SPONTANEOUS INVERTED EXCHANGE BETWEEN *X* AND *Y* IN *DROSOPHILA MELANOGASTER*

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(With Six Text-figures)

The following note describes a peculiar rearrangement which occurred spontaneously between the *X*- and *Y*-chromosomes in *D. melanogaster*. The *X*-chromosome involved was of the type known as scute^{S1} (sc^{S1}) an extreme type of scute which is characterized by a long inversion extending from a point immediately to the right of scute to one in the chromocentral region which lies between bobbed and the centromere.

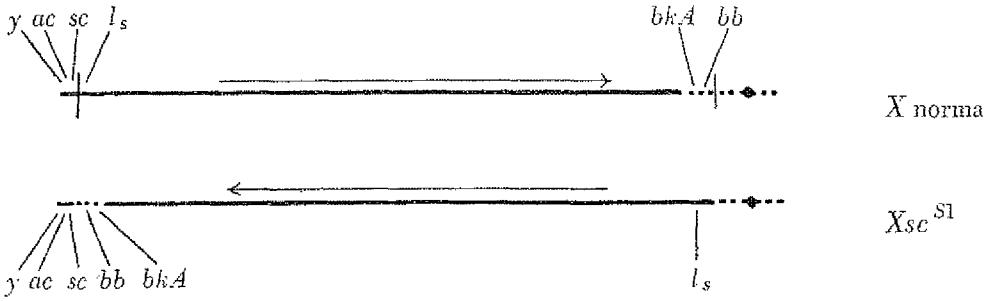


Fig. 1. Normal and sc^{S1} chromosomes, showing points of breakage: left break, between *sc* and l_s, the next gene to the right of *sc*; right break, between *bb* (with block *A*) and the centromere.

Thus in the sc^{S1} chromosome the scute locus which occupies its normal position (at the left or distal end of the chromosome) adjoins that part of the chromocentral region which contains bobbed and "Block A" (Muller & Gershenson, 1935). Fig. 1 shows the normal and the inverted order of the loci in question.

In forming the rearrangement with the *Y* the sc^{S1} chromosome became broken next to or within the chromocentral material to the right of scute, and the *Y*-chromosome was broken in its short arm in such a way that the "inert" chromatin in that arm and some or all genes necessary for fertility (Neuhaus, 1938) were separated from the centromere. The detached left end of the *X*, containing scute, became attached to the broken end of the *Y* near the centromere, being substituted for the short arm that had been removed. The piece of the *X* thus attached is too small to form a visible arm, hence the reconstituted

Y-chromosome (called the scute Y^L) appears rod-shaped in metaphase plates (Fig. 2). Whether the other pieces of the *X* and *Y* joined together is not known as they were not obtained.

The recombination by which the scY^L was formed may have taken place either in a male or in a female. The males of the stock in which the rearrangement occurred had contained the sc^{S1} chromosome and an ordinary *Y*; the females that were selected for breeding were triploids with attached-*X*-chromosomes homozygous for yellow, a free sc^{S1} chromosome, and very often, in addition, an ordinary *Y*-chromosome. The discovery of the new type of *Y*-chromosome was brought about by what appeared to be a sudden widespread contamination of the stock by wild-type flies. The breeding tests carried out on these supposedly foreign flies constitutes part of the genetical analysis of the scY^L chromosome, and the results are described below.

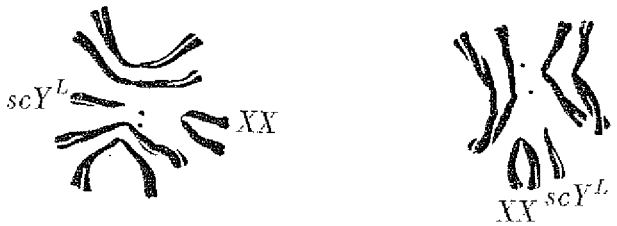


Fig. 2. Metaphase figure from female having attached-*X*-chromosomes and a scY^L.

