Genet. Res., Camb. (1964), **5**, pp. 328–329
With 1 *plate*
Printed in Great Britain

Short note

Cytology of the XXY mouse

By B. M. Slizynski*

Institute of Animal Genetics, Edinburgh, 9

(*Received* 16 *January* 1964)

This note gives some cytological observations on a male mouse, kindly provided by Mr J. H. Isaacson, which was presumed on genetical grounds to be of chromosomal constitution XXY. It was produced by a mating between a male of the inbred strain RIII/Fa and a non-bred female that was heterozygous for the sex-linked markers *tabby* (Ta) and *brindled* (Me^{br}) in repulsion. The phenotype of the mouse was that of a tabby heterozygote. It was therefore presumed to have the paternal X chromosome as well as one of the maternal X chromosomes. The observations described here refer to the somatic chromosome number, pachytene chromosomes, and so-called sex vesicle.

Recent investigations have shown that the mechanism of sex determination in mammals is not of the *Drosophila* type. In *Drosophila* the XXY fly is a fertile female, whilst in man the same chromosomal constitution results in a sterile male. Russell & Chu (1961) have shown that XXY in the mouse is, as in man, a viable male with normal external genitalia and small testes. The male described by these authors was sterile. No data on cytology of spermatogenesis were given.

Another case of proved XXY constitution in the mouse was described by Cattanach (1961). In his case, however, one of the X chromosomes contained a translocated piece of an autosome. Histological studies of the extremely small testes of two such males carried out by Cattanach showed the presence of seminiferous tubules but no spermatogenic cells. Somatic chromosome counts indicated that both these males were of the XXY constitution having forty-one chromosomes.

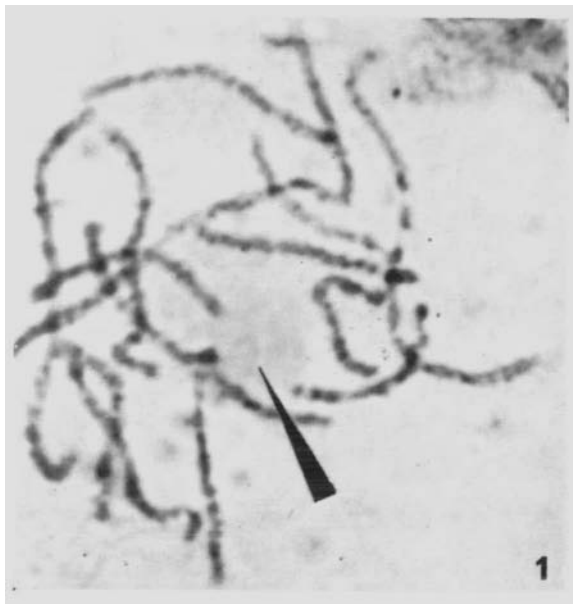
The phenotype of the XXY male described here was similar to that of Russell & Chu (1961); the external genitalia were normal but the testes were very small, about one-quarter of the normal size and less than one-tenth in weight.

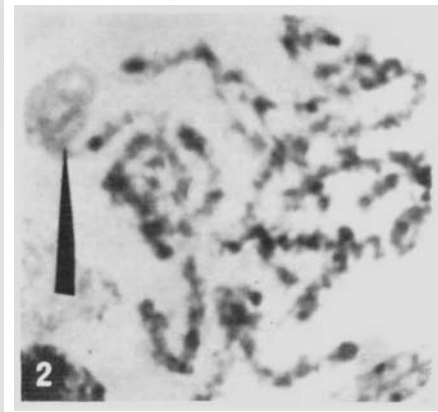
The chromosome counts in temporary preparations of somatic metaphase plates made by the lactic orcein technique, gave consistently forty-one chromosomes (six analysable nuclei were obtained, all of which had forty-one chromosomes). This, in conjunction with the genetical evidence, proves that this mouse had an XXY chromosome constitution.

In correlation with the small size of the testes, the seminiferous tubules were much thinner and their total length markedly smaller than in the normal mouse. In contrast to Cattanach's material, the testis was not completely devoid of spermatogenic elements. In the tubules there were many spermatogonial cells and a very large number of Sertoli cells, besides many spermatocytes.

Quite unexpectedly it has been found that some auxocytes entered meiotic prophase, progressing up to the stage of pachytene. No cells were found in later stages of meiosis. Altogether in the material of both testes no more than eighty cells were seen in leptotene, zygotene or in pachytene stages. The chromosomes of these cells were very difficult to squash, and therefore observations of the details of their morphology were not possible.

