Inheritance of the Robertsonian Translocation (1/21) in the Holstein-Friesian Cattle
I. Chromosome Analysis

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ABSTRACT. A new type of Robertsonian translocation (1/21) was found in Holstein-Friesian breed, and transmitted through the three generations, namely from the bull to 6 out of 11 daughters and 4 out of 7 grand-daughters/sons. Although the father of the bull was not available for cytogenetical studies, it was ascertained that 5 half-sibs of the bull had a normal karyotype. By C-banded karyotypes, the 9 translocation carriers within the three generations had a dicentric centromere in the translocated chromosome. It seems that no morphological effects have been noted to be associated with the translocation in all cases.—KEY WORDS: bovine, chromosome aberration, inheritance, Robertsonian translocation (1/21).

It has been reported that many types of chromosomal translocation frequently have negative effects on the reproductive performances in the domestic animals [3, 8, 26], and there are the inheritance for the translocation through the generations in some cases [4, 9, 14]. So, chromosomal translocations have gained an interest not only by cytogenetists in the field of domestic animals, but also by theriogenologists. Especially in bovine, the study of the 1/29 Robertsonian translocation reported by Gustavsson & Rockborn [11] was the trigger of subsequent findings of various types of chromosomal translocation in cattle [7, 17], because of the decrease in fertility. Now, it has been found in at least 38 breeds, distributed throughout the world, where cytogenetical studies have been made [4, 10]. However in the Holstein-Friesian breed, there are no reports about 1/29 translocation, and there is a few report about Robertsonian translocation involving the number 1 chromosome [4], and about the inheritance for any type of translocation.

Here we would like to report a new type of heritable Robertsonian translocation (1/21) through the generations in the Holstein-Friesian breed in Japan.

MATERIALS AND METHODS

A total of 24 Holstein-Friesian bulls, cows and heifers consisting of 3 generations were cytogenetically analyzed. It was composed of a bull (case 1), 5 half-sib bulls (cases 2–6), a son bull (case 7) and 10 daughter cows/heifer (cases 8–17) and 7 calves (cases 18–24) from the daughters of case 1 (Fig. 1).

For the purpose of chromosome analysis, the heparinized blood and a fragment of skin were aseptically collected, and were cultured in Eagle's MEM supplemented with fetal calf serum and phytohemagglutinin-M (2 µg/ml) (in the case of blood culture) at 38°C. At 72nd hours of blood culture, and at the time of the highest frequency in cell division of fibroblasts from the skin, the cultures were treated with colcemid (0.05 µg/ml). After the hypotonic treatment with 0.075 M KCl solution, chromosome preparations were made according to routine procedures. Over fifty metaphases per case were examined under a microscope and analyzed karyotypically. For further studies, the chromosomes were analyzed by G-banding [24] and C-banding [13] techniques.

RESULTS

The results of chromosome analysis from the blood and fibroblast cultures in the case 1 and the case 16 are shown in Table 1. In the case 1, the chromosome number of all 84 metaphases from blood samples was 59 which had one chromosome less than that in cattle (2n=60). All the 71 cells from fibroblasts showed the same karyotype as that in the results obtained from leucocytes. In the case 16 which was daughter cow of case 1, the total of 103 cells from leucocytes and 52 cells from fibroblasts had also the same karyotype as that in the father bull.
Table 1. Chromosome analysis of the bull and its daughter cow

<table>
<thead>
<tr>
<th>Cases</th>
<th>Number of cells with translocation (1/21)</th>
<th>Number of cells without translocation</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Leucocytes (%); Fibroblasts (%)</td>
<td>Leucocytes (%) ; Fibroblasts (%)</td>
</tr>
<tr>
<td>Bull (No. 1)</td>
<td>84 (100); 71 (100)</td>
<td>0 (0); 0 (0)</td>
</tr>
<tr>
<td>Daughter (No. 16)</td>
<td>103 (100); 52 (100)</td>
<td>0 (0); 0 (0)</td>
</tr>
</tbody>
</table>

Fig. 1. Heritable form of Robertsonian translocation (1/21) in Holstein-Friesian cattle
Number in circle or square shows the case number.

