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Robertsonian Translocation and Its Effect on Fertility in Japanese Black Cattle

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Abstract

To confirm the distribution of chromosome abnormalities and their effects on fertility, a cytogenetic survey was carried out in five different herds of Japanese Black Cattle. The chromosome abnormality most frequently observed was the 7/21 Robertsonian translocation. This translocation was observed in a local herd at a fairly high frequency of 10.9% due to continuous use of the carrier bulls in artificial insemination. Analyses of breeding data failed to detect any significant difference in fertility between the normal animals and those with the 7/21 translocation. Semen characteristics of the carrier culls with the 7/21 translocation appeared normal. However, meiotic studies on the heterozygous bulls revealed the formation of aneuploid secondary spermatocytes from adjacent segregation. Trisomic embryos were detected in the cytogenetic study of embryos sired by the heterozygous bulls with the 7/21 translocation. The 7/21 translocation in the heterozygous state seemed to be associated with a slight reduction in fertility. Also, the 1/29 Robertsonian translocation, which has been reported in many breeds throughout the world, was widely distributed in Japanese Black Cattle.

Discipline: Animal industry
Additional key words: reduced fertility

Introduction

Loss of fertilized ova occurs most frequently during the embryonic and fetal stages of development in domestic animals. Approximately 20% of the total embryonic and fetal loss is due to chromosome abnormalities, which are one of the major causes of reproductive inefficiency[14]. If chromosome abnormalities were associated with reproductive failure and were heritable, they could cause an even greater economic loss in Japanese Black Cattle because artificial insemination is widespread.

Considering the effect on fertility and hereditary transmission, the most significant abnormality in cattle may be the Robertsonian translocation. Though the Robertsonian translocation is not associated with malformation, it appears to be inherited in a Mendelian fashion and often leads to early embryonic loss.

In cattle, the Robertsonian translocation was first reported as the 1/29 translocation in the Swedish Red and White breed[4]. Since then, the same or similar types of Robertsonian translocations have been detected in other breeds. In Japan, the 1/29 and the 7/21 translocations were found in Japanese Black Cattle[7,16,17]. Several studies suggest that the 1/29 translocation may lead to a slight reduction in fertility[5,18,23]. However, the effect of the 7/21 translocation on fertility is still unknown.

This paper describes the results of a cytogenetic survey conducted in Japanese Black Cattle, especially the distribution of the 7/21 Robertsonian translocation and its effect on fertility.

Animals and cytogenetic analyses

The karyotype analyses were carried out in a total of 768 animals, 86 bulls and 682 cows, belonging to five herds of Japanese Black Cattle. Mitotic chromosomes were prepared using the whole blood culture method. Precise chromosome identification was attempted by using the G-band staining method. Heterochromatin was stained by using the BSG method. Testicular materials for meiotic studies were obtained from eight Japanese Black bulls at slaughter. Embryos for chromosome analyses were produced by the *in vitro* fertilization method described previously[3]. The chromosome slides were prepared using the gradual fixation-air drying method.

Distribution of the 1/29 and the 7/21 Robertsonian translocations

The results of karyotype analyses are shown in Table 1. In herd A, 10 out of 394 cows examined were 1/29 translocation carriers and 43 were 7/21 translocation carriers (Plate 1). One animal was a double heterozygote carrying the 1/29 and the 7/21 translocations. The frequencies of 1/29 and 7/21 translocation carriers were 2.5 and 10.9%, respectively. It was confirmed by pedigree analyses and karyotype analyses of the carrier bulls and their daughters that one bull heterozygous for the 1/29 translocation and eight bulls carrying the 7/21 translocation had been employed for artificial insemination. One of the bulls with the 7/21 translocation was a homozygote with 2n = 58, XY, t(7q 21q) t(7q 21q). All the bulls carrying the 1/29 and the 7/21 translocations have been retired, and their frequencies have tended to decrease since 1981[9]. However, the 1/29

