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A CYTOGENETIC SURVEY OF 1 101 AUSTRALIAN CATTLE OF 25 DIFFERENT BREEDS

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SUMMARY

The karyotypes of 1 101 cattle in Australia were examined in a population screening survey. XX : XY chimerism was found in the 5 animals of the *Hereford* and *Friesian* breeds. The incidence for *Friesian* cattle was 2 p. 100. A *Hereford* bull with 60,XY : 61,XXY mosaicism was identified. Autosomal trisomy was found in two bulls and one cow of the *Hereford* breed. Polyploidy was observed at a low level in samples from 577 animals. No polyploid cells were found in three muscular hypertrophic *Angus* animals. Chromosome associations were found in samples from 26 animals. Secondary constrictions were found in a proportion of the best quality cells in samples from 686 animals. No case of Robertsonian translocation, tandem fusion or of any of the other documented chromosomal aberrations was found.

INTRODUCTION

Population cytogenetic studies are directed at the estimation of the incidence of chromosomal aberrations. In cattle GUSTAVSSON (1969) found a high 14 p. 100 for a Robertsonian translocation in a select group of the *Swedish R* and *W* breed, HARVEY (1974) reviewed several reports to arrive at an estimated incidence of 1.53 p. 100 for the same defect in normal cattle of both *British* and *European* breeds and FECHHEIMER (1973) investigating only *British* breeds found no case of translocation carriers but did find a significant incidence of sex cell chimerism. No attempts have been made to arrive at estimates of the incidence of a further nine classes of chromosomal aberration mentioned in a review by HALNAN (1975).

This paper reports on the collected data from the study of 1 101 cattle in *Australia.*

MATERIALS AND METHODS

The survey comprised accumulated data during the period 1967 to 1975. Access to the animals studied was opportune and not by planned random selection. The greater part of the sample comprised 800 animals examined at the request of stud owners and the remainder were in herds belonging to public institutions. The details are presented in table 1.

TABLE I

Cattle Screened : By Breed and Sex

Bovins examinés : par race et sexe

Breed	Male	Female	Total
Afrikander	2	—	2
Angus	46	21	67
A.I.S. (1)	23	5	28
A.M.Z. (2)	1	—	1
Ayrshire	2	1	3
Banteng	7	1	8
Belmont Red	2	—	2
Brahman	15	5	20
Brahman × *Hereford*	9	9	18
Charolais	11	—	11
Friesian	126	48	174
Galloway	8	3	11
Hereford Horned	186	131	317
Hereford Polled	135	135	270
Hereford Crosses	14	1	15
Jersey	2	19	21
Lincoln Red S/H	2	—	2
Poll Shorthorn	1	—	1
Red Poll	5	3	8
Sahiwal	8	3	11
Santa Gertrudis	37	8	45
Simmental	1	—	1
Shorthorn	38	22	60
Sindhi	2	2	4
South Devon	1	—	1
	684	417	1 101

(1) *A.I.S.: Australian Illawarra Shorthorn.*
(2) *A.M.Z.: Australian Milking Zebu; Sahiwal* × *Jersey.*

The results were obtained by a peripheral blood lymphocyte method (HALNAN, 1976). A small number of cases were further studied by fibroblast cultures from other somatic tissues.

The chromosomes were stained by orthodox methods using orcein or Giemsa's stain and the work on banded chromosomes is not included.

The results were compiled from records of microscopic examination supported by photo-

micrography and karyotyping. In about 100 animals as few as 15 cells were studied ; for the remainder a minimum of 30 cells and as many as 300 cells were analysed.

A list of recognised chromosome aberrations, as described by HALNAN (1975), depicted the types of aberration looked for in the material.

RESULTS

1. — *General presentation*

The chromosomal aberrations observed and their incidence are summarised in table 2.

