

# Reciprocal X;1 translocation in a calf

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**Abstract** – A reciprocal translocation 60,XX,t(X;1)(42;13) was detected in a 2-month-old chimeric calf. A study of the parents of the calf strongly suggested the ‘de novo’ origin of the aberration. © Inra/Elsevier, Paris

**cattle / reciprocal translocation / X-autosomal / cytogenetics**

**Résumé** – Une translocation réciproque 60,XX,t(X;1)(42;13) sur un veau. Une translocation réciproque d’un veau chimérique de 2 mois a été détectée. L’examen de ses parents fait penser à l’origine de novo de l’aberration. © Inra/Elsevier, Paris

**bovin / translocation réciproque / X-autosomale / cytogénétique**

## 1. INTRODUCTION

Despite the high economic importance of cattle in farm animal breeding, very few data are available on reciprocal translocations in this species. Known interautosomal reciprocal exchanges are 60,XY,t(10;11)(41;14) in a bull showing a 60–90 days non-return rate of 30 % [11, 12], 60,XY,t(2q;20q),t(8q;27q) in an infertile bull [15] and 60,XY,t(8;15)(21;24) in a bull with a 60–90 days non-return rate of 25 % [13].

Further cases were 60,XY,t(1;8;9)(q43;q13;q26) with a reduction in 60–90 days non-return of 36.4 % [8], 60,XY,t(20;24)(q17;q25) with a reduction of 60–90 days non-return of 17 % [16] and 60,XY,t(8;13)(q11;q24) in a sterile bull [1].

In addition to these interautosomal reciprocal translocations, there are also a few known cases of X-autosome translocations [2, 3, 5, 6, 9, 14].

Now we have detected a further reciprocal translocation of the X-autosome type in cattle.

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## 2. MATERIALS AND METHODS

The 50 chimeric twins examined cytogenetically were all female chimeras of the Fleckvieh (Simmental) breed; they were 1–10 months old. Peripheral blood was withdrawn from the jugular vein and lymphocytes were isolated by the method described by Lin et al. [10]. The chromosomes were G-banded by the trypsin technique of Wang and Fedoroff [17]. G-banded preparations were karyotyped according to the Reading Conference recommendations [4]. The nomenclature of banding patterns was adopted from ISCNDA 1989 [7] for schematic representation.

## 3. RESULTS

A 2-month-old female chimera was found to possess 98 % female and 2 % male cells in a total of 200 screened cells. All of the 196 female cells were discovered to be heterozygous for the reciprocal translocation  $60,XX,t(X;1)(42;13)$  shown in *figure 1*. In contrast, the translocation was absent in the four XY cells. The translocation was balanced and the calf was healthy and appeared phenotypically normal at this age. We had no further data, i.e. later or section data regarding freemartinism.

While no blood sample from the male chimeric twin was available for cytogenetic investigation, samples of both parents of the female translocation carrier were investigated. Both of the parents presented normal karyotypes, i.e. they were free of any translocation in all the 150 blood cells investigated.

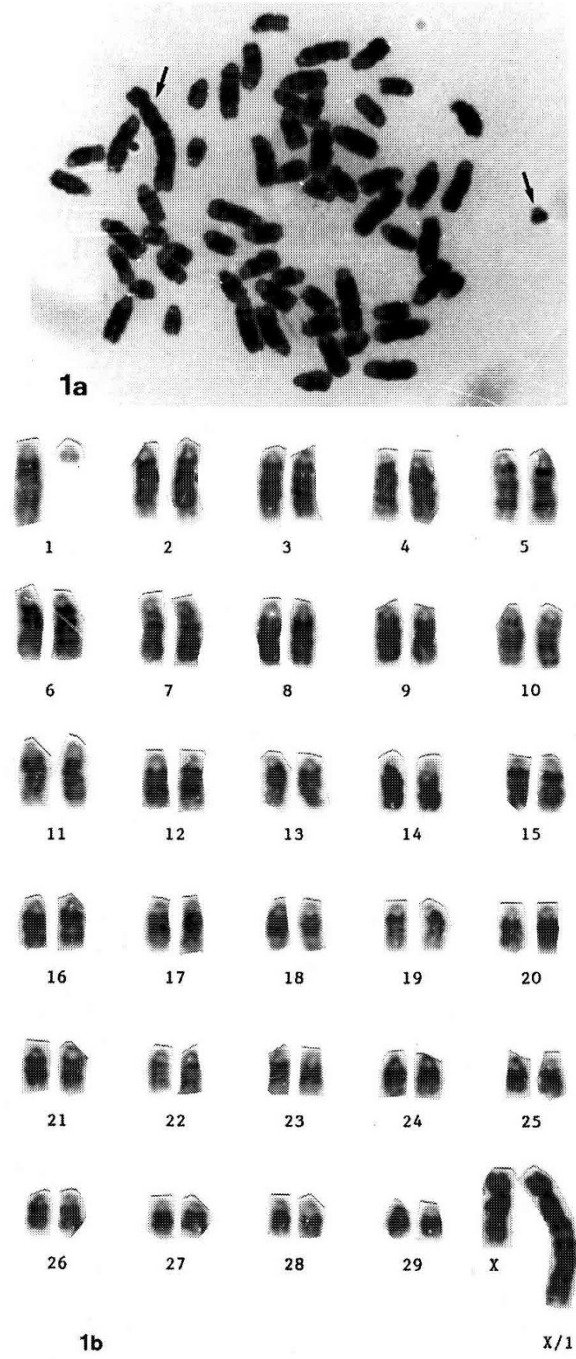
Therefore, a 'de novo' origin of the translocation was obvious.

## 4. DISCUSSION

The chimeric nature of our investigated female makes any conclusions regarding effects of the X;1-translocation on fertility impossible. However, it is justified to expect higher fertility depression from a reciprocal translocation as compared to the usually mild effects of Robertsonian translocations in cattle.

Because of the lack of the translocation in any cell of either parents, a 'de novo' origin of the rearrangement must be assumed. The mutational event probably occurred in the gametogenesis in the parental testis/ovary, or early in the embryogenesis of the calf. It is impossible to definitely differentiate between these possibilities, because no cytogenetic studies were performed in the parental gonads. Furthermore, the absence of any karyotypically normal XX cells makes a 'de novo' origin in late embryonic or fetal development rather improbable.

It may be expected that over the next few years new methods, such as chromosome painting, will speed up the detection of further reciprocal translocations in cattle, thus filling up the present gap in this field.



**Figure 1.** a) Metaphase of the 2-month-old chimeric calf. Arrows show the reciprocal translocation 60,XX,t(X;1)(42;13). b) Karyotype of 1a.

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