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A NEW CASE OF RECIPROCAL TRANSLOCATION rcp(13; 26) IN CATTLE

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S u m m a r y

The cytogenetic examination of 21 healthy phenotypically pedigree cows of the Black-and-White breed (3-6 month of third lactation) in farms of Novosibirsk oblast' revealed of animals-carriers of chromosomal rearrangement, a new type of reciprocal translocation rcp(13; 26). The cytological analysis of preparations found out the shorter considerably chromosome in karyotype of these animals. The event of inheritance of this mutation from parents to progeny was established.

Keywords: chromosome mutation, reciprocal translocation, Black-and-White breed, cattle.

Clinical cytogenetics of animals develops simultaneously with cytogenetic studies of a man (1). The most active researches were performed in late 1980ies. A large part of scientific reports about chromosomal mutations in animals is focused on assessing their effects on fertility. Today, it has been accumulated a sufficient data allowing to analyze the distribution of particular karyotype violations in different countries, breeds and populations.

There are two main groups of chromosomal abnormalities detected in cattle: chromosomal rearrangements (aberrations) and aneuploidy of sex chromosomes. The best studied and most frequent non-lethal karyotype violations in most cases are Robertsonian and reciprocal translocations that occur de novo or are inherited. The most common types of chromosomal aberrations is Robertsonian translocation resulting from a centric junction of two acrocentric non-homologous chromosomes and leading to the formation of metacentric or submetacentric chromosome. Cytogenetic studies conducted in different countries on different cattle breeds revealed the most frequent aberration – a centric fusion rob(1; 29) between the largest and the smallest chromosomes; this violation was observed for the first time in Swedish Red Pied cattle breed (2). The analysis of the available data showed significant differences in frequency of this type of Robertsonian translocation both between and within cattle breeds. More than 50 types of Robertsonian translocations are already known in cattle; the centric fusion was observed in all autosomes, but the 1st, 29th, 21st, 14th and 16th pairs are the most frequently involved in such structural rearrangements.

Recently, a special attention is paid to reciprocal translocations resulting in formation of a quadrivalent during the meiosis, which leads to production of unbalanced gametes. In such animals fecundity can be reduced by 50% and more. There are few reports about reciprocal translocations in cattle (Table 1). This mutation is referred to stable chromosome aberrations in most cases inherited over generations. Some types of reciprocal translocations contribute to phenotypic disturbances in animals. B. Mayr et al. (3) has reported about the case when artificial insemination with sperm of the bull-sire carrying rcp(10; 11) (41, 14) resulted in 30% cows returned to estrous after re-insemination, and in the case of the sire-carrier of rcp (8; 15) (21, 24) - in 25% cases (4). The translocations rcp(1, 8, 9) (q43; q13; q26) (5) and rcp(20, 24) (q17; q25) (6) in bulls-sires also resulted in re-return of inseminated cows to estrous in, respectively, 36,4 and 17,0% of cases. The carriers of translocations rcp(2q; 20q), t(8q; 27q) (7) and rcp (8, 13) (q11; q24) (8) were sterile males.