TABLE 2

Distribution of chromosomal aberrations according to breeds within the sample

Répartition des aberrations chromosomiques par race dans l'échantillon examiné

Breed/Sex		Chimmeris	Mosaic	Trisomy	Polyploidy	Associations	Secondary constrictions
Friesian	male	2			50		61
	female	2			25		27
Hereford	male	1	1	2	155	18	190
	female			1	133	7	146
Shorthorn	male				23	1	16
	female				8		13
Others					183		233
		5	1	3	577	26	686

2. — *XX : XY chimerism*

Five freemartins or cotwins were studied. A summary of the results is presented in table 3. None of the animals was available for dissection or histological study. The bull 3 was an AI donor and had a conception rate of 65 p. 100 which was 10 p. 100 below the average for other bulls in the same center. The heifer 4 had an enlarged clitoris, a small uterus and larger ovaries than normal, which did not manifest signs of cyclical activity although she did show a degree of oestrous behaviour. The bull 5 was used for natural service and found to have a greater return to service of his cows than normal, he was culled.

TABLE 3

Chimera observed, sex, age, cell ratios and status

No.	Subject	Sex	Age	Male Cells	Female Cells	Total Cells	Remarks
1	*Friesian*	F	18 mo	28	22	50	No signs oestrus
2	*Friesian*	M	18 mo	38	12	50	Sib to 1 castrated
3	*Friesian*	M	5 yr	44	6	50	Low conception rate
4	*Friesian*	F	15 mo	60	140	200	Showed oestrus
5	*Hereford*	M	18 mo	42	8	50	Culled low conception

3. — *Mosaic 60 XY : 61 XXY*

This was found in only one animal and the incidence of aberrant cells at 3 p. 100 was not very striking. Nevertheless the bull was found to have a depressed libido and poor sperm motility.

4. — *Autosomal Trisomy 61 XX and 61 XY*

Found in two bulls in which hip arthropathy was suspected and later confirmed and in one cow closely related to the arthropathic bulls. The proportion of hyperdiploid cells was low at 12 p. 100 (fig. 1). The chromosome involved could not be identified without banding.

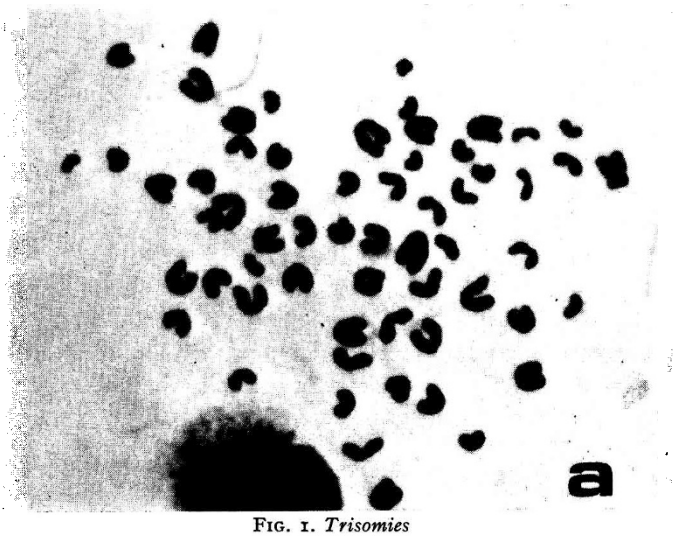

FIG. 1. *Trisomies*

a) Autosomal trisomy 61, XY from an arthropathic bull.

a) Trisomie autosomale 61, XY chez un taureau arthropatique

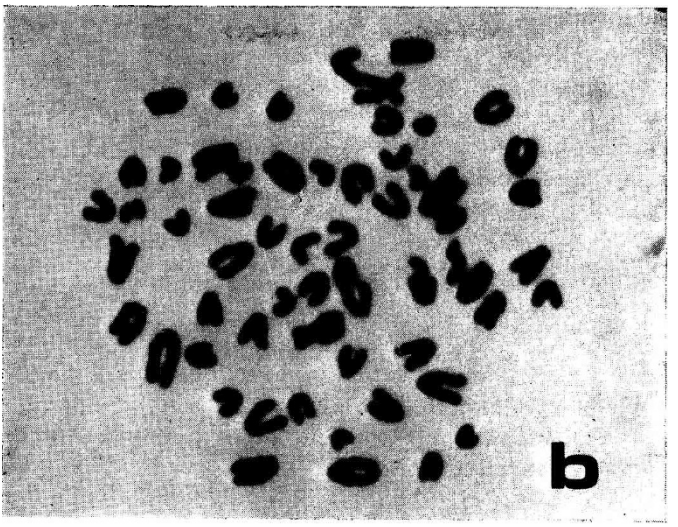

FIG. 1. (*suite*) — *Trisomies*

b) 61, XX from a cow related to the bull 1 *a* but not clinically diseased

b) Trisomie 61 XX chez une vache apparentée au taureau de 1 *a* mais indemne de la maladie