* Member of the external staff of the Medical Research Council.





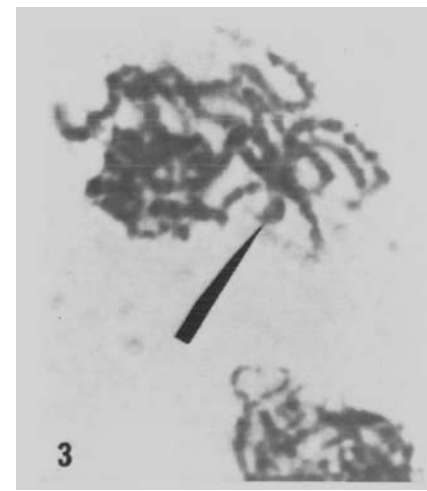


Fig. 1. Fully paired pachytene chromosomes of a normal male mouse. Note fuzzy outlines and changeable diameter of the chromosomes, large 'sex vesicle' with two dark lines inside.

Fig. 2. Fully paired pachytene chromosomes in embryonic oöcytes of a normal mouse. Note smooth outlines and evenness of the diameter of the chromosomes. There is a nucleolus attached to one pair of the chromosomes.

Fig. 3. Fully paired pachytene chromosomes of the XXY male mouse. Note smooth outlines and evenness of the diameter. The sex vesicle is smaller and more condensed with more darkly staining material inside.

https://doi.org/10.1017/S0016672300001269 Published online by Cambridge University Press

Autosomes of the pachytene stage in normal male mice are fuzzy and of very variable diameter (Fig. 1), in contrast to those of female mice, which have even contours and nearly uniform diameter (Fig. 2). The difference may be due to the occurrence of a uniform coiling and deposition of stainable material on the chromosomes in the oöcytes in contrast to a well-developed regional differentiation of the chromosomes for both these processes in the spermatocytes. The pachytene stage takes less time in oöcytes (not more than 4 days) than in the spermatocytes (at least 8 days).

The pachytene autosomes of the XXY male described here were in their external appearance more like those from the oöcytes than from normal spermatocytes. Their surface was not fuzzy and their diameter was uniform (Fig. 3); this was probably caused by the presence of two X chromosomes.

The sex chromosomes in meiotic prophase behave differently depending upon whether they are in oöcytes or in spermatocytes. In oöcytes they are similar in appearance to the autosomes. The only indication of any difference was given by Guenin (1948), who found that in the oöcytes of young female mice which had hormonally induced superovulation, the sex chromosomes were slightly retarded in the first anaphase. Meiotic prophase chromosomes of the embryonic mouse oöcytes when stained with methyl-green-pyronin revealed the presence of a nucleolus which was consistently attached to one pair of the chromosomes. It is probable that this pair represents the sex chromosomes (Fig. 2).

In the spermatocytes of the normal male the sex chromosomes appear as a globular structure which is not quite understood. According to Ohno, Kaplan & Kinosita (1957), the globular structure or the 'sex vesicle' in the mouse is actually a nucleolus, and the precociously condensed X and Y chromosomes are not inside it but are connected through the 'vesicle'. In the normal mouse the nucleolus is very large and shows clearly the X and Y chromosomes or their parts as two Feulgen positive dark lines (Fig. 1). The sex bivalent in the XXY animal was similar in appearance to that of the normal male, though much smaller, its Feulgen positive dark parts occupied much more of its volume and in general appearance it was more compact (Fig. 3). Owing to the difficulties of squashing the cells, nothing more can be said about its internal structure.

My thanks are due to Mr J. H. Isaacson for kindness in supplying the animal described here.

SUMMARY

The XXY male mouse, of otherwise normal chromosome complement, is characterized by small testes, few spermatogonia, and spermatocytes in which the first stage of meiosis up to the pachytene stage can occur, although with a very low frequency. Pachytene autosomes are morphologically similar to those of the oöcytes. The sex vesicle is smaller and more condensed than in the normal male.

REFERENCES

Cattanach, B. M. (1961). XXY mice. *Genet. Res.* **2**, 156–158.

Guenin, H. A. (1948). Les hétérochromosomes dans l'ovogenèse des Mammifères. *J. Genet.* **49**, 23–77.

Ohno, S., Kaplan, W. D. & Kinosita, R. (1957). Heterochromatic regions and nucleolus organizers in chromosomes of the mouse. *Exp. Cell Res.* **13**, 358–364.

Russell, L. B. & Chu, E. H. Y. (1961). An XXY male in the mouse. *Proc. nat. Acad. Sci., Wash.*, **47**, 571–575.