1. Types of reciprocal translocations recorded in cattle (literature data).

Translocation	Author of report	Year	Mutation carrier: breed, gender	Phenotypic manifestation
rcp(10; 11)(41; 14)	B. Mayr e.a. (3)	1979	?, bull	Reduced fecundity
t(2q-; 20q+), t(8q-; 27q+) (double translocation)	G.G. De Schepper e.a. (7)	1982	?, bull	Sterility
rcp(8; 15)(21; 24)	B. Mayr e.a. (4)	1983	Grey Alpine, bull	Phenotypically normal
t(X; 23)(p+; q□)	P.K. Basrur e.a. (9-11)	1992, 2001, 2001	Crossbred Limousine ½ Jersey, cow	Reduced fertility
rcp(1; 8; 9)(q43; q13; q26)	A. Kovacs e.a. (5)	1992	American Brown Swiss, bull	Reduced fecundity
rcp(1; 8)(?; ?)	K. Christensen e.a. (12)	1992	American Brown Swiss, bull	Reduced fecundity
rcp(20; 24)(q17; q25)	M. Andersson e.a. (13), D.A. Villagomez e.a. (6)	1992 1993	?, bulls	Reduced fecundity
rcp(8; 13)(q11; q24)	H.A. Ansari e.a. (8)	1993	?, bull	Sterile
rcp(X; 1)(42; 13)	B. Mayr e.a. (14)	1998	Simmental, heifer	Phenotypically normal
rcp(12; 17)(q22; q14)	A. Ducos e.a. (1)	2000	Montbeliarde, bull	Reduced fecundity
rcp(1; 5)(q21→qter; q11→ q33)	L. Iannuzzi e.a. (15)	2001	Grey Alpine, bull	Sterile
rcp(Y; 9)(q12.3; q21.1)	L. Iannuzzi e.a. (16)	2001	Chianina, bull	Sterile
rcp(11; 21)(q28; q12)	L. Molteni e.a. (17)	2007	Chianina, bull	Reduced proliferation of generative cells
rcp(9; 11)(q27; q11)	L. De Lorenzi e.a. (18)	2007	Marchigiana, bull	Reduced fecundity
rcp(2; 4)(q45; q34)	M. Switonski e.a. (19)	2008	Ayrshire, bull	Reduced fecundity
t(Y; 21)(p11; q11)	M. Switonski e.a. (20)	2011	Holstein-Friesian, bull	Phenotypically normal, reduced testosterone level
t(4; 7)(q14; q28)	L. De Lorenzi e.a. (21)	2010	Marchigiana, bull	Phenotypically normal

Along with inter-autosomal translocations, there are several examples of X-autosomal translocations (9, 10, 11, 14). An interesting fact was observed at cytogenetic examination of 50 Simmental twins - chimerical females (14). The 2-month-old heifer was a carrier of a chimerical germ cell XX:XY; all the germ cells with XX sex chromosome contained the reciprocal translocation rcp(X; 1) (42; 13), whereas in XY germ cells no alteration was found. This was a balanced translocation, and the heifer was healthy and phenotypically normal for its age.

The purpose of this work was identification of carriers of chromosomal abnormalities in populations of cattle and the analysis of detected mutations and their phenotypic effects.

Technique. During an annual cytogenetic monitoring in cattle farms of Novosibirsk oblast' (2008-2010), there were examined 21 pedigree Black-and-White cows (phenotypically healthy animals being at the 3rd-6th month of the 3rd lactation).

Chromosomes were recorded in cultured samples of peripheral blood leukocytes of the cows. Lymphocytes (0,5 ml) were cultured for 48 h in RPMI-1640 medium (5 ml) added with 15-20% autologous serum and concanavalin (0,1 ml). In 1,5 hours before the end of culturing, the medium was added with colchicine solution. Hypotonic treatment was performed using 0,56 M KCl solution for 20 min at 37 °C, then the cell culture was pre-fixed with subsequent fixation in methanol-acetic mixture. Chromosomes were routinely stained with Giemsa solution and examined under a microscope Biolar 1DP5AZ (Poland, magnification ×1000). G-banding was performed according to Seabright's technique (22) with modifications.

Results. During the primary cytogenetic monitoring of cattle populations, there was detected the cow carrying a chromosomal rearrangement (presumably, a reciprocal translocation) while a normal set of chromosomes *2n*=60 (Fig. 1).

During the period of economic use, this cow gave birth to two phenotypically healthy calves (bull-calf and heifer) and one pregnancy was interrupted by abortion. Animals-carriers of a translocation (especially reciprocal) in the heterozygous state during meiosis produce normal gametes along with gametes containing excessive or insufficient number of chromosomes; such chromosomal mutations are phenotypically manifested as increased fetal mortality.

It's notably that the heifer born by this cow has inherited the mother's mutation (Fig. 2).

Fig. 1. Metaphase plate of Black-and-White cow (2*n* = 60, XX) **the carrier of an aberrant chromosome** (circled). Giemsa stain, magnification ×1000.

