Génét. Sél. Evol., 1987, **19** (3), 381-386

Note

A new reciprocal translocation involving chromosomes 1/14 in a boar

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Summary

A case of a boar with reduced fertility and carrier of a reciprocal balanced translocation involving chromosomes 1 and 14 is presented. High resolution chromosomes are analyzed and the break-points of the rearrangement are compared to those of the other cases of translocations in pigs involving chromosomes 1 or 14 or both.

Key words : Boar, chromosome abnormality, breakpoint.

Résumé

Une nouvelle translocation réciproque impliquant les chromosomes 1 et 14 chez un verrat

Une nouvelle translocation réciproque impliquant les chromosomes 1 et 14 est décrite chez un verrat présentant une prolificité réduite. Les chromosomes sont analysés avec une haute résolution et les points de cassure sont comparés à ceux décrits chez le porc dans d'autres cas de translocations impliquant les chromosomes 1 et 14 (l'un des deux ou les deux simultanément).

Mots clés : Verrat, anomalie chromosomique, point de cassure.

I. Introduction

The structural chromosome abnormalities which most frequently affect the reproductive performance of the domestic pig seem to be reciprocal translocations.

As regards the chromosomal complement, the carrier of a reciprocal balanced translocation may produce at meiosis 3 classes of gametes : normal, balanced and unbalanced. Fertilisation of an unbalanced gamete produces an embryo bearing duplications and defects, which rarely survives. This leads to reduced litter size and hence a reduced fertility is the frequent consequence of heterozygosity due to a reciprocal translocation in the pig.

To date, 19 cases of translocations in pigs have been reported in the literature (POPESCU & BOSCHER, 1986).

The purpose of this work is to describe a translocation involving chromosomes 1 and 14 studied with high resolution banding.

II. Material and methods

The boar investigated was a one and a half year old *Large White* (n° 6438) with a normal body conformation. The boar was first employed in artificial insemination but the litters produced were small. The semen picture, however, appeared quite normal, as was the boar's libido. Later, when used in natural matings, this reproductive performance impairment was again observed. The boar's fertility was reduced by 35 p. 100 (table 1) compared to all the other matings recorded in this breeding farm up to December 31, 1982.

TABLE 1

Comparison between the number of piglets for each litter produced by boar n° 6438 and by other farm boars

Boars	Litter n°	Mean and s.d. of live piglets for each litter
Boar n° 6438	65	6.58 ± 1.37
Other boars (23)	1 845	10.02 ± 0.42

Given this reduced fertility, a cytogenetic analysis of the boar was carried out. The offspring were not available for investigation because they had not been marked, as they belonged to litters not reaching a sufficently high number to be registered in the Herd-Book. The boar was slaughtered immediately after the beginning of the cytogenetic study due to a severe limb injury.

The cytogenetic study was carried out on lymphocyte cultures set up in the conventional way. The chromosomes were investigated by GTG (SEABRIGHT, 1972) and RBA techniques (BrdU in a final concentration of 30 μg/ml was added 7 h before harvest) (DUTRILLAUX *et al.*, 1973). The karyotype was established according to the recommendations of the Reading International Conference 1976 (FORD *et al.*, 1980).

III. Results

The RBA and GTG banding showed the presence of a reciprocal translocation between the long arm of a chromosome 1 and a chromosome 14 in all 30 metaphases scored (fig. 1).

