A CYTOGENETIC STUDY OF THE YELLOW-SCUTE REGION OF THE X CHROMOSOME IN DROSOPHILA MELANOGASTER

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INTRODUCTION

A DETAILED study of the *y-sc* region in *Drosophila melanogaster* has been made by means of an analysis of a series of changes in the loci yellow (*y*), achaete (*ac*), and scute (*sc*), which are included in that region.

The purposes of this study were (a) to determine the bands of the salivary gland chromosomes with which these loci are associated, (b) to find out the relationship between cytological and genetical changes in this region, and (c) to compare the cytogenetic findings for the *y* and *sc* loci with those for other loci in the X chromosome.

These points will be discussed after the presentation of a preliminary description of the changes studied.

MATERIALS AND METHODS

With a few exceptions, noted in the descriptions, the changes were obtained by irradiating wild type (Swedish-b) males with X-rays at a dosage of 2,500–3,000 roentgen units and picking up induced changes in the *y* and *sc* loci among the F_1 females from the cross *y sc* or *y sc w* females × irradiated males.

The *y* or *sc* mutant females thus obtained were mated with *y Hw w* males, and kept in balanced stocks.

Slides were made from the salivary glands of heterozygous female larvae of the constitution mutant/*y Hw w*, and were stained either with aceto-carmine or with acetic orcein.

The equipment used in analyzing these slides consisted of a 90X, 1.3 N.A. apochromatic objective, with an oil immersed 1.4 N.A. condenser, 12.5X compensating oculars, and a Bausch and Lomb research lamp with the green Wratton filter number 61.

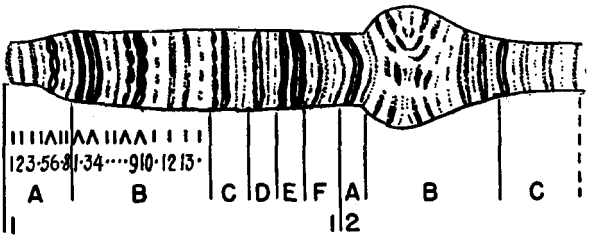

FIGURE 1.—Diagram of left end of the salivary gland X chromosome, with bands numbered after BRIDGES' (1938) map.

DESCRIPTION OF CHANGES

The collection of changes studied can be divided into three groups, as follows: I. Genetic changes not associated with chromosomal aberrations (seven cases). II. Genetic changes associated with chromosomal aberrations (19 cases). III. Cytological changes without genetic effect (two cases).

The cytological analyses are based on BRIDGES' (1935, 1938) maps of the salivary gland chromosomes. The *y-sc* region of the X chromosome is shown in figure 1. Some of the changes included in this collection were analyzed and published previously by DEMEREC and HOOVER (1936a).

I. GENETIC CHANGES WITHOUT CYTOLOGICAL ABNORMALITY

(*a*) *Yellow locus*

1. y^{260-4} (yellow 260-4) Found by DEMEREC 1938, unpublished. From X-rayed *sn B* male. Not lethal. Phenotypically like y^2; *sc* not affected.
2. y^{260-12} (yellow 260-12) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. Phenotypically like y^1; *ac* and *sc* not affected.
3. y^{260-24} (yellow 260-24) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. Phenotypically like y^1; *ac* and *sc* not affected.
4. y^{260-28} (yellow 260-28) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Viability of males reduced. Phenotypically like y^1; *ac* also affected, but not *sc* or *svr* (silver).
5. y^{260-30} (yellow 260-30) Found by BISHOP 1940, unpublished. From X-rayed Sw-b male. Not lethal. Phenotypically like y^1; *ac*, *sc* and *svr* not affected, but *f* (forked) also affected.
6. y^{260-31} (yellow 260-31) Found by FANO 1941, unpublished. From X-rayed Sw-b male (12,000 r). Lethal in homozygous and hemizygous condition. y^{260-31}/y^1 phenotypically like y^1; *ac* also affected. Salivaries show a piece of IIL (24-29) inserted into X about 9A. Aberration independent of change at *y* locus. Genetic tests show that y^{260-31} does not cover the recessive lethal associated with Df 260-25, therefore the recessive lethal in y^{260-31} is associated with the change at the *y* locus and not with the rearrangement.