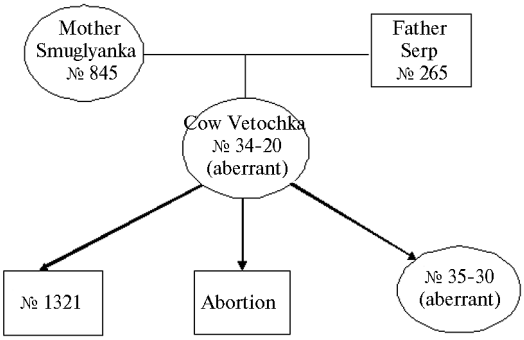

Fig. 2. Genealogical scheme of Black-and-White cow the carrier of aberrant chromosome

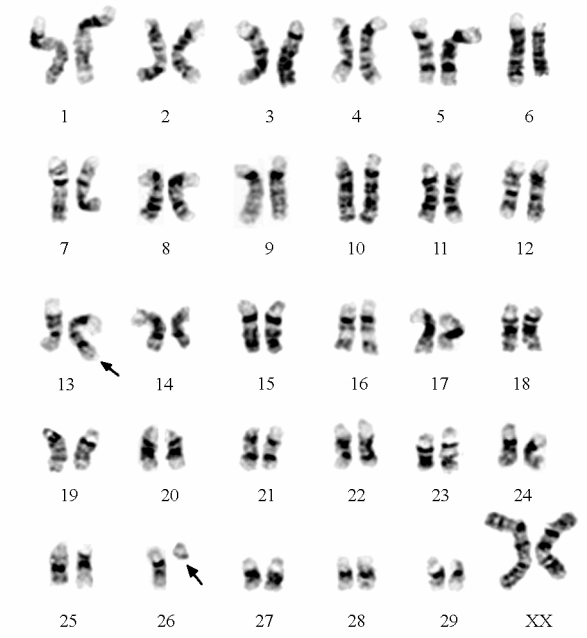

Fig. 3. GTG-stained chromosomes of cow Vetochka № 34-20 heterozygous for reciprocal translocation rcp(13; 26). Arrows show aberrant chromosomes. Magnification ×1000.

2. Number of cases when cattle chromosomes participate various types of reciprocal translocations

Number of cases	Pair of chromosomes, №
0	3; 6; 14; 16; 18; 19; 22; 25; 26; 28; 29
1	5; 7; 10; 12; 13; 15; 17; 23; 24; 27
2	2; 4; 20; 21; X; Y
3	9; 11
4	1
5	8

Unfortunately, examination of karyotype of the bull-calf born by this cow wasn't performed since the retirement of this calf from the herd. It is also unclear if this mutation was occurred de novo or inherited from one of the parents – a bull-sire Serp № 265 or from mother Smuglyanka № 845. Cytogenetic examinations of six daughters of this bull-sire showed the absence of this chromosomal aberration in their cells. Differentiated staining of chromosomes № 34-20 in lymphocytes obtained from the cow Vetochka revealed the rearrangement involving 13th and 26th pairs (Fig. 3). While the normal number of chromosomes (*2n* = 60), GTG-staining showed the presence of an aberrant chromosome whose size and stained bands suggested it as a product of the reciprocal translocation between 13th and 26th pairs of chromosomes. Another product of this rearrangement was a shortened chromosome of the 26th pair.

The cow Vetocka № 34-20 and its daughter (№ 35-30) were heterozygous carriers of this chromosomal aberration. At present time, there are being performed the attempts to establish breakage sites of chromosomes participating this translocation using various staining techniques.

Reciprocal translocations occur at a simultaneous breakage in two or more non-homologous chromosomes and a mutual exchange by resulting fragments having equal or different length. During a meiosis, homologous genes and blocks of genes conjugate, and if the presence of reconstructions, a normal shape of homologues-conjugants will be changed (23). As a rule, reciprocal translocations appear in the heterozygous state. In cattle karyotype, most of reciprocal translocations occur in autosomes (60 %) and in gonosomes (Table 2), which facts were discovered mainly because of their effects on carrier's fecundity (Table 1). It is important to highlight that carriers of these translocation, as a rule, were bulls. Probably, this fact is associated with a significant role of bulls-sires in formation of the gene pool of cattle populations and it highlights the importance of cytogenetic examination of bulls-sires used in artificial insemination.