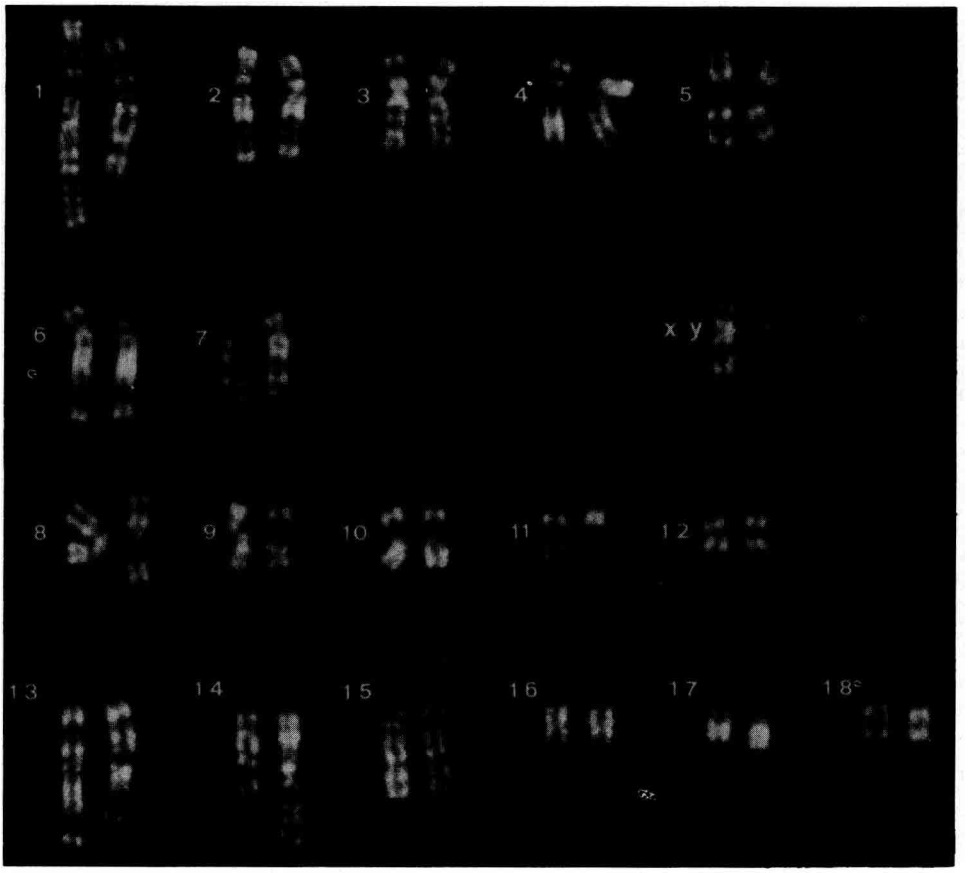

FIG. 1

R-banded karyotype of the boar carrying the 1/14 translocation.

The possibility of analysing high — quality chromosomes with G — banding pattern in more detail than that described by HANSEN (1977), led us to modify the nomenclature diagram of chromosomes 1 and 14, dividing the bands in sub-bands in agreement with the system adopted for human cytogenetics (ISCN, 1981).

TABLE 2

The different reciprocal translocations described in the pig involving chromosomes 1 and 14

No.	Rearrangement	Authors
1	trcp 1 p − ; 6 q +)	LOCNISKAR *et al.* (1976)
2	trcp (13 q − ; 14 q +)	HAGELTORN *et al.* (1976)
3	trcp (6 p + ; 14 q −)	MADAN *et al.* (1978)
4	trcp (4 q + ; 14 q −)	POPESCU & LEGAULT (1979)
5	trcp (1 p − ; 16 p +)	FORSTER *et al.* (1981)
6	trcp (1 p − ; 8 q +)	GUSTAVSSON *et al.* (1982)
7	trcp (1 q + ; 14 q −)	GOLISCH *et al.* (1982)
8	trcp (1 p + ; 14 q −)	GUSTAVSSON & SETTERGREN (*in* POPESCU *et al.*, 1984)
9	trcp (1 q − ; 17 q +)	GUSTAVSSON & SETTERGREN (*in* POPESCU *et al.*, 1984)
10	trcp (5 p − ; 14 p +)	POPESCU *et al.* (1984)
11	trcp (1 ; 7)	GUSTAVSSON (*in* POPESCU & BOSCHER, 1986)
12	trcp (1 q − ; 14 q +)	Present case