Genetical analysis

Normally the only wild-type flies in the stock bottle were the triploids; detachment of the *X*'s could, however, produce a yellow male or a wild-type female (of constitution y/sc^{S1}), and either of these, if not detected and removed, would give rise to further wild-type females and also to yellow males. The wild-type diploid females which were found in large numbers were supposed to have arisen in this way. When the process of purifying the stock was begun, a few yellow males were found, but also unexpectedly a number of wild males, whose presence could normally be explained only by contamination. Many of these wild males and females, however, showed some slight but consistent differences from the normal wild, and for this reason it was decided to test their actual constitution.

The females were mated to sc^{S1} males: if they were the result of detachment of the *X*'s they should be sc^{S1}/y; but if they were the result of contamination they might be either pure wild or $sc^{S1}/+$., as far as

the *X*-chromosome was concerned. A combination of both conditions might have given rise to females that were $y/+$. In all cases the characters exhibited by their sons would reveal the constitution of the mothers.

In one or two cases the females tested proved to be sc^{S1}/y and carried a *Y*-chromosome as well. Many gave only sc^{S1} sons, but *both yellow and wild daughters*. These wild daughters, when mated to their scute brothers, gave the same results as their mothers. Another group of females, also wild in appearance and mated to sc^{S1} males, gave daughters that were scute or wild, and sons that were yellow scute *or wild*. When the wild daughters were mated to their scute brothers the same result was repeated. From these results it was clear that the apparently wild-type females (at least many of them) still carried attached-*X*'s marked by yellow, but that the yellow was being "covered" in some way; the wild-type allele of yellow was probably translocated to either an autosome or to the *Y*-chromosome, it was not clear at this stage which of these possibilities represented the actual position. This question was settled when a wild male taken at random from the stock bottle was tested.

This male was mated to an unrelated female of a stock in which the females carried attached-*X*-chromosomes marked by yellow and apricot (yw^a), and the long arm of the *Y* (Y^L). It has been shown (Stern, 1929) that both arms of the *Y* are necessary for fertility in the male; in this stock the short arm is attached to the *X*-chromosome which is transmitted by the males from father to son, and the long arm is supplied by the mother. The males are therefore fertile. But if the females are outcrossed to males which carry ordinary *X*- and *Y*-chromosomes they produce sons which are sterile since they receive only the Y^L. When a yellow apricot female of this stock was mated to the wild male which was being tested, the progeny obtained consisted of apricot (non-yellow) daughters, and yellow sons. From this result it was apparent that the wild male in question carried yellow on his *X*-chromosome which his sons received, but that the normal allele of yellow was attached to his *Y*-chromosome which his daughters received. If it had been carried on an autosome, it would have been equally distributed among his sons and daughters, half of each receiving it; (some of the sons would have been wild-type and some of the daughters yellow).

Three types of matings were made with these apricot females which carried the yellow-suppressing *Y*-chromosome, and the yellow males which had received the long arm of the *Y* only from their yellow apricot mother:

(1) backcross of apricot female to wild father;

(2) F_1 apricot female by yellow brother (expected to be sterile);

(3) outcross of apricot female to unrelated wild male from stock (Oregon K).

The results were as follows:

from (1): apricot females and wild males;
from (2): yellow apricot females and wild males;
from (3): yellow apricot females and wild males.

