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NEW DATA ON PERICENTRIC INVERSION IN CATTLE *(BOS TAURUS L)* *

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SUMMARY

We report a cytogenetic study on 27 females, all progeny of a *Norman* bull previously found to be a carrier of pericentric inversion. Eleven have a normal karyotype and 16 are pericentric inversion carriers. This abnormality thus seems to be transmitted as a simple, dominant character. The chromosomes are paired according to the R-band method after BUDR marking, and the abnormal chromosome is situated in the 14th pair according to relative length. Its centrometric index is 0.30. All the animals studied are phenotypically normal but the sire of these progeny has a low fertility rate.

We previously described pericentric inversion in a *Norman* bull (POPESCU, 1972). Because of the rarity and interest of this abnormality, we continued the study on a lot of that animal's progeny to understand how it is transmitted. The band method was used to identify the abnormal chromosome.

MATERIAL AND METHODS

We studied 27 females, all daughters of the bull carrying the abnormality. They were phenotypically normal but there is no data on their fertility. However, the bull had a low fertility rate because the non-return rate at 60-90 days, calculated on 398 first artificial inseminations, was 58.64 p. 100.

We first looked for the abnormal chromosome in the usual preparations obtained with the method of de GROUCHY *et al.* (1964) using whole blood. The second culture series was carried out with the blood of two animals identified as carriers of the abnormality; the best of these slides were studied by the band method.

R-bands were studied by the BUDR incorporation procedure proposed by DUTRILLAUX *et al.* (1973 a) for humans and adapted to cattle chromosomes (POPESCU, 1975 a). Eight hours before culture was stopped, the BUDR (Sygma) was put into contact with the cells in a final concentration of 200 µg/ml of medium. The slides were prepared in the usual way and stained 20 minutes in a solution of 0.5 p. 100 orange acridine, washed and mounted in a buffer solution at 6.7 pH. We studied the slides using a Leitz-Ortholux microscope equipped with a Pleom

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illuminator having an HBO 200 lamp and BG 12, KP 490 and K 510 filters. Photos were taken with a Leitz-Orthomat camera on Ilfort Pan F film.

We obtained the C-bands using SUMNER's method (1972) slightly modified (POPESCU, 1974).

RESULTS

Out of the 27 females studied, 11 had a normal karyotype (60, XX) and 16 presented a medium-sized submetacentric chromosome probably caused by pericentric inversion (fig. 1 a, b, c).

1

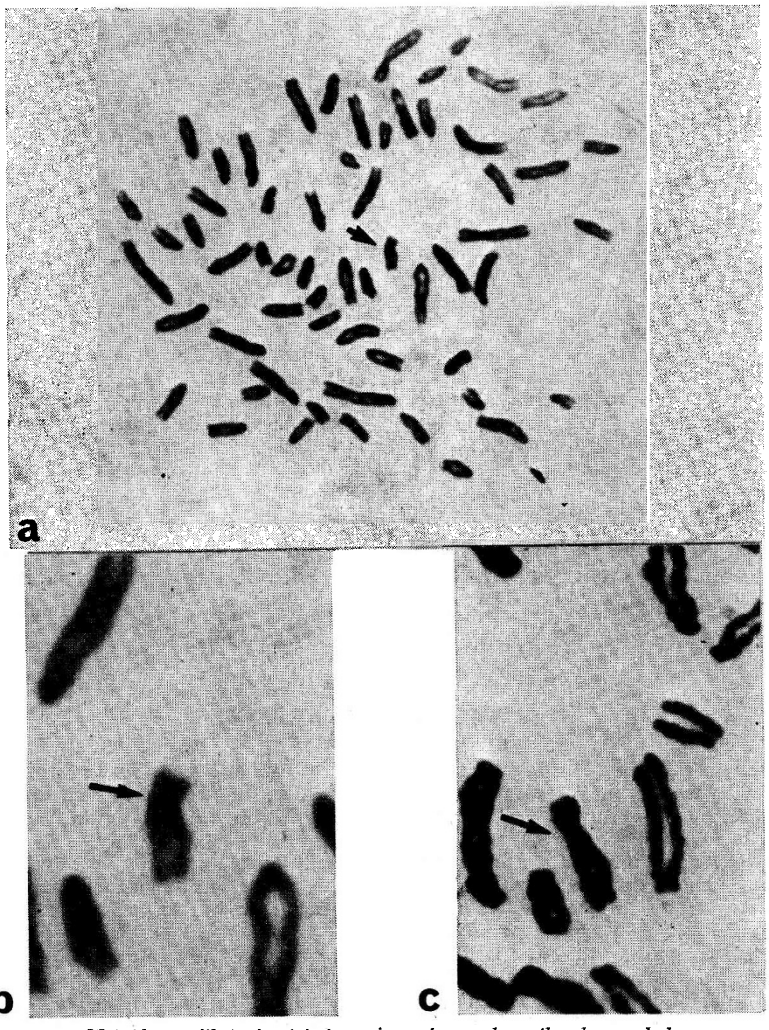

FIG. 1 a. — *Metaphase with pericentric inversion. Arrow shows the abnormal chromosome.*
b. c. — *Abnormal chromosome of two different cells.*