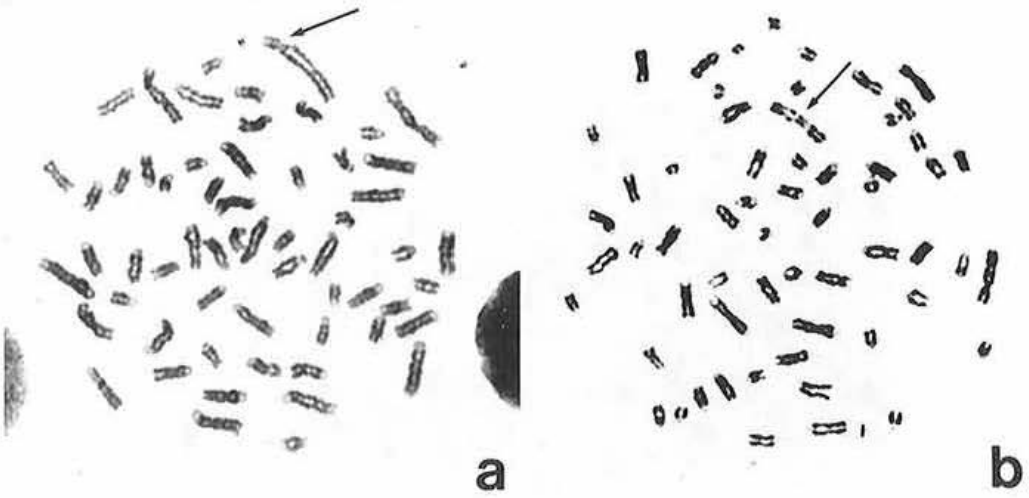

Plate 1. G-banded metaphases of cells carrying the 1/29(a) and the 7/21(b) translocation
Arrow shows a translocation chromosome.

Table 1. Frequencies of the 1/29 and the 7/21 translocation carriers in Japanese Black cows

Karyotype[a)]	Herd					Total[b)]
	A	B	C	D	E	
60, XX, Normal	342	54	41	99	73	609 (89.3)
59, XX, t_1	9	1	3	2	1	16 (2.3)
59, XX, t_2	36	2	4	7		49 (7.2)
58, XX, t_2t_2	6			1		7 (1.0)
58, XX, t_1t_2	1					1 (0.1)

a): t_1; t(1q 29q), t_2; t(7q 21q).
b): Figures in parentheses show the percentage of the carriers.

and the 7/21 translocations were also observed in other herds though the frequencies were lower than that in herd A. These results indicate that the 1/29 and the 7/21 translocations were widely distributed in Japanese Black Cattle. In British White cattle[2)] and Barrosa Cattle[22)], the 1/29 translocation was observed with a higher frequency of more than 50% due to the continuous use of the carrier bulls in artificial insemination. It is thus possible to assume that the number of carrier animals may increase markedly in a local herd of Japanese Black Cattle.

Origin of the 1/29 and the 7/21 Robertsonian translocations

The 7/21 translocation was not observed in Mishima Cattle, a native breed in Japan, which mainly contributed to the development of Japanese Black Cattle[12)]. This translocation has not been reported in breeds other than Japanese Black Cattle. While, the 1/29 translocation has been found in many breeds throughout the world[6)], several breeds such as Brown Swiss Cattle and Simmental Cattle have been imported for the improvement of Japanese

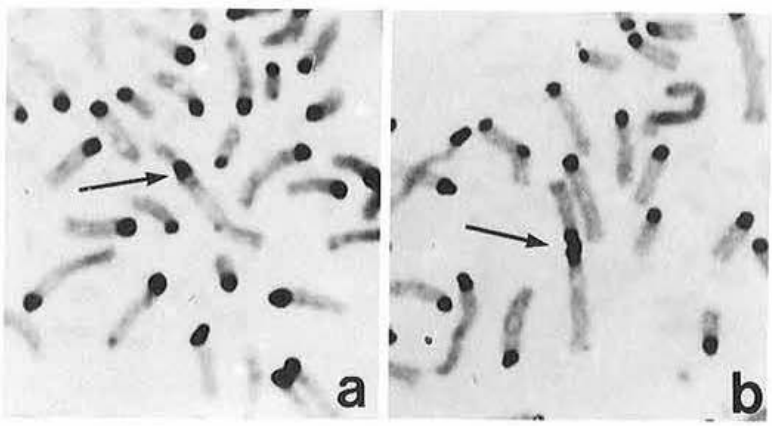

Plate 2. C-banding patterns of cells carrying the 1/29(a) and the 7/21(b) translocations
Arrow shows a translocation chromosome.

Black Cattle in Japan. Based on the C-banding analysis, it was confirmed that the 1/29 and the 7/21 translocations involved monocentric and dicentric chromosomes, respectively (Plate 2)[7]. The single C-band has been recognized by some researchers as evidence for an old, well-established translocation[13].

Based on its distribution and C-banding pattern, it is suggested that the 7/21 translocation has occurred recently in Japanese Black Cattle, while the 1/29 translocation has been introduced through the European breeds.