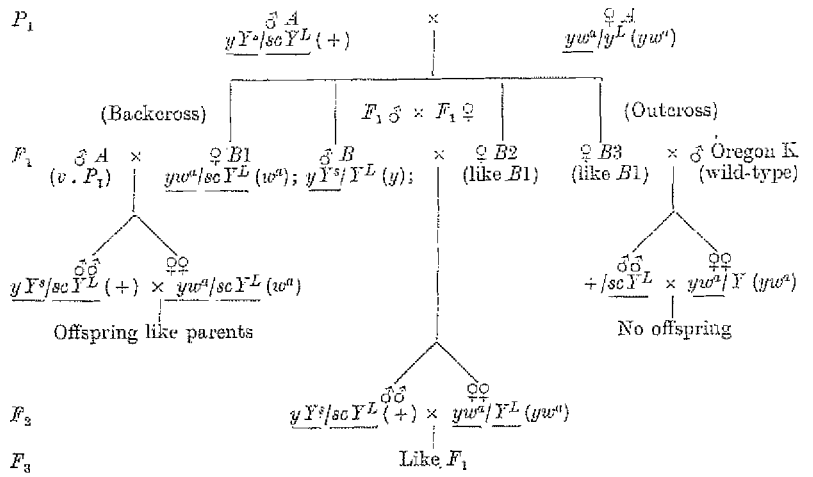
The following conclusions were drawn: (*a*) both male and female offspring from mating (1) carried the yellow-suppressing *Y*-chromosome (when inbred these formed a true-breeding stock); (*b*) since the F_1 yellow males used in mating (2) were fertile though they had received only the long arm of the *Y* from their mother, they must have received the short arm at least from their father; and since they were all phaenotypically yellow, the normal allele of yellow must have been on the long arm which their father had contained. The long arm of the *Y* was therefore segregating independently of the short arm and of the *X*, but with the wild-type allele of yellow attached to it, while the short arm of the *Y* was evidently attached to the *X*.

Flies of all three lines were inbred and the above conclusions were confirmed by the results. The F_2 from mating (1) were like the parents; that from mating (2) were like the grandparents (apricot females and yellow males); the male offspring from mating (3) were entirely sterile. They had received only the long arm of the *Y* from their mother and an ordinary *X* from their Oregon K father: hence they lacked the Y^S. Both the males and the females were remated to unrelated flies and the females were proved to be fertile. This series of matings is shown fully in Fig. 3, as it illustrates the distribution of the yellow suppressing *Y* (scY^L).

The wild-type males in the triploid stock were thus proved to be in reality yellow males which had resulted from separation of the attached-*X*'s. As has been shown by Kaufman (1933) separation of attached-*X*'s is usually accompanied by crossing-over with the *Y*-chromosome in such a way that the short arm of the *Y* becomes attached to one of the *X*'s and the long arm to the other. In the present case the free *X*-chromosome which arose in this way had the short arm of the *Y* attached to it. For such males to be fertile they must possess in addition either the long arm or a complete *Y*. That the particular male tested carried only the reconstituted scY^L suggests that the latter chromosome had been formed

before separation of the attached-X's occurred, and was widely distributed in the stock, and hence readily became associated with its complementary portion, the short arm, when this became available. This hypothesis is borne out by the fact that many attached-X yellow females that were wild-type in appearance were observed in the stock before yellow or wild males were found, as has been said above.

The likelihood that the scY^L had arisen by crossing-over in the inert material at the left end of the sc^{S1} chromosome made it plausible to



Explanation of symbols: yY^s = X-chromosome marked by yellow, attached to short arm of Y. scY^L = long arm of Y with fragment of X attached bearing the sc^{S1} gene and normal alleles of all genes left of it. yw^a = attached X's marked by yellow and apricot. Y^L = long arm of Y. + = wild-type. The stroke / is used to separate the X from the Y or from that portion of it which segregates independently. Symbols enclosed in brackets indicate the phaenotype.

Fig. 3. Genetic distribution of scY^L.

assume from the start that the fragment of X on the scY^L chromosome included the sc gene and the normal alleles of all genes to the left of scute but no genes to the right of it. By crossing males containing the scY^L to females heterozygous for lethal^{J1} (left of yellow) and to females homozygous for achaete and scute, it was proved that the fragment covered these genes as expected; crosses have not been made, however, involving all the genes to the left of yellow, but it is unlikely that any of these are missing on the scY^L.

