

## A new Robertsonian translocation in *Blonde d'Aquitaine* cattle, rob(4;10)

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**Summary** – The cytogenetic study of a population of *Blonde d'Aquitaine* cattle revealed the presence of a Robertsonian translocation. The chromosomes involved in this abnormality were determined using G (GTG), R (RBG) and C (CBG) banding techniques. The chromosomes in question were identified as chromosomes 4 and 10. The existence of 2 paternal half-sisters carrying the abnormality suggests that it originates from the sire.

**cattle / chromosome / Robertsonian translocation**

**Résumé** – Une nouvelle translocation chez les bovins *Blonde d'Aquitaine*, rob(4;10). L'étude cytogénétique d'une population de bovins *Blonde d'Aquitaine* a permis de trouver une nouvelle translocation robertsonienne. Les chromosomes impliqués dans cette anomalie ont été déterminés à l'aide des techniques de marquage G (GTG), R (RBG) et C (CBG). Les chromosomes concernés sont le 4 et le 10. L'existence de deux vaches porteuses demi-sœurs de père indique une origine vraisemblablement paternelle de l'anomalie.

**bovin / chromosome / translocation robertsonienne**

### INTRODUCTION

Robertsonian translocations are the most commonly reported chromosome anomalies in cattle; the most widespread is the 1;29 translocation detected for the first time by Gustavsson and Rockborn (1964), and reported later with high frequency in numerous breeds worldwide (Popescu, 1977; Popescu and Pech, 1991). The 1;29 translocation is widespread in the *Blonde d'Aquitaine* breed since the frequency of the heterozygous and homozygous carriers ranges from 14 to 24% (Queinnec et

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*al*, 1974; Cribiu, 1985; Frebling *et al*, 1987). In contrast, as in other breeds, other Robertsonian translocations have been reported only as sporadic cases (Berland *et al*, 1988; Cribiu *et al*, 1989). The present report describes a new Robertsonian translocation observed in Blonde d'Aquitaine cattle.

## MATERIALS AND METHODS

### Animals

Karyotypes were prepared from one phenotypically normal Blonde d'Aquitaine cow carrying the new translocation (the proband), its mother and 3 half-sisters (1 maternal half-sister and 2 paternal half-sisters), belonging to private farms near Toulouse, southwest France. The pedigree is shown in figure 1.

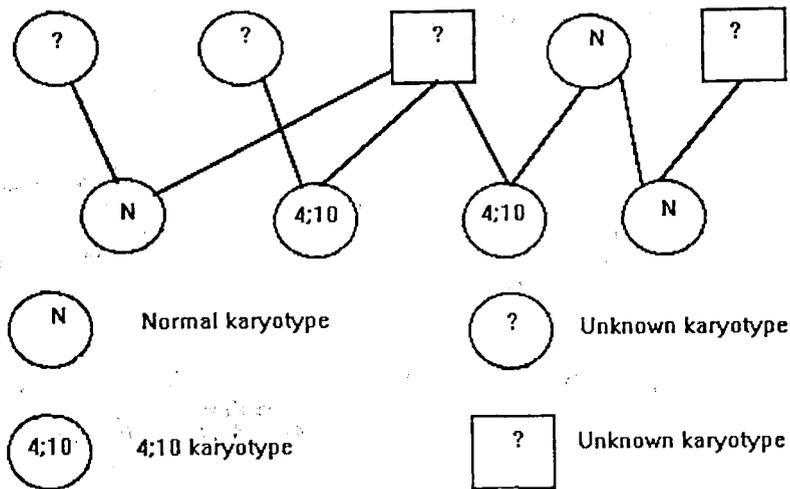


Fig 1. Pedigree showing the inheritance of the 4;10 Robertsonian translocation.

### Methods

The karyotype of each cow was determined using whole blood cultures (Grouchy *et al*, 1964) and primary skin cell cultures (Chaffaux *et al*, 1986). The peripheral blood was cultured at 37°C for 72 h in Ham's F12 medium supplemented with 20% fetal bovine serum, 2 mM glutamine, 100 µg/ml streptomycin and concanavalin A (final concentration: 0.1 µg/l). Colcemid (final concentration: 0.03 mg/l) was added to the culture 60 min before harvesting. Tissue biopsies were performed under local anesthesia on the rump. Primary fibroblast cultures were initiated from skin fragments, disrupted and digested in a trypsin solution (2.5 g/l) and grown

in a CO<sub>2</sub> incubator as monolayer cultures in Falcon dishes (75 cm<sup>2</sup>) containing a medium similar to that previously described for lymphocyte cultures.