(*b*) *Scute locus*

1. sc^{260-16} (scute 260-16) Found by SUTTON 1938, unpublished. From X-rayed Sw-b male. Lethal in homozygous and hemizygous condition. *sc* does not always show in sc^{260-16}/sc females; *y* not affected.

II. GENETIC CHANGES ASSOCIATED WITH CHROMOSOMAL ABERRATION

(*a*) *Yellow locus*

1. y^{260-2} (yellow 260-2) DEMEREC and HOOVER 1936 (published as Deficiency 260-2). From X-rayed Sw-b male. Lethal in homozygous and hemizygous condition. y^{260-2}/y^1 phenotypically like y^1; *y* and *ac* affected, but not *sc*. Deficiency for bands 1A1 to 1B1.2 inclusive.

2. y^{260-10} (yellow 260-10) SUTTON 1940 (published as Df (1) 260-10). From X-rayed Sw-b male. Not lethal. Males and homozygous females phenotypically like y^1; also achaete. *y* and *ac* affected, but not *sc*. Deficiency for bands 1A1, 2.

3. y^{260-11} (yellow 260-11) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. Males sterile. Males and homozygous females phenotypically like y^1; *y* affected but not *ac* and *sc*. T(X; IIIR). Break in X between 1B1.2 and 3.4, in IIIR between 85F1 and 5.

4. y^{260-13} (yellow 260-13) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal, fertility of males reduced. Body wild type, bristles yellow in males and homozygous females. *ac* and *sc* not affected. T(X; IIL). Break in X between 1A4 and 5.6, in IIL at 36D. Stock lost.

5. y^{260-21} (yellow 260-21) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Lethal in homozygous and hemizygous condition. y^{260-21}/y^1 phenotypically like y^1; *sc* not affected; not tested for *ac*. Inversion, with breaks between 1A5.6 and 7 and between 5D7.8 and E1.2. T(X; IIIL) also present, with break in X at 6C and in IIIL at 70E-F.

(*b*) *Scute locus*

1. sc^{260-14} (scute 260-14) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. *sc* affected, but not *y*, *ac*, or *svr*. Males and homozygous females scute. Inversion, with breaks between 1B1.2 and 3.4 and between 11D3 and 8.

2. sc^{260-15} (scute 260-15) Found by DEMEREC 1938, unpublished. From X-rayed Sw-b male. Viability reduced. Males sterile. *sc* affected, but not *y* or *ac*. Males and homozygous females scute. T(X; IIIL). Break in X between 1B3.4 and 5, in IIIL between 71C and D.

3. sc^{260-17} (scute 260-17) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. *sc* affected, but not *y*, *ac* or *svr*. Males and homozygous females scute. T(X; IIL). Break in X between 1B1.2 and 3.4, in IIL about 31C.

4. sc^{260-18} (scute 260-18) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. Males sterile. Males and homozygous females scute. *sc* affected, but not *y*, *ac* or *svr*. T(X; IIR; IIIL). Breaks in X between 1A5-6 and 1B1.2 and between 7A1.2 and B1.2; in IIR chromocenter between 41D and E; and in IIIL chromocenter at 80C. New order: Tip of X-1A5.6/41D-sp.a. II-21 (Tip of IIL); tip of IIR-41E /1B1.2-7A/80C-sp.a. III-tip of IIIR; tip of IIIL-80C/7B-sp.a. X. (sp.a. = spindle attachment.)