Whether the scY^L contained any genes to the right of scute was investigated by using the sc^{19}. This is a translocation of a small section of the X, including yellow achaete scute and the gene immediately to

the right of scute (l_s) from the X to the second chromosome. If this gene were also present on the sc^{Y^L}, males having the sc^{Y^L} and the deficient X but lacking the translocation would survive. As no such males were obtained it was concluded that the sc^{Y^L} contains no genes to the right of scute.

Phaenotypical effects

sc^{S1} males and females containing sc^{Y^L} are phaenotypically distinguishable from those which do not. Phaenotypically sc^{S1} is an extreme type of scute; with sc^4 and sc^{L8} it formed the material for the studies on "position effect" in the work of Muller & Prokofyeva (1934) and Muller & Raffel (1938), and has been very minutely described by them. It shows a reduction of all bristles of the so-called "scute character" as distinguished from those of the "achaete character", though even the latter are occasionally affected by sc^{S1}; there is, moreover, a slight Hairy-wing effect which causes the bristles along the wing veins to be more conspicuous than usual, and the dorso-central bristles to be increased in number. Other frequent characteristics of sc^{S1} are shaven abdomen and nicked wings. Homozygous sc^{S1} females are sterile.

The presence of the sc^{Y^L} chromosome in a sc^{S1} male or female considerably reduces the degree of expression of the character, restoring most of the scutellar bristles in particular, and suppressing the shaven abdomen and nicked wing effects; on the other hand, the number of extra dorso-central bristles is increased as is the expression of Hairy-wing. There is also a tendency for bristles to be longer and thicker, and the body colour is definitely darkened. These characteristics appear to be accentuated by additional doses of sc^{Y^L}. The general viability of sc^{S1} males and females is increased by the presence of sc^{Y^L} and the females are rendered fertile.

Females with attached-X-chromosomes marked by yellow and a sc^{Y^L} covering yellow often differ from wild-type females in having one or two extra dorso-central bristles, and a body colour slightly lighter than normal. The extra bristles, however, are not a constant feature of these hyperploid females, at least when only one dose of sc^{Y^L} is present; it is much more constant as well as more pronounced in the males of the stock, which have an X-chromosome marked by yellow with the Y^S attached and a sc^{Y^L}. This fact is probably due to the relatively higher grade of hyperploidy represented by the combination of the sc^{Y^L} with only one complete X-chromosome instead of with two as in the female.

The presence of the $sc\,Y^L$ is often associated with mosaicism of the "eversporting displacement" type. This is most readily seen in flies carrying yellow on their X-chromosomes and a $sc\,Y^L$. The same mosaicism is found in sc^{S1}, sc^8, and similar scutes in which the loci in the scute region have been placed in a position adjoining the chromocentral region. The phenomenon is well known and has been interpreted as due to a variable condition of the genes of the chromocentral region which is reflected in variations in position effect observable over sizable areas of the body (Noudjin, 1938). It is of interest that this effect is exerted on the loci in question whether they lie in proximity with the chromocentral region of the X or with that of the Y^L.

POSITION EFFECT

In order to study the position effect, if any, on the scute gene in its new position on the Y-chromosome, an X-chromosome lacking the scute locus was obtained. This was done by combining (through crossing-over in a heterozygous female) the left part of an X containing the sc^8 inversion with the right part on one containing the sc^4 inversion. Since the left break of the sc^8 inversion occurred immediately to the left of scute, and that of sc^4 immediately to the right of scute (the right break of both being in practically the same position), crossing-over between these two inversions occurring in the middle region of the X produces two abnormal chromosomes, one which lacks the scute locus entirely and one which contains it in duplicate (at both the right and left ends of the chromosome). The deficient chromosome is lethal and hence cannot usually be obtained in males. By crossing females heterozygous for it to males containing the $sc\,Y^L$ a number of males were obtained in which the deficiency was covered by the $sc\,Y^L$. These males were very similar in all respects to ordinary sc^{S1} males showing that in its new position the sc^{S1} gene has much the same effect as in its normal position on the X. These observations were, however, of a general nature, since a more exact study would require the preparation of stocks so constituted as to make possible the distinction between differences due to autosomal modifiers and those possibly due to variations within or between the two positions of scute.