G-banding was achieved using a modification of the technique of Seabright (1971). The C-bands were obtained by the barium hydroxide/saline/Giemsa (BSG) technique (Summer, 1972). To induce R-banding, 5-bromo-2-deoxyuridine was added to the medium at a final concentration of 10 or 20 µg/ml. The cultures were incubated at 37°C until the number of mitotic round cells reached a maximum, about 8 to 9 h after BrdU addition (Hayes *et al*, 1991). In order to obtain RBG-bands, the cells were treated according to the procedure described by Hayes *et al* (1991) and fluorochrome-photolysis-Giemsa (FPG) staining was performed as described by Viegas-Péquignot *et al* (1989).

The chromosomes were identified, paired and arranged according to the recommendations of the Reading Conference (1976) and the ISCNDA (1989).

## RESULTS

In classically stained metaphases, the karyotypes of the cow and 1 paternal half-sister included 59 chromosomes: the 2 X chromosomes, 56 acrocentric and one large submetacentric chromosome. The G- and R-bands showed that chromosome pairs 4 and 10 are involved in the translocation (figs 2, 3). The C-banding

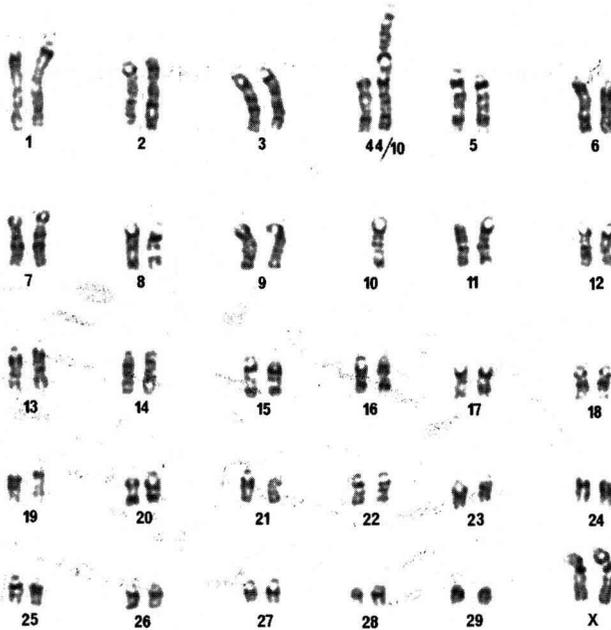


Fig 2. GTG-banded karyotype with the 4;10 translocation.



Fig 3. RBG-banded karyotype with the 4;10 Robertsonian translocation.



Fig 4. CBG-banded metaphase showing constitutive heterochromatin as 2 blocks in the 4;10 translocation (arrow).

technique revealed the presence of 2 constitutive heterochromatin blocks in the pericentromeric region of the 4;10 translocated chromosome (fig 4).

Among the other 3 animals examined, the mother, 1 paternal half-sister and 1 maternal half-sister were normal with a diploid chromosome number ( $2n$ ) of 60.

## DISCUSSION AND CONCLUSION

This chromosome abnormality is the fourth Robertsonian translocation reported in the Blonde d'Aquitaine breed. The first translocation was the 1;29 translocation which is known to have a wide distribution among AI bulls (Queinnec *et al*, 1974) and heifers (Frebling *et al*, 1987). Chromosomes implicated in the second and third translocations were identified as the 21 and 27, and the 9 and 23, respectively (Berland *et al*, 1988, Cribeu *et al*, 1989). These 2 rob(21;27) and rob(9;23) have been observed only in one Blonde d'Aquitaine bull and its progeny respectively.

Robertsonian translocations are the result of the fusion of 2 acrocentric chromosomes. Two types of Robertsonian translocations have been described in the Blond d'Aquitaine breed, depending on the presence of one block (1;29 translocation) and 2 blocks (21;27 and 9;23 translocation) of juxtacentromeric constitutive heterochromatin revealed by the CBG-banding technique (Berland *et al*, 1988; Cribeu *et al*, 1989). The presence of these 2 blocks would suggest the mechanism by which this translocation arose. The breakpoints involved the short arms, which are extremely limited in size, of both chromosomes 4 and 10 in the centromeric region; the fusion gave rise to a submetacentric chromosome and 2 minute fragments (short arms) which were lost during the subsequent cell divisions (Eldridge, 1974).

