a, b, c; Soldatović et al., 1989; etc). These disorders of reproductive performance usually range from low fertility, abortions, stillbirths, early and sudden death of calves to births of offspring with congenital malformations.

Prophylactic measurements consisting of cytogenetical monitoring attach crucial importance to the detection of those individuals-carriers of balanced changes in cattle karyotypes. Phenotypically the cows are normal but they could transmit the chromosomal aberrations present in their karyotypes to their offspring and spread them within the population. These carriers of balanced karyotypic changes must be eliminated from reproduction at the proper time.

In this way, it will be possible to prevent the beginning of a drastic gene des-balance in certain populations of cattle (Ernest and Zhigachev, 1990).

#### MATERIAL AND METHODS

One of the milk producers from Mladenovac had a four year old Simmental cow. It was phenotypically normal and had normal fertility and high milk production. However, all of its three pregnancies ended by the birth of a calf that was unable to survive. The calved died 30 minutes after birth. The results of clinical inspection, rectal palpation and microbiological examinations were pathologically negative. These methods were unable to detect all disorders that could be considered as causes of calf mortality. This was the reason for the karyotypic test of this cow.

The karyotypic test employed short term cultures of lymphocytes and was performed according to the method of Moorhead et al. (1960), using whole peripheral blood. Blood samples were taken by jugular vein puncture of the investigated cow and its mother. A large number of metaphases was examined for each cow and the best samples were chosen for the karyotypic test (74 for the investigated cow and 62 metaphases for its mother).

#### RESULTS AND DISCUSSION

The presence of one large submetacentric autosome unusual for the cattle karyotype was observed on all cytogenetically tested metaphases of the problematic cow. At the same time the deficiency of two acrocentric autosomes was detected in all metaphases. Thus, the changed diploid number of chromosomes was  $2n=59$ , but, NF was 62. This cytogenetical finding suggested that the present karyotypic aberration of the problematic cow was a heterologous

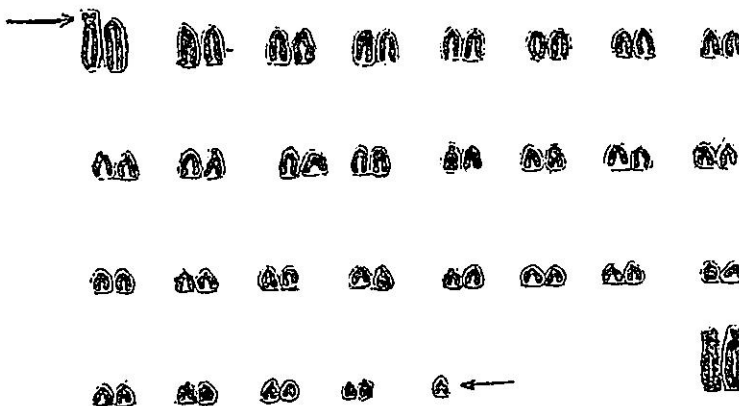


Figure 1. Robertsonian fusion in a Simmentaleow

Robertsonian translocation of two acrocentric autosomes. However, a identification of two translocated autosomes by the banding technique was not possible because the investigated cow was taken to a slaughter house.

The karyotype of its mother was also examined. It was normal without any structural or numerical changes of the chromosomes. The diploid number of its chromosomal set was  $2n=60$  and all autosomes were acrocentric. The appearance of a few aneuploid cells could be considered as a methodological error during the procedure of lymphocyte preparation.

On the basis of the detected chromosomal anomalies including the history of calvings and the calfs mortality, it was clear that the problematic cow was the carrier of a balanced change a heterologous Robertsonian translocation. This karyotypic anomaly did not cause phenotypical changes. Moreover, high milk production was an important characteristic of this cow, so that it was chosen as a bull mother. Never the last difficulties appeared at its calving. All of its three calves died at birth.

Since its mother had a normal chromosomal set, we could consider that the transformed karyotype-Robertsonian translocation originated from a de novo process in the gametogenesis of this cow. However, it was not possible to eliminate the possibility that the transformed karyotype had been inherited from the father. In this case, the father would have had the same type of karyotypic changes as its daughter and had to be a balanced carrier, if this was true then the father would have had reproductive problems but they were not registered.