(case 1). In the metaphases stained with giemsa solution, there was a larger submetacentric chromosome than the X chromosomes, which was not observed normally (Fig. 2). There were none of cells with normal karyotype (2n=60, XX or XY) in any metaphases examined from both tissues of both cases. By G-banded staining, the abnormal submetacentric chromosome was identified to be formed by Robertsonian translocation between chromosome No. 1 and No. 21 (Fig. 3).

Although the father bull of case 1 could not investigated chromosomally, it was ascertained that 5 half-sib bulls of the case 1 by different mother cows had a normal karyotype (Fig. 1). The mother cow of case 1 could not examined chromosomally because of breeding in U.S.A.

Out of a son bull (case 7) and 9 daughter cows/heifer (cases 8–15, 17) excluding case 16, 5 daughters (case 10–13, 17) had a same Robertsonian translocation as that in the father bull (case 1) and a half-sib cow (case 16) in their blood samples. Furthermore, 4 calves, namely case 19 and 20 from the daughter cow (case 11), case 23 from case 13 and case 24 from case 16 showed the same type of translocation as that in the mother cows in their blood samples.

The centromeric regions of 1/21 translocated
HERITABLE TRANSLOCATION (1/21) IN CATTLE

Fig. 4. C-banded chromosome pair of 1/21 translocation (left) and No. 1 (right). The number shows Case 1 bull (1), daughters (cases 10–13, 16) and grand-daughter/sons (Cases 19, 20, 23). See Fig. 1.

chromosome were stained with C-banded technique, and had two dark dots in the bull (case 1), its daughters (case 10–13, 16) and 3 calves (cases 19, 20, 23) (Fig. 4).

It seemed that no morphological effects have been noted to be associated with the translocation in all the carriers.

DISCUSSION

The Robertsonian translocation is the most frequent type of chromosomal abnormalities found in cattle [4] and sheep [1]. And in the limited amount of research on chromosomes of cattle, over 25 types of Robertsonian translocation have been found in many breeds [4].

Although, out of many cases, the translocations involving the number 1 chromosome were found for 1/4, 1/23 and 1/28 in Czechoslovakian cattle [16], 1/25 in Simmental [25], 1/29 in Japanese Black [12, 18] and 1/29 in over 38 different breeds [4], only a few case had been reported until now in Holstein-Friesian breed [21].

Since it seems that the Robertsonian translocation caused by centric fusion between two acrocentric chromosomes transmit from heterozygous parents to their progenies in a standard Mendelian pattern as a dominant [4], it is expected that about a half number of offspring has a same type of Robertsonian translocation as that of ancestors. In the study of 5/21 translocation from Japanese Black by Masuda et al. [19], the 10 out of 15 offsprings from the bull with translocation showed the same type of translocation as that in the father bull, furthermore the one son bull out of them produced 8 offsprings with same translocation. And it is shown that there is the positive inheritance with apparent segregation in a 1:1 ratio in other translocation [9, 14, 20]. In this study, a half-sib offspring from 1/21 translocation bull had the same type of translocation, and three daughters with 1/21 produced 4 calves with same translocation. We have not been able to examine all calves which were produced from other presumable daughters with 1/21. However we are afraid that more offsprings from the bull and cows with 1/21 will have the same translocation, and they may be distributed over Japan with any effects as shown in the 1/29 translocation. King et al. [15] reported trisomic embryos sired by bull heterozygous for the 1/29 translocation as a cause of increased embryonic mortality. And Popescu [23] found 2 of 52 embryos lacking the No. 1 chromosome. However, it has not been clear about the possibility that the 1/21 translocation would be associated with the fertility, because the daughters are too young to estimate their fertility. This will be examined later.

By C-banded technique, all the carriers through the 3 generations had a dicentric chromosome, with two blocks of heterochromatin in the centromeric regions. In the case of 1/29 translocated chromosome, the single C-band observed has been accepted as evidence of an old, well-established translocation, whereas Robertsonian translocations of a more recent origin seem to have larger, or dicentric centromere by C-banding [2, 5, 6, 19, 22]. From the above concept, the 1/21 translocation in our cases may occur newly. If the size of heterochromatin stained by C-banding in translocated chromosome is the most noticeable characteristics for the origin of translocation, the dicentric centromere in our cases will be changed to monocentric one in the future. It will be reasonable to continue to investigate the nature of heterochromatin in our cases in vitro and in vivo.

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REFERENCES