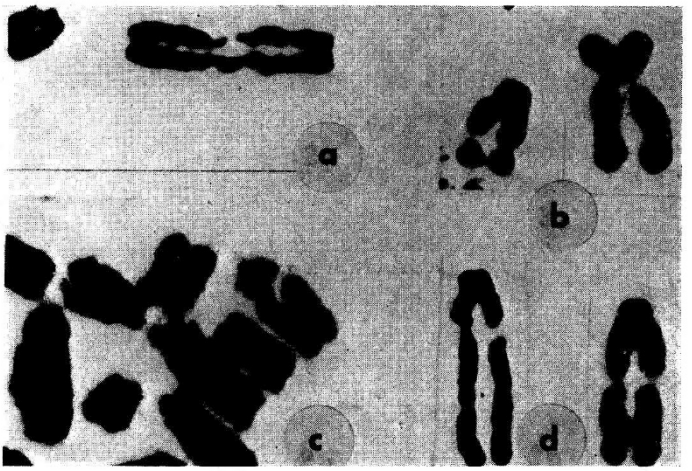

FIG. 2. — *Associations and constrictions*

a) Chromosome association between two autosomes
b) A small to medium size autosome with secondary constrictions giving the appearance of satellites with an X chromosome at the same magnification × 4000
c) Chromosome association between three autosomes × 4000
d) Secondary constrictions in large autosomes from two different animals demonstrating positional variation.

Associations et constrictions

a) Association entre deux autosomes
b) un autosome petit à moyen avec des constrictions secondaires ressemblent à des satellites, avec un chromosome X au même grossissement × 4000
c) Association entre trois autosomes × 4000
d) constrictions secondaires sur de grands autosomes de deux animaux différents avec différentes localisations.

5. — *Polyploidy*

Polyploidy was recorded as present in an animal on the finding of only one cell. In a select sample of 50 animals the level of incidence was determined, it varied from less than 0.1 p. 100 to 1.0 p. 100 of the total lymphocytes. The higher figures for the incidence of polyploidy were always found where the mitotic index was low. In three double muscled *Angus* animals no polyploid cells were found.

6. — *Associations*

Associations between two autosomes (fig. 2 *a*) or three autosomes (fig. 2 *c*) were encountered only in a low percentage of the mitotic cells of the order of 5 p. 100. The possibility of correlation with disease was investigated and no significant evidence was found. However, in 33 progeny of one bull 3 half sibs were found to carry the same triad configuration as the sire.

7. — *Secondary Constrictions*

The study of secondary constrictions is to be the subject of a later paper. Two examples only are presented here of seven types recognised so far (HALNAN, unpublished data) (fig. 3 *b* and *d*).

DISCUSSION

1. — *XX : XY Chimerism*

The incidence in *Friesian* cattle at 2.2 p. 100 agrees closely with the 2 p. 100 derived from the FECHHEIMER (1973) data. The subnormal fertility in the three bulls studied here is reason enough for reconsidering the practice of using cotwins in breeding programmes.

2. — *Mosaicism 60 XY : 61 XXY*

The very small percentage of aneuploid cells suggests that no major departure from normal genotype had occurred in this animal. The fertility problem is regarded as a fortuitous event and the conditions are noted without presumption. This case differs from the mosaic described by RIECK *et al.* (1969).

3. — *Autosomal trisomy 61 XX or 61 XY*

Reported trisomies amount to six cases (HERZOG and HÖHN, 1968 and 1971 ; MORI *et al.*, 1969 and DUNN and JOHNSON, 1972) if the three cases recorded here are included this brings the total to 9. The total number of cattle examined cyto-

genetically may be estimated from the literature mentioned by HALNAN (1975) together with the results reported here and the figure arrived at after allowing for 400 unreported normals was about 5.200. Thus the incidence of autosomal trisomy is .17 p. 100 which compares closely with human studies at .15 p. 100 of live births (HAMERTON, 1971). In both human and animal studies skeletal deformities are recognised as one of the phenotypic concomitants of autosomal trisomies.

