Genet. Res., Camb. (1997), **69**, *pp.* 237–238. *With 2 figures* Printed in the United Kingdom © 1997 Cambridge University Press

SHORT PAPER

A new chromosomal polymorphism by duplication of a heterochromatic region in cattle

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Summary

A new hereditary duplication located at the hetechromatin region of the 14th chromosome is described by CBG, CDD and RE banding in a family of Charolais cattle.

Chromosomal rearrangements, different from Robertsonian translocations, are hard to detect and generally more deleterious in the heterozygous state (White, 1978), affecting reproductive performance in the carriers (Searle, 1988). However, heterochromatic additions and deletions appear to be common in mammals (Qumsiyeh, 1994). In Bovidae, heterochromatic polymorphisms and differences in the repetitive DNA composition have been elucidated by CMA_3/DA/DAPI CDD (Mayr *et al*., 1985) and by restriction endonuclease (RE) banding (Hidas, 1994), respectively. In this work, a new hereditary anomaly of an autosomal heterochromatic region is reported in cattle.

Fixed metaphase chromosomes were obtained from peripheral blood of five males and six females belonging to a family of Charolais cattle. In all metaphase cells the expected chromosome number was observed (2n = 60). In four animals, C-banding (Sumner, 1972) showed an anomalous autosome bearing a large heterochromatin block (Fig. 1*a*). It was identified by CDD banding (Schweizer, 1980) as number 14, but an additional subcentromeric band CMA_3 positive, similar to the centromeric region of the primary constriction, was observed (Fig. 1*b*, *c*). This anomaly was transmitted from a heterozygous dam, showing abortions and returns on heat, to three offspring (Fig. 1*f*). Additional blocks of heterochromatin can appear as a result of Robertsonian translocations (Miyake *et al*., 1991); however, we have not found the chromosome from which the segment could have been translocated. For this reason, translocation is ruled out as the origin of the anomaly described.

RE banding (Mezzanotte *et al*., 1985) was obtained after digestion with *Hae* III, *Alu* I, *Hin*f I, *Rsa* I, and *Dde* I restriction endonucleases. *Alu* I, *Dde* I and *Rsa* I treatments did not induce any distinctive effects on polymorphic zone. However, after digestion with *Hae* III and *Hin*f I endonucleases we could clearly differentiate two blocks in the C-banded region. *Hae* III digestion showed that the additional block was located over separated chromatids (Fig. 1*d*), indicating that the additional heterochromatin was not associated with a new constriction. After *Hin*f I treatment we found an intermediate region darkly stained, separating two more digested chromosomal areas (Fig. 1*e*). The ability of the restriction endonucleases to attack some heterochromatic blocks, depending on the number and distribution of restriction targets, allows the detection of differences in the composition of the repetitive DNA (Hedeman *et al*., 1988). Thus, the polymorphic region CMA_3 positive contained evenly distributed *Hae* III sites, whereas two heterochromatic regions are left after treatment with *Hin*f I. These effects could indicate that the additional block originated from a duplication of the heterochromatin in the pericentromeric region of chromosome 14 and, therefore, in some way resembles a dicentric chromosome.

According to Vig & Schroeter (1993) duplications can result from unequal crossing-over. The early death of the embryos carrying other anomalies such as deletions or higher orders of duplications could explain both the absence of offspring harbouring complex rearrangements and the observed reproductive failures of the dam.

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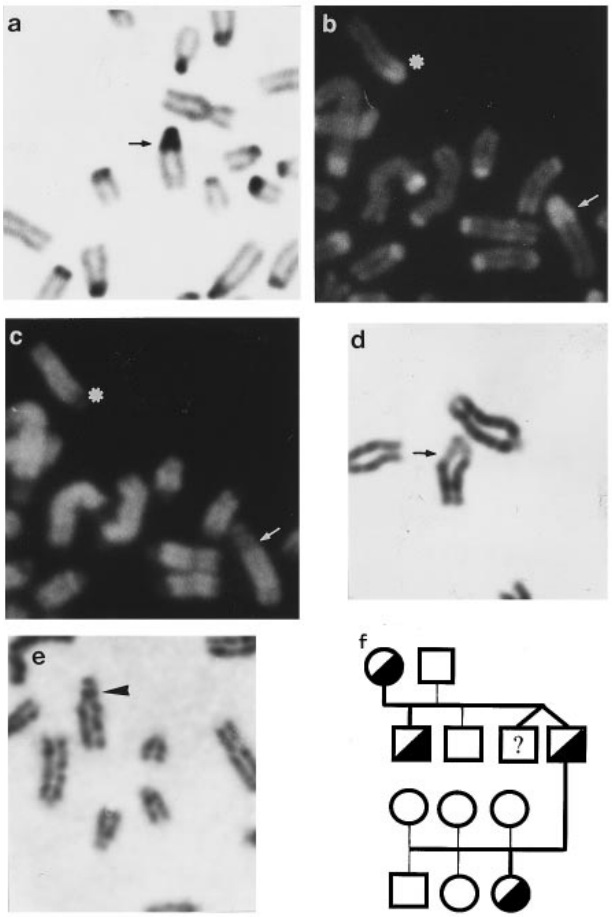

Fig. 1. (*a*)–(*e*) Partial metaphase spreads showing chromosome 14 carrier of the duplicated heterochromatin block after C-banding, CMA_3 stain, DAPI counterstain, and RE banding with *Hae* III and *Hinf* I, respectively (arrows show the duplicated region, arrowhead marks a resistant area to *Hinf* I and asterisks indicate the homologous region not affected). (*f*) Mendelian inheritance of this anomaly in a family of Charolais cattle (◒ or ◪, heterozygous carrier; ○ or □, normal; ⊡, not investigated).

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