

Two abnormal chromosomes found in one cell line of a mosaic cow with low fertility

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Summary

A morphologically normal cow was cytogenetically investigated due to its inefficient reproduction. The female, 11 years old, had produced 3 calves with long birth to conception intervals characterized by the presence of follicular cysts. Chromosome complement was found to be 60, XX/60, XX, t (12 q ; 15 q), inv (6). Marker chromosomes of the abnormal line were monocentric. Blood typing showed that the origin of this line was not due to chimerism associated with vascular anastomosis. Insufficient data was available to associate statistically the chromosomal abnormalities with lowered fertility.

Key words : Abnormal chromosomes, chimerism, fertility, cow.

Résumé

Mise en évidence de deux anomalies chromosomiques dans une lignée cellulaire d'une vache à fertilité réduite

Cette étude cytogénétique concerne une vache morphologiquement normale mais à fertilité réduite. Cette femelle âgée de 11 ans, avait mis bas trois veaux espacés par de longs intervalles entre vêlage et fécondation ; elle présentait également des kystes ovariens. Le caryotype de cette vache était le suivant : 60, XX/60, XX, t (12 q ; 15 q), inv (6). Les chromosomes marqueurs de la lignée cellulaire anormale étaient des monocentriques. Dans cette lignée leucocytaire anormale, on a mis en évidence 2 remaniements, identifiables grâce aux mesures chromosomiques et aux bandes G et C, à une translocation robertsonienne 12-15 et une inversion péricentrique du chromosome 6. L'étude des groupes sanguins a révélé que cette lignée cellulaire ne résulte pas d'un chimérisme d'anastomose vasculaire. L'information recueillie n'était pas toutefois suffisante pour établir la relation entre ces anomalies et la chute de fertilité.

Mots clés : Anomalie chromosomique, chimérisme, fertilité, vache.

I. Introduction

Several structural and numerical chromosome abnormalities have been described in *Bos taurus*. The 1/29 translocation (GUSTAVSSON & ROCKBORN, 1964) is the only structural abnormality in which lowered fertility has unequivocally been demonstrated in heterozygous carriers (GUSTAVSSON, 1980). It was demonstrated that 56-day non-return rates of translocation female carriers were lowered (GUSTAVSSON, 1969; REFSDAL, 1976), daughter groups of heterozygous bulls had increased culling rates (GUSTAVSSON, 1971 a) and the incidence of 1/29 in a group of heifers diagnosed as repeat breeders was increased (GUSTAVSSON, 1971 b).

A definite relationship with female fertility has not been found in other structural abnormalities, namely the 2/4 (POLLOCK, 1972; POLLOCK & BOWMAN, 1974), 13/21 (HARVEY & LOGUE, 1975), 14/24 (DI BERARDINO *et al.*, 1979), 25/27 (DE GIOVANNI *et al.*, 1979), 5/21 (MASUDA *et al.*, 1980), and 7/12 (HANADA *et al.*, 1981).

In a cytogenetic survey of females with disturbed fertility, a cow with an abnormal complement was found. The karyologic study is described here.

II. Material and methods

One single-born, morphologically normal *Holstein-Friesian* cow was submitted to cytogenetic analysis due to its inefficient reproduction. The female was eleven years old and has produced three calves, with long birth to conception intervals. The *post-partum* periods were characterized by the presence of follicular cysts, and high milk production (mean = 7,800 kg). Cysts were treated after the first and second parturitions but treatment was unsuccessful after the third one and the animal was slaughtered. Cytogenetic analysis was carried out on two different occasions according to the technique described by HALNAN (1977) with some modifications. Lymphocytes were cultured for 72 hours. Mitosis was arrested adding vinblastine sulphate (Velbe, Eli Lilly Argentina) (1 μ g/ml) 45 minutes before sacrifice of the culture. Slides were air-dried and stained with 2 p. 100 Giemsa. G-bands were obtained by trypsin digestion (SEABRIGHT, 1971), and C-bands with the Ba(OH)₂ technique (SUMNER, 1972). Ten abnormal metaphases were photographed and chromosomes measured. Blood typing was performed in order to investigate the possibility of a secondary chimerism due to the presence of a foetus which died *in utero* (STORMONT, 1954).