CONCLUSION

It is conceivable that the rearrangement above described took place as the result of independent breakages of the X- and Y-chromosomes in the positions noted, followed by recombination, as ordinarily happens

in translocations, but it is alternatively conceivable that the recombination took place by means of a process of crossing-over since more or less homologous regions of the *X*- and *Y*-chromosomes were involved and it is known that these regions do undergo occasional crossing-over, even in the male, at least when they lie in their normal positions in chromosomes not containing an inversion. But if the rearrangement took place by crossing-over, either the chromocentral regions involved must have paired in inverted position with regard to each other, or the union must have occurred between adjacent instead of diagonally opposite ends of the two chromosomes, for the general order of the homologous regions in question, of the sc^{S1} and the *Y*-chromosomes, are such that a normal crossing-over in this region would have given only dicentric and acentric chromosomes (see Figs. 4–6). In view, however, of the tendency of all the parts of all chromocentral regions to pair with one another more or less indiscriminately, as observed in salivary gland preparations, as though they were partially homologous, such reversed pairing seems more likely in this case than ordinarily. In fact this generalized conjugation as well as the high tendency to breakage on the part of chromocentral regions makes it doubtful whether in their case a sharp distinction between crossing-over and abnormal processes of recombination, such as translocation, "unequal crossing-over", etc., can legitimately be made.

It is evident that the scY^L chromosome is useful for some studies in which it is desirable to follow the distribution of a *Y*-chromosome (at least in cases where some modification of the latter may be used instead of the ordinary *Y*), since its possession of convenient markers such as y^+, ac^+, allow its presence to be readily detected if the corresponding recessives occur in the *X*-chromosomes.

Summary

1. The case is reported of a chromosome termed scY^L formed by rearrangement between the *Y*-chromosome and an *X* containing the sc^{S1} inversion. The rearranged chromosome has on one side of its centromere the whole of the long arm of the normal *Y* and on the other side a small section invisible in mitotic stages, containing the left distal end of the *X*, with the scute gene and the normal alleles of genes to the left of it.

2. If the breakage and reunion occurred by a process of crossing-over, the chromocentral regions of *X* and *Y* here involved must have been in inverted position in regard to each other or else adjacent instead of

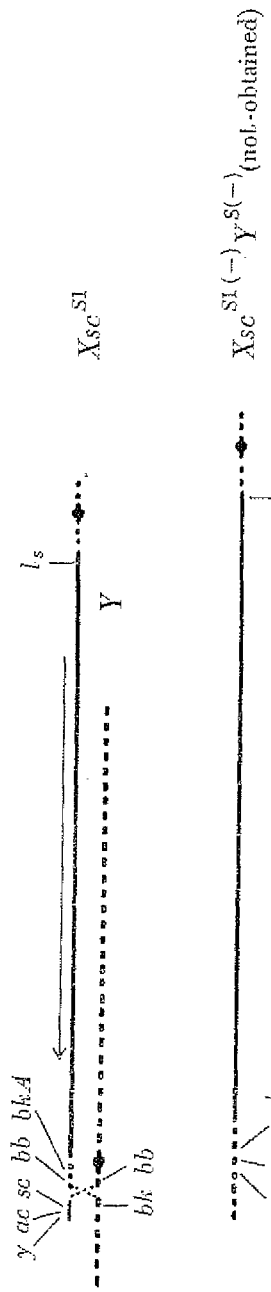


Fig. 4. Reversed pairing between Y^S and the inert material to the right of *sc*, resulting in the formation of the "scY^L".

diagonally opposite ends of the conjugating chromosomes must in this case have become attached together.