FIG. 1. a. — *Métaphase porteuse de l'inversion péricentrique. Le chromosome anormal est marqué par une flèche.*
b. c. — *Le chromosome anormal des deux cellules différentes.*

The band pattern is sufficiently clear to correctly pair the autosomes (fig. 2 a, b). The abnormal submetacentric chromosome had a mean centrometric index of 0.30, comparable to that found in the sire of these progeny (POPESCU, 1972). In the karyotype in which chromosomes are paired using R-bands and arranged in decreasing order according to relative length, the abnormal chromosome is found in the 14th pair (fig. 2 b).

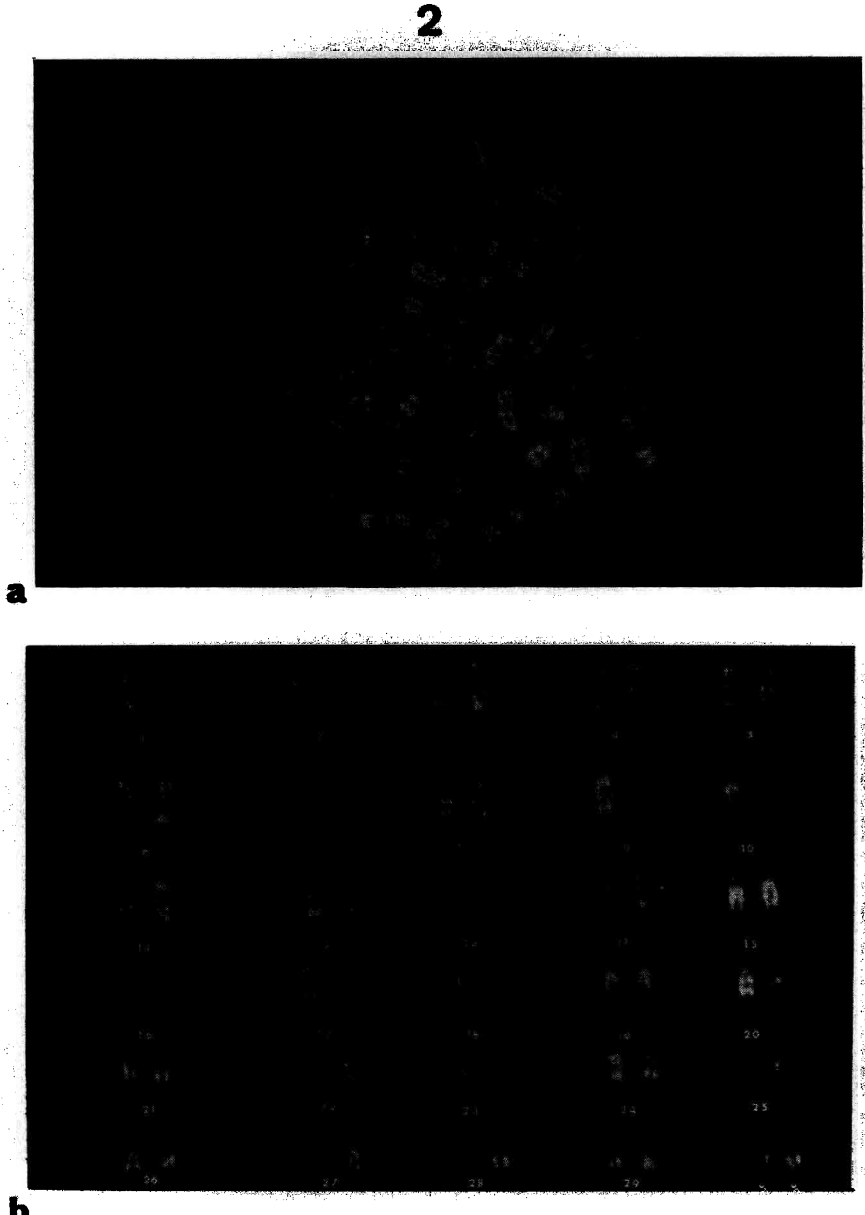

FIG. 2. — *R-bands.* a) *Metaphase.* b) *Karyotype of the same cell.*
FIG. 2. — *Bandes R.* a) *Métaphase* b) *Caryotype de la même cellule.*

In the metaphases treated by the C-band method, the constitutive heterochromatine appears normally distributed in the pericentromeric region of normal autosomes, but it is completely absent on the abnormal chromosome (fig. 3).