III. Results and discussion

The chromosomal analysis showed the existence of two cell lines. One was normal (60, XX) and the other presented 60 chromosomes including a Robertsonian rearrangement and a chromosome presumably having a pericentric inversion (fig. 1). The

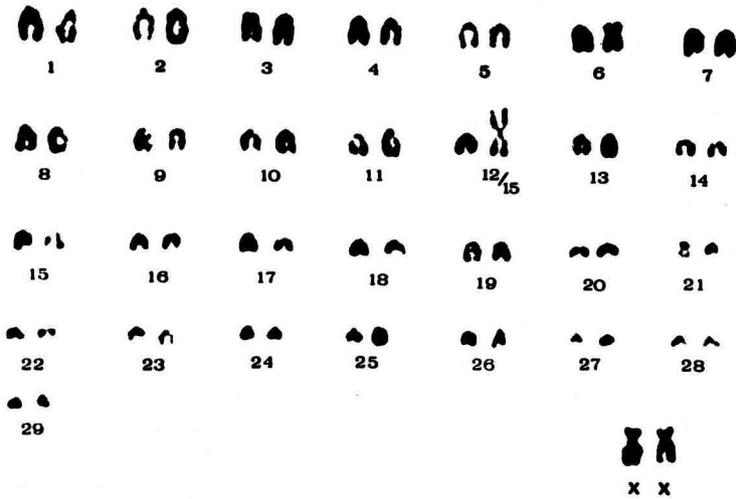


FIG. 1

*Giemsa stained karyotype of the abnormal cell line of the subfertile cow.
Caryotype de la lignée cellulaire anormale, coloré au Giemsa.*

two marker chromosomes were present in 17 (9.35 p. 100) of 182 metaphases examined. Measurements of chromosomes allowed location of the pericentric inversion in the sixth pair and the arms of the translocated chromosome were found to be between the 10-18th and 8-13th pairs for the short and long arms respectively. The comparison of G-banded karyotypes with the Reading standard (FORD *et al.*, 1980) showed that the short arm of the translocation corresponded to pair 15 while the long arm coincided with pair 12 (fig. 2). The chromosome with the pericentric inversion

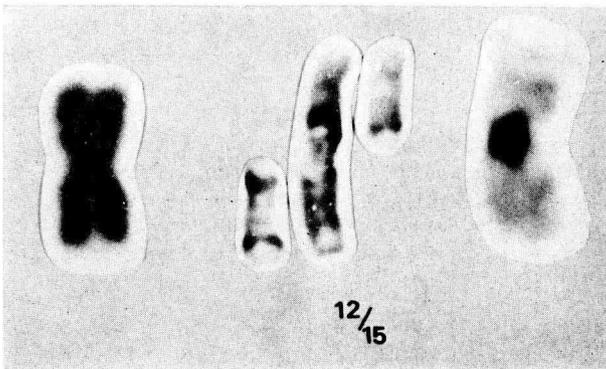


FIG. 2

Giemsa stained (left), G-bands (center), and C-bands (right) of the 12/15 translocation present in the abnormal line.

Translocation 12/15 caractérisant la lignée cellulaire anormale, coloré au Giemsa (à gauche), montrant le dessin des bandes G (au centre) et des bandes C (à droite).

could not be identified by G-bands. Thus, the chromosome complement of this animal could be described as 60, XX/60, XX, t (12 q; 15 q), inv (6). In addition, the abnormal line contained two chromosomes No. 15 and, therefore, could be assumed as trisomic for this pair.

The treatment with Ba(OH)₂ showed that both the translocated chromosome and the pericentric inversion presented a single block of centromeric heterochromatin, i.e. they were monocentric (fig. 2 and 3).

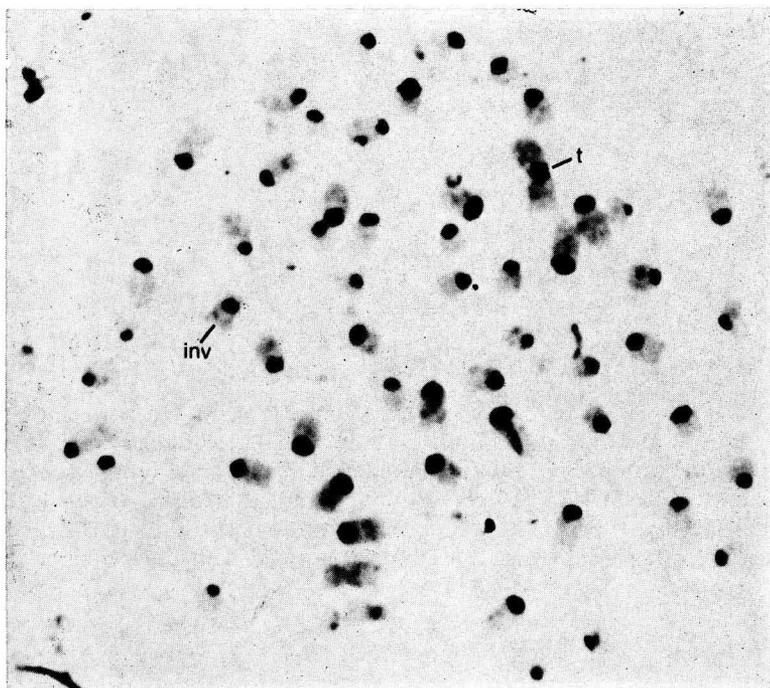


FIG. 3

C-bands of the abnormal line ; *t* = 12/15 translocation ;
inv = pericentric inversion of chromosome No. 6.