5. sc^{260-20} (scute 260-20) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. *sc* affected, but not *y*, *ac* or *svr*. Males and homozygous females scute. T(X; IIIL). Break in X between 1A8 and 1B1.2, in IIIL preceding 61A1.2, at extreme tip.

6. sc^{260-22} (scute 260-22) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. *sc* affected, but not *y*, *ac* or *svr*. Males and homozygous females scute. Inversion with breaks between 1B1.2 and 3.4 and between 1E1.2 and 3.4.
7. sc^{260-23} (scute 260-23) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Not lethal. *sc* affected, but not *y* or *svr; ac* not adequately tested. Males and homozygous females scute, apparently not achaete. T(X; ?). Break in X between 1B1.2 and 3.4. Stock lost.
8. sc^{260-26} (scute 260-26) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Viability of males reduced, fertile. *sc* affected, but not *y*, *ac* or *svr*. T(X; IIL; IIR). Break in X between 1B3.4 and 5; in IIL between 27D1.2 and 3; and in IIR at 41A, 41F and 58B. New order: tip of X-1B3.4/41F3-58B/1B5-sp.a. X; tip of IIR-58B/41F-41A/27D-sp.a. II-40/27D-tip of IIL.
9. sc^{260-27} (scute 260-27) Found by SUTTON 1939, unpublished. From X-rayed Sw-b male. Males sterile, with reduced viability. *sc* affected, but not *y*, *ac* or *svr*. Males and homozygous females scute. T(X; IIL; IIR). Breaks in X between 1A8 and 1B1.2, at 15E and at 19F (chromocenter); in IIL at 33-34; and in IIR at 57B-C. New order: Tip of X-1A8/20A-sp.a. X; tip of IIL-33/15E-19F/1B1.2-15E/57B-sp.a. II-34/57-tip of IIR.
10. sc^{260-29} (scute 260-29) Found by SUTTON 1940, unpublished. From X-rayed Sw-b male. Not lethal, males sterile. *sc* affected, but not *y*, *ac* or *svr*. Males and homozygous females scute. T(X; IIL; IIIL). Break in X between 1A5.6 and 1B1.2; in IIL between 22A and B, and between 34A and B; and in IIIL between 75C and E. New order: Tip of X-1A8/34A-22B/34B-sp.a. II-tip of IIR; tip of IIL-22A/75E-sp.a. III-tip of IIIR; tip of IIIL-75C/1B1.2-sp.a.X.
11. *l*(1) 272-13 (lethal (1) 272-13). Found by DEMEREC 1940, unpublished. From X-rayed *y* male. Lethal in homozygous and hemizygous condition. *l*(1)272-13/*sc* phenotypically scute. *sc* affected, but not *y*, *ac*, *svr*. Complex rearrangement of X, with four breaks: first between 1A5.6 and B1.2; second between 11A6.7 and 8.9; third between 11F1.2 and 12A1.2; fourth between 18A3.4 and B1.2. New order: Tip of X-1A5.6/12A1.2-18A3.4/11A6.7-1B1.2/11A8.9-11F1.2/18B1.2-sp.a.X.

(*c*) *Yellow and scute loci*

1. Df(1)*svr* LVM (Deficiency (1) silver L. V. Morgan). Lethal in homozygous and hemizygous condition. *y*, *ac*, *sc* and *svr* affected. Females heterozygous for Df(1) *svr* L.V.M. and for *y*, *ac*, *sc* or *svr* show the recessive character. Deficiency for bands 1A1-1B9.10 inclusive (presence of 1B11, 12 uncertain).
2. Df(1)260-1 (Deficiency (1)260-1) DEMEREC and HOOVER 1936. Lethal in homozygous and hemizygous condition. *y*, *ac* and *sc* affected, but not *svr*. Females heterozygous for Df(1)260-1 and for *y*, *ac*, or *sc* show the recessive character. Deficiency for bands 1A1-1B3.4 inclusive.