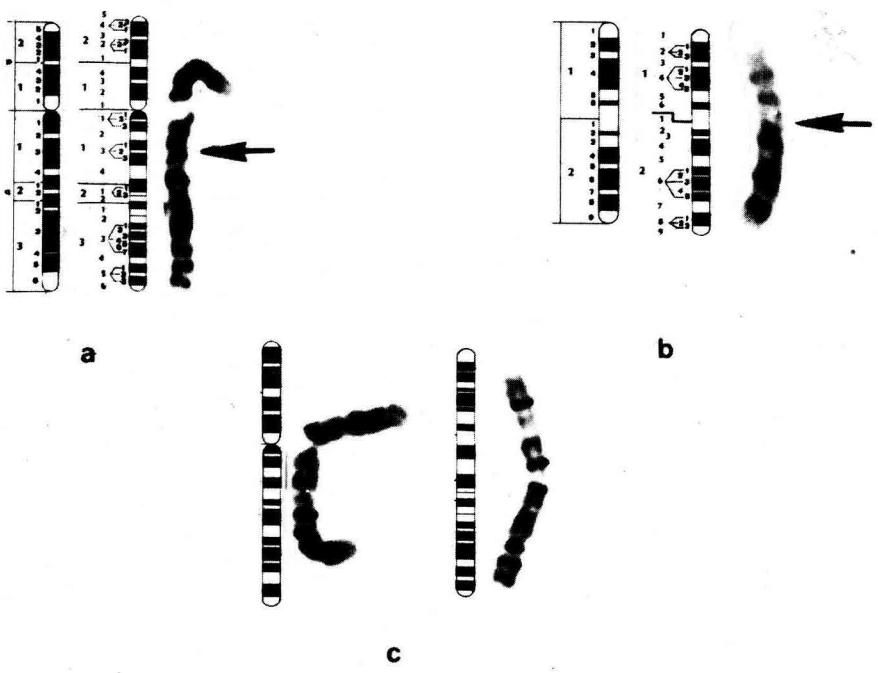

FIG. 2

Partial karyotype with high-resolution G-banded chromosomes demonstrating the breakpoint locations.

a) Normal chromosome 1 (right) with ideograms at high-resolution (middle) and according to HANSEN (left).

b) Normal chromosome 14 with the same schemes.

c) Derivative chromosomes 1 and 14 with high-resolution ideograms. The arrows indicate the breakpoints at 1 q 13.2 and 14 q 21.

According to this new schematic representation of chromosomes, the locations of the breakpoints of the rearrangement are designated as 1 q 13.2 and 14 q 21 (fig. 2). The karyotype of the boar is therefore 38 XY, t (1 ; 14) (q 13.2 ; q 21).

IV. Discussion

The case described, the first in Italy, shows a new reciprocal translocation between the long arm of a chromosome 1 and a chromosome 14.

Although increasingly reported over recent years, the number of balanced rearrangements discovered in the domestic pig throughout the world is still small. To date in fact the published cases, including the present one, amount to 20 in all (POPESCU & BOSCHER, 1986).

Such a small number of observations does not allow us to draw definite conclusions, but it is nevertheless surprising to note that the chromosomes involved in our case, i.e. 1 and 14, are repeatedly represented among the reported translocations. As table 2 shows, chromosomes 1 and 14 are, in fact, involved in 8 and 7 rearrangements respectively (3 times in a 1/14 exchange) ; this is equivalent to an involvement in 40 and 35 p. 100 of the total published instances. In addition it is interesting to examine the location of respective breakpoints.

Comparing our case with the previously documented observations concerning chromosome 14 (excluding case 8 of table 2), the breakpoints are seen to be very close or identical in 3 cases. As shown in fig. 3, which reports the breakage locations proposed by authors or inferred from the published iconography, band q 21 is involved both in our case and in that described by GOLISCH *et al.* (1982), while POPESCU & LEGAULT (1979) localise the exchange site in 14 q 16, i.e. in the band immediately proximal to q 21.

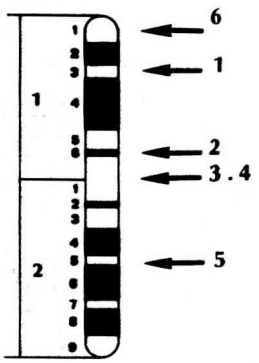

FIG. 3

HANSEN's diagrammatic representation of chromosome 14 with the indication of the breakpoints found in the documented cases (ours included) reported so far :
1. q 13 (MADAN et al., 1978)
2. q 16 (POPESCU & LEGAULT, 1979)
3. q 21 (GOLISCH et al., 1982)
4. q 21 (our case)
5. q 25 (HAGELTORN et al., 1976) (inferred by iconography)
6. q 11 (POPESCU et al., 1984) (inferred by iconography) (detail in the text).

For chromosome 1, the breakpoint in the only detailed report concerning the long arm (table 2, case 7) is completely dissimilar to that found in our case (q 32 vsq 13.1), but in the 3 documented cases involving arm 1 p (table 2, cases 1, 5 and 6) the break sites fall in a small segment near the centromere (bands p 11 or 12, p 11 and p 13 respectively) so that again it is possible that two or all of these are coincident.

Although scanty, these data are sufficient to suggest a non-random distribution of breakage points in spontaneous rearrangements in the pig, both between and within chromosomes. In order to verify this hypothesis, it is clearly necessary to collect a vast number of observations and to use high-resolution chromosome preparations to accurately localise breakpoints.

Received January 7, 1985.
Accepted February 3, 1987.

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