3. In its new position on the *Y*-chromosome the scute gene has an

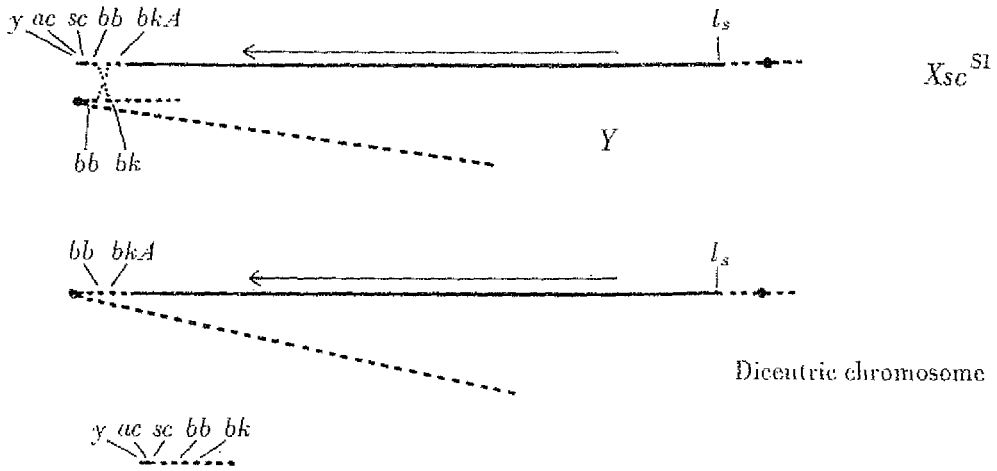


Fig. 5. Normal pairing resulting in the formation of a dicentric chromosome and an acentric fragment. Both would be eliminated.

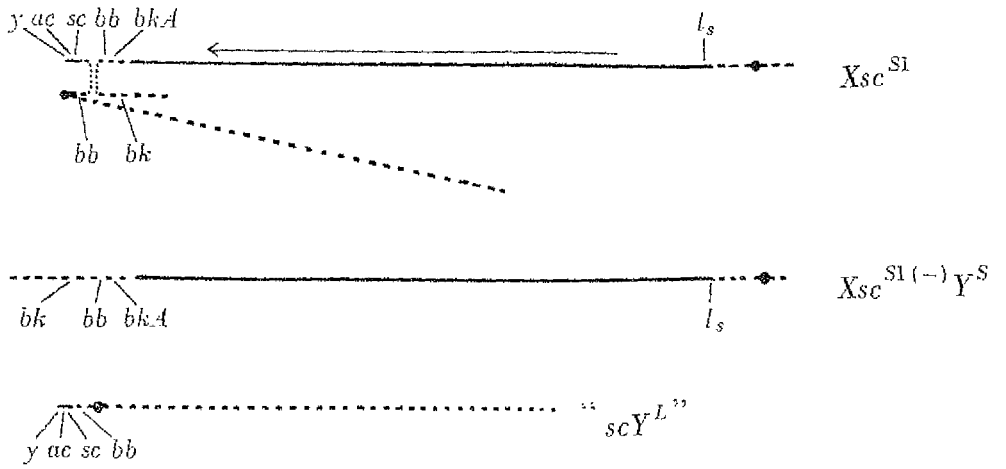


Fig. 6. Normal pairing followed by fusion of adjacent instead of diagonally opposite elements: such a process would give the same results as reversed pairing.

expression similar to that on its old position on the *X*-chromosome. Moreover the position effect is of a mosaic nature in the *Y* as in the *X*.

4. When the scY^L forms a duplication of *sc*, the scute effect is re-

duced and the *Hw* effect accentuated, the body colour is darkened and the homozygous sc^{S1} female (usually sterile) is rendered fertile; bristles are longer and thicker, and general viability is increased. In non-scute flies the scY^L adds bristles in the dorsocentral region, and gives a slight *Hw* effect.

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