3. Tp(1)260-25 (Transposition (1)260-25) SUTTON 1940. From X-rayed Sw-b male. Lethal in homozygous and hemizygous condition. Extreme *sc*, shows variegation for *y* and *ac*. Tp(1)260-25/*sc* females are scute, Tp(1)260-25/*y ac* females show variegation for *y* and *ac*. Transposition; terminal bands 1A1-1B1.2 transferred to X chromocenter. The deficiency for these terminal bands obtained by crossing-over was published as Df(1)260-25 (SUTTON 1940).

III. CYTOLOGICAL CHANGE WITHOUT GENETIC EFFECT

1. Df(1)260-5 (Deficiency (1)260-5) DEMEREC and HOOVER 1936a. Detected cytologically in salivary gland chromosomes of an induced *ct* (cut) stock. Not lethal. *y*, *ac* and *sc* not affected. Males and homozygous females wild type. Deficiency for bands 1A1-4 inclusive.

2. Df(1)260-19 (Deficiency (1)260-19) SUTTON 1940. Detected cytologically in salivary gland chromosomes of a stock of Stern (*yy*/g^2B). Not lethal. *y*, *ac* and *sc* not affected. Males and homozygous females wild-type. Deficiency for bands 1A1, 2.

DISCUSSION

Location of genes in the salivary gland chromosomes

The yellow and achaete loci

In Df(1)260-5, bands 1A1-4 are lost, but the *y*, *ac* and *sc* loci are unaffected. The conclusion can therefore be drawn that all of these loci lie to the right of 1A4, in spite of the fact that in y^{260-10} yellow seems to be affected by the loss of only 2 bands, 1A1, 2 (see below). From each of the stocks sc^{260-20} and sc^{260-27} it was possible to derive a stock hyperploid for the bands 1A1-8, but not for the bands immediately to the right of 1A8. By mating hyperploid females with *y ac* males and backcrossing the hyperploid F_1 females with *y ac* males, it was shown that the duplication for bands 1A1-8 covers the *y* and *ac* loci. Both of these loci must therefore lie within the region 1A5-8. This means that Hairy wing (*Hw*), which is associated with a duplication of 1B1.2 (DEMEREC and HOOVER 1939), is not a duplication of the *ac* locus, as was previously suggested.

The scute locus

In Df(1)260-2 the *sc* locus is unaffected, although bands 1A1-1B1.2 have been lost in this stock. The *sc* locus must therefore lie to the right of 1B1.2. In all the gross rearrangements in which *sc* is affected, the break in this region is either between 1A8 and 1B1.2, between 1B1.2 and 3.4, or between 1B3.4 and 5. No *sc* change was obtained in which the break was further to the right than 1B3.4–5. Since the breaks are clustered around 1B1.2 and 3.4, and since 1B1.2 is excluded by the evidence of Df(1)260-2, 1B3.4 is indicated as the position of the *sc* locus.

"Point mutations" and "position effects" at the yellow and scute loci

Table 1 shows the frequency of mutations associated with different types of rearrangement at the loci of *y* and *sc*. The data for *y* and *sc* are taken from this paper and comparable data for other loci have been provided by DEMEREC and HOOVER (1936b), DEMEREC, KAUFMANN, and SUTTON (1939), and DEMEREC (1940).

In six of the 14 *y* changes, no detectable aberration was found. Three of the remaining eight *y* changes were due to actual deficiency for the *y* locus, and one was associated with a deficiency which did not include the *y* locus, while four were due to gross rearrangements. On the other hand, only one of the 15 *sc* changes showed no aberration, and 12 were due to gross rearrangements or "position effects."