The analysis of participation of various pairs of chromosomes in translocations suggests that autosomes of the 8^{th}, 1^{st}, 9^{th} and 11^{th} pairs are the most frequent participants in such violations, and then – chromosomes of the 2^{nd}, 4^{th}, 20^{th} and 21^{st} pairs and sex chromosomes. Therefore, some of the chromosomes are more predisposed to breakage and translocations. About 40 % autosomes are not involved in reciprocal translocations; at least there are no available reports about it. L. Molteni et al. (17) assumes a low rate of reciprocal translocations in *Bos taurus* as a result of acrocentric type of all its autosomes, so a routine staining doesn't allow revealing morphological changes in chromosomes except the cases when they can be visually detected (abnormally lengthened or shortened chromosomes).

The need for cytogenetic screening in populations of pedigree cattle is indicated by the following fact. In most countries, the frequency of chromosomal abnormalities in dairy cattle (eg, a common Robertsonian translocation between chromosomes of the 1^{st} and the 29^{th} pairs) significantly decreased, while in beef cattle no similar changes were observed (1). Moreover, beef cattle breeds show a trend to increased rate of reciprocal translocations (Table 1). Probably, this fact became the result of less accurate selection of pedigree animals, which confirms the importance of cytogenetic evaluation of these individuals (24).

Thus, the literature data and findings of this research suggest an association between the detected chromosomal violations and reduce in reproductive qualities of cattle. In some cases, animals - the carriers of such violations - can be clinically healthy, but they have significantly reduced fecundity owing to inferiority of gametes. It is possible to prevent the spread of chromosomal abnormalities in cattle population using cytogenetic certification of individuals, especially those involved in formation of pedigree bloodstock, new lines, types and breeds.