The origin of the translocation is uncertain, since the karyotype of the sire is unknown. It is probable that the 4;10 translocation originated from the sire since it was found in 1 paternal half-sister and not in the mother and the maternal half-sister.

As with a majority of Robertsonian translocations found in animal populations, the 4;10 translocation does not seem to be associated with phenotypic characteristics. In the absence of fertility records, a reduced fecundity in heterozygotes resulting from anaphase I nondisjunction and/or changes in the pattern of recombination in such individuals, cannot be excluded. For example, the 1;29 translocation produces in certain breeds a reduced fertility in the daughters of carrier bulls (Gustavsson 1969; Refsdal 1976).

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## REFERENCES

- Berland HM, Sharma A, Cribiu EP, Darré R, Boshier J, Popescu CP (1988) A new case of Robertsonian translocation in cattle. *J Hered* 79, 33-36
- Chaffaux S, Cribiu EP, Crespeau F (1986) Un cas rare d'hermaphrodisme vrai latéral chez une chienne 78, XY. *Rec Méd Vét* 162, 463-470
- Cribiu EP (1985) Fréquence de la translocation 1/29 dans les centres d'insémination artificielle français. *Elev insémin* 209, 17-22
- Cribiu EP, Matejka M, Darré R, Durand V, Berland HM, Bouvet A (1989) Identification of chromosomes involved in a Robertsonian translocation in cattle. *Genet Sel Evol* 21, 555-560
- Eldridge FEA (1974) A dicentric Robertsonian translocation in a Dexter cow. *J Hered* 65, 353-355
- Frebling J, Foulley JL, Berland HM, Popescu CP, Cribiu EP, Darré R (1987) Résultats de l'enquête sur la fréquence de la translocation 1/29 en race bovine Blonde d'Aquitaine. *Bull Tech. CRZV Theix INRA* 67, 49-58
- Grouchy de J, Roubin M, Passage E (1964) Microtechnique pour l'étude des chromosomes humains à partir d'une culture de lymphocytes sanguins. *Ann Génét* 7, 45
- Gustavsson I, Rockborn G (1964) Chromosome abnormality in three cases of lymphatic leukemia in cattle. *Nature (Lond)* 203, 990
- Gustavsson I (1969) Cytogenetics, distribution and phenotypic effects of a translocation in Swedish cattle. *Hereditas* 63, 68-169
- Hayes H, Petit E, Dutrillaux B (1991) Comparison of RBG-banded Karyotypes of cattle, sheep and goat. *Cytogenet Cell Genet* 57, 51-55
- ISCNDA (1989) *International System for Cytogenetic Nomenclature of Domestic Animals* (Di Berardino D, Hayes H, Fries H, Long S, eds) *Cytogenet Cell Genet* 53, 65-79 (1990)
- Popescu CP (1977) Les anomalies chromosomiques des bovins (*Bos taurus* L). État actuel des connaissances *Ann Génét Sél Anim* 9, 463-470
- Popescu CP, Pech A (1991) Une bibliographie sur la translocation 1/29 des bovins dans le monde (1964-1990). *Ann Zootech* 40, 271-305
- Queinnec C, Darré R, Berland HM, Raynaud JC (1974) Étude de la translocation 1/29 dans la population bovine du Sud-Ouest de la France : conséquences zootechniques. In: *1<sup>er</sup> Congrès Mondial de Génétique Appliquée à l'Élevage Animal*. Madrid, 7-11 octobre; Garsi, Madrid, 131-151
- Reading Conference (1976) Proceedings of the First International Conference for the Standardization of Banded Karyotypes of Domestic Animals, Reading, August 2-6, 1976 (Ford CE, Pollock DL, Gustavsson I, eds) *Hereditas* 92, 145-162 (1980)
- Refsdal AO (1976) Low fertility in daughters of bulls with 1/29 translocation. *Acta Vet Scand* 17, 190-195
- Seabright M (1971) A rapid banding technique for human chromosomes. *Lancet* 2, 971-972
- Summer AT (1972) A simple technique for demonstrating centromeric heterochromatin. *Exp Cell Res* 75, 304-306

Viégas-Péquignot E, Dutrillaux B., Magdelenat H, Coppey-Moisan M (1989) Mapping of single copy DNA sequences on human chromosomes by *in situ* hybridization with biotinylated probes: enhancement of detection sensitivity by intensified fluorescence digital-imaging microscopy. *Proc Natl Acad Sci, USA* 86, 582-586