4. — *Polyploidy*

The incidence of polyploidy in the population was high, in keeping with the earlier suggestion that it is an artifact of culture (HALNAN, 1976). The incidence of polyploid cells within individual normal animals agreed with the 2-6 p. 100 found by DARRÉ *et al.* (1970). Several correlations have been suggested for abnormally high levels of polyploid cells such as muscular hypertrophy (POPESCU, 1968 and DARRÉ *et al.*, 1970). in-breeding (ZARTMAN and FECHHEIMER, 1967) and neurological deformities (HERZOG and HÖHN, 1971). The failure to find polyploid cells in 3 muscular hypertrophic animals may indicate breed differences rather than contrary evidence of correlation.

5. — *Associations*

FORD and MADAN (1972) reported studies of chromosome associations describing them as branched chromosomes in preference to the term « endoreduplication selective » (LEJEUNE *et al.*, 1966). The chromosome associations described here bear a close resemblance to those described for man. Moreover, as in man, they were found here to be heritable.

6. — *Secondary Constrictions*

The work on secondary constrictions, achromatic regions or gaps in human studies has been summarised by HAMERTON (1971). Secondary constrictions seem to be involved in the formation of satellited chromosomes of the D and G groups where they are accepted as nucleolar organisers. In cattle the secondary constrictions in small autosomes (fig. 2 *b*), bear a resemblance to the human satellited condition. Therefore, it will be interesting to see if these cattle chromosomes are also the site of nucleolar organisers. The other types of secondary constriction in cattle, figure 2 *d*, resemble the type found in the Nos. 2, 3, 9 and 13-15 chromosomes of man. No mention was made by HAMERTON (1971) of the heritability of these secondary constrictions in man, however, in cattle they have been seen in the same position on the same chromosomes in the progeny of the propositus and in animals from related family lines (HALNAN, unpublished data). The possible occurrence in relation to phenotypic defects (HALNAN, 1972) is being further studied. That minor aberrations of this type should not be disregarded because they have been seen only in cells of the highest quality is encouraged by the remarks of BOBROW (1974) in relation to small differences observable in human banded preparations.

7. — *Robertsonian Translocation and Other Aberrations*

Robertsonian translocations and several other chromosomal aberrations noted previously (HALNAN, 1975) were not encountered. This study, like that of FECHHEIMER (1973), was mainly concerned with cattle breeds collectively described as *British*. The results suggest a lower incidence of chromosome aberrations than among the continental *European* breeds, with the exception of chimerism and autosomal trisomy.

CONCLUSION

The results have shown a low incidence of major chromosomal aberrations in the cattle studied, as the mosaics and trisomies were found only in a minority of the cells. Chimerism was found at levels similar to those expected from previous publications. Polyploidy is a condition which requires closer study as present evidence does not correlate polyploidy with phenotypic variation in mammals. The same conclusion applies to associations and secondary constrictions. However, if it is remembered that polyploidy is rarely observed *in vivo*, whereas, associations and secondary constrictions are, then the latter may have a definite role to play in the identification of phenotype variation.

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RÉSUMÉ

EXAMEN CYTOGÉNÉTIQUE DE 1 101 BOVINS AUSTRALIENS DE 25 RACES DIFFÉRENTES

Au cours d'une enquête cytogénétique sur la population bovine australienne 1 101 caryotypes ont été réalisés. Le chimérisme XX : XY a été trouvé chez 5 animaux *Hereford* et *Frisons*. La fréquence en race *Frisonne* était de 2 p. 100. Un taureau *Hereford* présentait une mosaïque 60 XY : 61 XXY. La trisomie autosomique était présente chez deux taureaux et une vache de race *Hereford*. Des échantillons en provenance de 577 animaux montraient un faible taux de polyploïdie. Aucune cellule polyploïde n'a cependant été trouvée chez trois *Angus* culards. Des associations de chromosomes existaient chez 26 animaux. Sur des échantillons provenant de 686 animaux on a trouvé des constrictions secondaires sur des cellules présentant les meilleures images. Aucun cas de translocation robertsonienne, de fusion en tandem ou d'autres aberrations chromosomiques déjà décrites n'a été repéré.

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