REFERENCES

1. Ducos A., Revay T., Covacs A., Hidas A., Pinton A., Bonnet-Garnier A., Molteni L., Slota E., Switonski M., Arruga M.V., Van Haeringen W.A., Nicolae I., Ghaves R., Guedes-Pinto H., Anderson M. and Ianuzzi L., Cytogenetic Screening of Livestock Populations in Europe: an Overview, *Cytogen. Genome Res.,* 2008, vol. 120, no. 1-2, pp. 26-41.
2. Gustavsson I., Distribution and Effects of the 1/29 Robertsonian Translocation in Cattle, *J. Dairy Sci*., 1979, vol. 62, no. 5, pp. 825-835.
3. Mayr B., Themessel H., Wöckl F. and Schleger W., Reziproke Translokation 60, XY, t(10; ll)(41; 14) beim Rind, *Z. Tierzucht. Züchtgsbiol*., 1979, vol. 96, pp. 44-47.
4. Mayr B., Krutzler H., Auer H. and Schleger W., Reciprocal Translocation 60, XY, t(8; 15)(21; 24) in Cattle, *J. Reprod. Fert.,* 1983, vol. 69, no. 2, pp. 629-630.
5. Kovacs A., Villagomez D.A.F., Gustavsson I., Lindblad K., Foote R.H. and Howard T.H., Synaptonemal Complex Analysis of a Three-Breakpoint Translocation in a Subfertile Bull, *Cytogen. Cell Gen*., 1992, vol. 61, pp. 195-201.
6. Villagomez D.A., Andersson M., Gustavsson I. and Plöen L., Synaptonemal Complex Analysis of a Reciprocal Translocation, rcp(20; 24)(q17; q25), in a Subfertile Bull, *Cytogen. Cell Gen*., 1993, vol. 62, pp. 124-130.
7. De Schepper G.G., Aalbers J.G. and Te Brake J.H., Double Reciprocal Translocation Heterozygosity in a Bull, *The Veterinary Record*, 1982, vol. 110, no. 9, pp. 197-199.
8. Ansari H.A., Jung H.R., Herdiger R., Fries R., König H. and Stranzinger G., A Balanced Autosomal Reciprocal Translocation in an Azoospermic Bull, *Cytogen. Cell Gen*., 1993, vol. 62, pp. 117-123.
9. Basrur P.K., Piheiro L.E.L., Berepubo N.A., Reyes E.R. and Popescu P.C., X Chromosome Inactivation in X Autosome Translocation Carrier Cows, *Genome*, 1992, vol. 35, no. 4, pp. 667-675.
10. Basrur P.K., Reyes E.R., Farazmand A., King W.A. and Popescu P.C., X-Autosome Translocation and Low Fertility in a Family of Crossbred Cattle, *Animal Reprod. Sci.,* 2001, vol. 67, no. 1, pp. 1-16.
11. Basrur P.K., Koykul W., Baguma-Nibasheka M., King W.A., Ambady S. and Ponce de León F.A., Synaptic Pattern of Sex Complements and Sperm Head Malformation in X-Autosome Translocation Carrier Bulls, *Mol. Reprod. Develop.,* 2001, vol. 59, no. 1, pp. 67-77.
12. Christensen K., Agerholm J.S. and Larsen B., Dairy Breed Bull with Complex Chromosome Translocation: Fertility and Linkage Studies, *Hereditas*, 1992, vol. 117, pp. 119-202.
13. Andersson M., Aalto J. and Gustavsson I., Embryo Quality and Andrological Study of Two Subfertile Bulls versus Five Control Bulls with Normal Fertility, *Theriogenology,* 1992,vol. 38, no. 4, pp. 623-631.
14. Mayr B., Korb H., Kiendler S. and Brem G., Reciprocal X;1 Translocation in a Calf, *Gen. Sel. Evol*., 1998, vol. 30, no. 3, pp. 305-308.
15. Iannuzzi L., Molteni L., Di Meo G.P., Perucatti A., De Lorenzi L., Incarnato D., De Giovanni A., Succi G. and Gustavsson I., A New Balanced Autosomal Reciprocal Translocation in Cattle Revealed by Banding Techniques and Human-Painting Probes, *Cytogen. Cell Gen*., 2001, vol. 94, pp. 225-228.
16. Iannuzzi L., Molteni L., Di Meo G.P., De Giovanni A., Perucatti A., Succi G., Incarnato D., Eggen A. and Cribiu E.P., A Case of Azoospermia in a Bull Carrying a Y-Autosome Reciprocal Translocation, *Cytogen. Cell Gen*., 2001, vol. 95, pp. 225-227.
17. Molteni L., Perucatti A., Iannuzzi A., Di Meo G.P., De Lorenzi L., De Giovanni A., Incarnato D., Succi G., Cribiu E., Eggen A. and Iannuzzi L., A New Case of Reciprocal Translocation in a Young Bull: rcp(11; 21)(q28; q12), *Cytogen. Genome Res*., 2007, vol. 116, pp. 80-84.
18. De Lorenzi L., De Giovanni A., Molteni L., Denis C., Eggen A. and Parma P., Characterization of a Balanced Reciprocal Translocation, rcp(9; 11)(q27; q11) in Cattle, *Cytogen. Genome Res*., 2007, vol. 119, no. 3-4, pp. 231-234.
19. Switonski M., Andersson M., Nowacka-Woszuk J., Szczerbal I., Sosnowski J., Kopp C., Cernohorska H. and Rubes J., Identification of a New Reciprocal Translocation in an AI Bull by Synaptonemal Complex Analysis, Followed by Chromosome Painting, *Cytogen. Genome Res.,* 2008, no.12, pp. 245-248.
20. Switonski M., Szczerbal I., Krumrych W. and Nowacka-Woszuk J., A Case of Y-Autosome Reciprocal Translocation in a Holstein-Friesian Bull. *Cytogen. Genome Res*., 2011, vol. 132, no. 1-2, pp. 22-25.
21. De Lorenzi L., Kopecna O., Gimelli S., Cernohorska H., Zannotti M., Béna F., Molteni L., Rubes J. and Parma P., Reciprocal Translocation t(4; 7)(q14; q28) in Cattle: Molecular Characterization, *Cytogen. Genome Res*., 2010, vol. 129, no. 4, pp. 298-304.
22. Grafodatsky A. and Radzhabli S., *Khromosomy sel'skokhozyaistvennykh i laboratornykh mlekopitauschikh: Atlas* (Chromosomes of Farm and Laboratory Mammalians: Atlas), Novosibirsk, 1988.
23. Dubinin N.P., *Obschaya genetika* (General Genetics), Moscow, 1986.
24. Kochneva M.L., Chromosomal Instability in Healthy and Anomalous Pigs, *S.-kh. biol.,* 2003, no. 2, pp. 69-72.