Thus while "point mutations" and changes due to aberrations occurred with about equal frequency at the *y* locus, the *sc* locus behaved as if it were rela-

TABLE 1

Frequency of different types of cytological change associated with genetic changes at the yellow and scute loci.

	y	*sc*
No aberration	6	1
Deficiency	4(3)	2
Rearrangement—with euchromatin	3	7
—with heterochromatin	1	5
Total	14	15

tively stable to "intragenic" changes and relatively susceptible to "position effects." The *sc* locus appears to be more extreme than the *ct* (cut) locus in this respect, and much more so than the *N* (Notch) locus. The yellow phenotype (and scute) can be produced by a point mutation, a deficiency for the locus, or a position effect, all three types being represented in the material under review. The stock y^{260-10} requires some consideration from this point of view, since the change from y^+ to *y* is associated with a deficiency for only two bands, 1A1 and 2. It has been shown above that the *y* locus lies to the right of 1A1, 2, in the region 1A5-8. The yellow phenotype in y^{260-10} therefore cannot be due to deficiency for the *y* locus.

It is difficult to regard this case as being due to a "position effect" caused by the absence of neighboring genes in 1A1, 2, because a similar deficiency (Df (1) 260-19) has no effect on the y^+ phenotype. In spite of the difficulties of cytological analysis in such cases (in relatively unstretched chromosomes, and when the two homologues are superimposed, the deficiency cannot be seen), the author believes that these are both genuine deficiencies, and not the pseudo-deficiencies described by GOLDSCHMIDT and KODANI (1943). They appear consistently the same in all cells where the tips of the two homologues are

well spread and stretched. The analysis was made from a study of several pairs of glands from larvae of the constitutions y^{260-10}/y *Hw w* and Df (1) 260-19/+. The change at the *y* locus in y^{260-10} must therefore be attributed to a point mutation, occurring in one of two ways. (1) The deficiency and the change at the *y* locus may have been quite independent of one another. (2) They may both be results of a single event caused by irradiation. The latter possibility must be taken into account since it has been shown (DEMEREC and FANO 1941) that deficiencies covering several bands may be caused by such a single event.

Among the *sc* rearrangements there are four involving heterochromatin and eight involving euchromatin. Heterochromatin is known to affect loci at some distance from the translocation point, but in euchromatic rearrangements the effects are limited to a shorter distance. Of the euchromatic *sc* rearrangements, five out of eight have a break immediately adjacent to the *sc* band, 1B3.4, either to right or left, and the other three have a break removed to the left of 1B3.4 by one band. Not all breaks immediately adjacent to 1B3.4 are effective in producing *sc* changes, however, as evidenced by y^{260-11} in which there is a break between 1B1.2 and 3.4, and the *y* locus, but not the *sc* locus, is affected.

In this sample of *y* and *sc* changes, the effective range for breaks which affect the *y* locus is the region between 1A2 and 1B3.4, and that for the *sc* locus is the region between 1A8 and 1B5. The data for the *ct* and *N* loci show narrow limits for the "sensitive region" similar to the limits for *sc*. A comparison of "sensitive regions" for several different loci has been made by DEMEREC (1940).

MULLER (1935) suggested the possibility that "position effects" were due to interaction between gene products rather than between the neighboring genes themselves. If this were so, the greater extent, for some genes, of the surrounds ing region in which breaks can produce effective changes could be explained on the further assumption that these genes form primary products in higher concentrations than do other genes; and these products therefore diffuse more rapidly away from the center of production, and are thus enabled to come in contact with and interact with the products of other genes beyond the point of breakage. An alternative assumption would be that in the case of these genes the interacting substances are not the immediate products of genes synthesis, but later products in the chain of reactions between the gene and its ultimate phenotypic expression, so that the time interval before their formation allows the intermediate gene products to diffuse over a greater distance.

SUMMARY

A cytogenetic analysis was made of 28 cytological or genetic changes in the distal region of the X chromosome, in which the loci *y*, *ac*, and *sc* are included.

It is shown that the yellow and achaete loci are associated with bands 1A5-8 of the salivary gland X chromosome, while the scute locus is associated with the 1B3.4 band.

The different types of changes at the *y* and *sc* loci are discussed.

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