Theoretically, this cow, as the carrier of the karyotypically change-heterologous Robertsonian translocation, could form 6 different types of gametes: a, b, c, d, e and f (Figure 2.). One sixth of the gametes (a) would be karyotypically normal. One sixth of the gametes (d) would possess the balanced karyotypic change and it could be expected that calves developed from them would be phenotypically normal, as their mother. However these

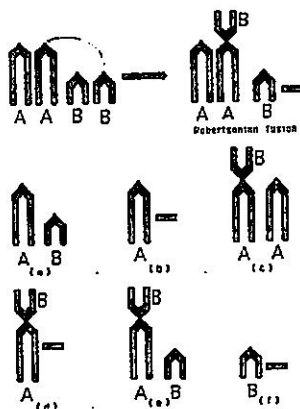


Figure 2. Types of gametes that could be formed in the progress of gametogenesis

calves would transmit that karyotypic aberration to their offspring. However, 4/6 of the gametes might form unbalanced zygotes (b, c, e and f). The chromosomal set of gametes b and f would be reduced by one autosome. Their zygotes would be monozomic, avital and not able to survive. In the process of fertilisation gametes c and e would form trisomic zygotes, also unable to survive. The consequences of a trisomy could be early death of the embryo or birth of a congenitally malformed calf.

Based on these considerations it could be expected that only 1/3 of the calves would possess the balanced karyotype. One sixth of the calves would be karyotypically and phenotypically normal but one sixth of the calves would be carriers of the balanced karyotypic change Robertsonian translocation as their mother.

Data obtained from the literature (Gustavsson, 1969; Balzak and Eldridge, 1977; Gustavsson, 1977; Popescu et al. 1984; Swartz i Vogt, 1983; Soldatović et al., 1986a, b, c; Soldatović et al., 1989; etc) also suggested that Robertsonian translocations in cattle frequently resulted in various reproductive disorders.

#### CONCLUSION

The karyotypic test of 74 metaphases of the investigated cow detected the presence of a heterologous Robertsonian fusion of two acrocentric autosomes. The types of gametes that could be formed in the gametogenesis process and the types of zygotes that could be developed from these gametes were predicted theoretically.

Based on these considerations it could be concluded that the investigated cow which attracted our attention was the carrier of a balanced karyotypic change. Its reproductive performance should have been reduced by 2/3 but, 1/3 of its calves would be phenotypically normal. Among the phenotypically normal calves 1/2 would be carrier of the same balanced karyotypic change as their mother.

This work also emphasizes the great importance of cytogenetical detection of those individuals that could spread balanced karyotypic changes all over the population.

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#### ROBERTSONOVA FUZIJA U KARIOTIPU SIMENTALSKE KRAVE-BIKOVSKJE MAJKE

B. SOLDATOVIĆ, Z. STANIMIROVIĆ, MARIJANA VUČINIĆ, D. ĐOKIĆ I MARIJANA VUČIĆEVIĆ

#### SADRŽAJ

Ovaj rad predstavlja prikaz jednog od aktuelnih problema koji učestalo prate visoku proizvodnju u govedarstvu, a odnose se na pojavu Robertsonovih translokacija u kariotipu govečeta.

Citogenetička ispitivanja sprovedena su na kravi simmentalske rase staroj četiri godine. Ova krava odlikovala se normalnim fenotipskim karakteristikama, između ostalog, normalnom plodnošću i visokom mlečnošću. Upravo na osnovu ovih osobina krava je odabrana za bikovsku majku. Naime, telad su uginjavala 30 minuta nakon rođenja. To je bio razlog da se pristupi analizi kariotipa ove krave. Analizom kariotipa otkriveno je prisustvo heterologe Robertsonove fuzije dva akrocentrična autozoma.

Na osnovu izvršenih analiza može se zaključiti da problematična krava simmentalske rase predstavlja nosioca balansirane kariotipske promene, koja se može prenositi na potomstvo, te na taj način širiti kroz određenu populaciju.