Topographie des bandes C de la lignée anormale ; t = translocation 12/15 ;
inv = inversion péricentrique du chromosome 6.

Blood typing showed complete hemolysis for the groups detected, and thus chimerism due to vascular anastomosis could be discarded. Unfortunately cytogenetic investigation of cells of different embryological origin could not be performed.

Several structural (GUSTAVSSON *et al.*, 1968 ; ELDRIDGE, 1974, 1980 ; POPESCU, 1977 ; ELLSWORTH *et al.*, 1979 ; MASUDA *et al.*, 1980 ; MORAES *et al.*, 1980) and numerical (RIECK *et al.*, 1970 ; NORBERG *et al.*, 1976) abnormalities have been found in bovine females with or without disturbed fertility. In some cases only a few ani-

mals with a particular abnormality have been studied so the correlation between abnormal complements and fertility was not completely established.

The presence of follicular cysts is of common occurrence in cows with high milk yield and its genetic origin has also been recognized (ROBERTS, 1971). Due to this facts and until more data related to the chromosome abnormality described here are available, it is difficult to associate this abnormal complement to the reproductive disorders found.

The origin of the abnormal cell line and the presence of two simultaneous chromosomal rearrangements are worth further considerations.

Chimerism due to the presence of a female co-twin dead *in utero* could be excluded because of lack of erythrocyte chimerism evidenced through blood typing (STORMONT, 1954). The difference between mosaicism and primary chimerism (McCLAREN, 1976) could not be established with the methods employed. Pure autosomal trisomic embryos rarely survive until birth and are malformed (HERZOG *et al.*, 1977). It has been stated that mosaics including this condition survive more easily and even attain a normal appearance (GUSTAVSSON, 1980). The trisomy detected in pair 15 in the abnormal line of the animal described here could be an example of the latter statement.

The presence of two simultaneous chromosome abnormalities is not a frequent event (DI BERARDINO *et al.*, 1979). Animals with two different centric fusions have been described among cattle (DARRÉ *et al.*, 1974; DI BERARDINO *et al.*, 1979; MASUDA *et al.*, 1980; HANADA *et al.*, 1981) and sheep (BRUÈRE *et al.*, 1976). Also two different and simultaneous reciprocal translocations have recently been reported in a bull (DE SCHEPPER *et al.*, 1982) and pigs (GUSTAVSSON *et al.*, 1982). Among cattle, all cases in which two Robertsonian translocations have been found in one animal were a combination of 1/29 and a translocation not previously described, reflecting, in part, the widespread occurrence of the former. When pedigrees of these animals were analyzed it was generally found that each translocation came from different parental lines. The double reciprocal translocation found by DE SCHEPPER *et al.* (1982) was not traced back among the relatives. Unfortunately, as parents and calves of the cow described here were not available for study, it is not possible to speculate about the parental origin, a *de novo* appearance or transmission of the abnormal chromosomes found.

The dicentric chromosomes may be of more recent origin than monocentrics, according to several authors (NIEBUHR, 1972; EVANS *et al.*, 1973; POPESCU, 1973; ELDRIDGE & BALAKRISHNAN, 1977). In view of this hypothesis the t(12q; 15q) found should be assumed as an ancient one. Anyway, there is no reason to assume that a *de novo* translocation could not be established between the long arm of an acrocentric chromosome and a broken point opposite to the long arm of a different acrocentric chromosome (ELDRIDGE & BALAKRISHNAN, 1977).

Pericentric inversions have been rarely found in cattle. SHORT *et al.* (1969) described a marker chromosome in the male line of a pair of heterosexual twins, which might represent a pericentric inversion. POPESCU (1972, 1976) has described a pericentric inversion of chromosome number 14 which lacked C-bands. The presence of C-bands in the inversion described here could reflect the fact that in the present

case the rearrangement could have taken place by a different mechanism than the one reported by POPESCU (1976).

In view of these findings it would be important to study the chromosomal complements of the relatives in order to analyze the transmission and segregation of the abnormal chromosomes.

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