Effect of the 7/21 Robertsonian translocation on fertility

The analyses of breeding data failed to demonstrate any significant departure from normal fertility in the 7/21 translocation carrier animals. Also semen characteristics of the carrier bulls appeared normal. The seminiferous tubules seemed to be normally developed with a histologically normal spermatogenesis.

However, a trivalent configuration and cells with unbalanced karyotype were observed in the meiotic study of heterozygous bulls with the 7/21 translocation[8]. Out of 253 cells analyzed, 10 were hyperhaploid cells carrying an excess chromosome. The percentage of nondisjunction (N.D.) calculated from the frequency of the hyperhaploid cells was 7.9% in the heterozygous state as compared with 2.3% in the normal bull (Table 2)[10].

Then chromosome analyses were performed on 6–8 days old embryos (blastocysts) produced by *in vitro* fertilization in order to determine whether monosomic and trisomic embryos resulted from the fertilization of gametes with unbalanced karyotype. The results are shown in Table 3. In the group sired by normal bulls, all the embryos subjected to the analyses showed a normal chromosome complement, 2n = 60,XY and 2n = 60,XX. While, a total of 36 embryos were suitable for chromosome analyses in the group sired by the 7/21 translocation heterozygous bulls. Of these embryos, 19 had normal chromosome complements and 15 embryos showed a balanced karyotype with 59 chromosomes including the 7/21 translocation chromosome. However, 2 (5.6%) were trisomic embryos with the unbalanced karyotype, 2n = 60,XY,t and 2n = 60,XX,t[11].

In cattle, more information has been obtained in systematic studies on the 1/29 Robertsonian translocation. In the meiotic study on heterozygous bulls with the 1/29 translocation, secondary spermatocytes with unbalanced karyotype were observed with frequencies of 6.4%[15] and 11.2%[20]. Furthermore it was confirmed by the analyses of breeding data that the 1/29 translocation caused a slight reduction in

Table 2. Distribution of euploid and aneuploid cells in the meiosis of bulls carrying the 7/21 translocation

Karyotype[a]	No. of bulls	NF/2[b]			N.D.[c] (%)
		29	30	31	
60, XY	5	36	219	3	2.3
59, XY, t_2	2	32	211	10	7.9
58, XY, t_2t_2	1	7	32		0

a): t_2; t(7q 21q).

b): NF(Nombre fondamental); No. of chromosome arms.

c): $\text{N.D. (\%)} = \dfrac{2 \times \Sigma(n+1)}{\Sigma(n-1)+\Sigma n+\Sigma(n+1)} \times 100.$

Table 3. Chromosome analyses of embryos sired by the normal and the 7/21 translocation heterozygous bulls

No. of embryos	Karyotype of sire	
	Normal	Hetero.
Processed (A)	49	81
Analyzed (B)	23	36
Balanced karyotype		
Normal	23	19
Hetero.	–	15
Unbalanced karyotype		
Monosomy	–	–
Trisomy	–	2
B/A (%)	46.9	44.4

fertility of the carrier bulls[1] and daughters sired by the heterozygous bulls[5,18,23]. This phenomenon was ascribed to the early death of trisomic and monosomic embryos resulting from adjacent segregation of the trivalent chromosome at the first division[13,21,25]. The meiotic study revealed the presence of aneuploid secondary spermatocytes in heterozygous bulls with the 7/21 translocation. The meiotic behavior appeared to be similar to that of the 1/29 translocation. Trisomic embryos with unbalanced karyotype were also found in the cytogenetic study of embryos sired by the 7/21 heterozygous bulls. These results suggest that the 7/21 translocation in the heterozygous state may be associated with a slight reduction in fertility.

Conclusion

It was demonstrated that the 1/29 and the 7/21 translocations were widely distributed in Japanese Black Cattle. The presence of gametes and embryos with unbalanced karyotypes indicates the possibility of a slight reduction in fertility in the 7/21 translocation heterozygous animals. The 1/29 and the 7/21 translocations did not confer any advantage for well-defined characteristics. Moreover, the widespread and continuous use of carrier bulls in artificial insemination should not be encouraged. The importance of introducing eradication programs in cattle breeding has been demonstrated clearly for the Swedish Red and White breed[24] and routine investigations are now currently performed in several other countries, including Norway and Great Britain. It is important to introduce an eradication program for the 1/29 and the 7/21 translocations in Japanese Black Cattle.

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