3

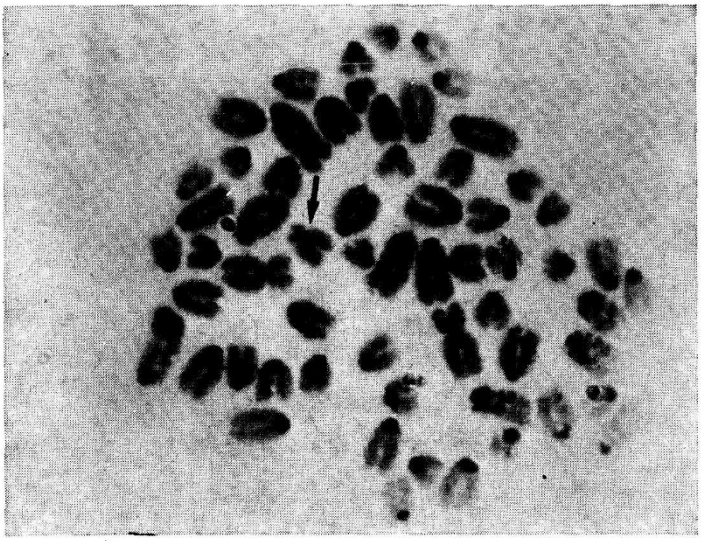

FIG. 3. — *C-bands. Arrow shows abnormal chromosome.*

FIG. 3. — *Bandes C. Le chromosome anormal est marqué par une flèche.*

DISCUSSION

In spite of a slight excess of heterozygotes (16 to 11 homozygotes) the observed ratio is not statistically different from a 1 : 1 ratio (χ^2 = .925,1 d. f.) and we may admit that the abnormality is transmitted as a simple, mendelian, autosomal, dominant character. Its pattern of transmission was previously found in man (De la CHAPELLE *et al.*, 1974).

Using BUDR a sa marker permitted the abnormal chromosome to be identified in the 14th pair. According to centromeric index, the breaking point preceding the inversion was situated in the lower third of the long arm.

The constitutive heterochromatine is usually found on all *Bos taurus* autosomes in the pericentromeric region (POPESCU, 1973; HANSEN, 1973; SCHNEDL and CZAKER, 1974), and its absence on the abnormal chromosome is difficult to explain. This chromosome may have been either poor in constitutive heterochromatine, or was lacking it, before inversion. In some autosomal pairs in cattle, there are wide differences in the amount of constitutive heterochromatine found in the two homologues (POPESCU, 1974, 1975, 1976).

A study of percentric inversion in man has shown the size of the abnormal chromosome and the position of the breaking points to be determining factors in the formation of unbalanced gametes by « aneusomie de recombinaison »

(DUTRILLAUX *et al.*, 1973 b). Unbalanced gametes carryng duplications-deficiencys (LEJEUNE and BERGER, 1965) cause many abnormalities and malformations (DUTRILLAUX *et al.*, 1973; VAN DER LINDEN, 1975, FRAISSE, 1975).

Besides these effects a pericentric inversion, may disturb by chromosomic interaction, the normal meiotic process of other chromosomes, thus causing malsegregation. Several cases of trisomy associated with pericentric inversion can thus be explained (CATTI, 1975).

The fertility disorders often manifested by pericentric inversion carriers may be induced by early embryonic mortality, but also by the direct effect of the abnormality on spermatogenesis (BOUÉ *et al.*, 1975) and the male sex carrying seems more severely affected by disorders than the female (De la CHAPELLE *et al.*, 1974; BOUÉ *et al.*, 1975).

The incidence of percentric inversion in man is estimated at 0.07×10^3 by DUTRILLAUX *et al.*, (1973) and at 1×10^3 by De la CHAPELLE *et al.* (1974). Only one case in cattle has been described previously (SHORT *et al.*, 1969). However, pericentric inversion may be more frequent in this species but is not observed if the inversed segment or the abnormal chromosome is small.

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RÉSUMÉ

NOUVELLES DONNÉES SUR UNE INVERSION PÉRICENTRIQUE CHEZ LES BOVINS (*Bos taurus* L.)

Une étude cytogénétique a été entreprise sur 27 femelles toutes descendantes d'un taureau *Normand*, trouvé précédemment porteur d'une inversion péricentrique. Onze avaient un caryotype normal et 16 étaient porteuses de l'inversion péricentrique, ce qui indique que cette anomalie est transmise comme un caractère simple dominant. Par la méthode des bandes R, après marquage au BUDR, les chromosomes ont été appariés et selon la longueur relative le chromosome anormal a été placé dans la 14e paire. Son index centromérique était de 0,30. Les animaux étudiés étaient tous phénotypiquement normaux, mais le père de ce groupe de demi-germains avait une fertilité réduite.

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