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NOTES AND COMMENTS

CHROMOSOME BREAKAGE: A POSSIBLE MECHANISM FOR DIVERSE GENETIC EVENTS IN OUTBRED POPULATIONS

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SUMMARY

Evidence is summarised to support the hypothesis that genetic characteristics frequently assayed in natural populations, including mutator activity, sterility, male recombination, and distortion of segregation, may often be expressions of the same general event—chromosome breakage in outbred populations.

1. HYPOTHESIS AND DISCUSSION

THE interesting article on female sterility in *Drosophila melanogaster* by Kearsey, *et al.* (1977) and other recent articles (Bucheton, *et al.*, 1976; Picard, 1976) have stimulated us to discuss a variety of phenomena that at first appear diverse, but that may in fact be expressions of the same basic phenomenon.

A number of events are observed when some wild populations of *D. melanogaster* are crossed to laboratory strains. These events are termed hybrid dysgenesis by Kidwell and Kidwell (1976) and Sved (1976) and include recombination in males, sterility in hybrids, mutator activity, and segregation distortion. We now believe that all of these events are caused by chromosome breakage in outcrossed lines. The evidence for this hypothesis is summarised here.

Recombination in outcrossed *D. melanogaster males*, which was first reported by Hiraizumi in 1971, has now been reported in numerous populations world wide (Hiraizumi, 1971; Hiraizumi, *et al.*, 1973; Slatko and Hiraizumi, 1973, 1975; Voelker, 1974; Waddle and Oster, 1974; Yamaguchi and Mukai, 1974; Cardellino and Mukai, 1975; Kidwell and Kidwell, 1975*a*, 1975*b*, 1976; Sochacka and Woodruff, 1976; Sved, 1976; Yamaguchi, 1976; Woodruff and Thompson, 1977*a*). In contrast, most laboratory mutant and wild type lines, such as the commonly used *Canton-S* wild type, do not undergo recombination in heterozygous males (Slatko and Hiraizumi, 1973; Kidwell and Kidwell, 1975*a*; Woodruff and Thompson, 1977*a*). From detailed analysis of many of the male recombination lines, it is clear that many, if not most, of them also show other genetic events. These events include mutator activity, the induction of unique chromosome rearrangements, segregation distortion, and sterility.

The point we would like to emphasise is that the evidence now strongly suggests that these events are probably common in many natural populations and that they are caused by chromosome breakage. Thus, even in studies in which one is looking at sterility or at mutation rates, the other events may

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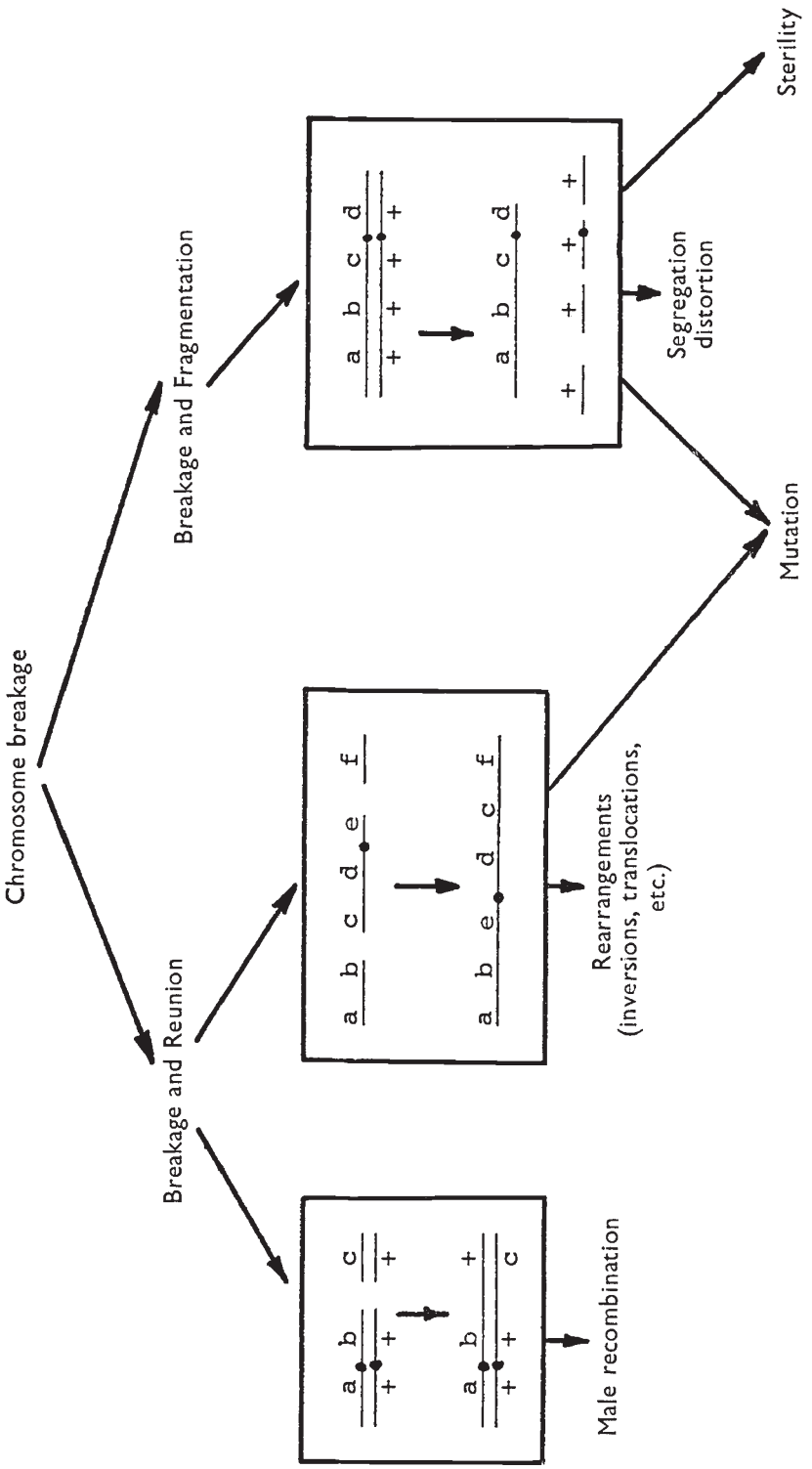

FIG. 1.—Possible consequences of chromosome breakage in outbred natural population lines. A wide diversity of genetic events in natural populations can be attributed to breakage.

be occurring and should not be ignored in planning or interpreting experiments. This is summarised in fig. 1.

Hiraizumi (*e.g.*, Hiraizumi, *et al.*, 1973) and others (Voelker, 1974; Yamaguchi and Mukai, 1974; Cardellino and Mukai, 1975; Yamaguchi, 1976), for example, have suggested that male recombination may be caused by chromosome breakage and reunion. It is certainly clear that many natural populations which show male recombination when outcrossed to marker stocks also undergo frequent chromosome breakage. This is apparent from the recovery of rearrangements in analyses of salivary gland chromosomes from male recombination lines (Voelker, 1974; Yamaguchi and Mukai, 1974; Cardellino and Mukai, 1975) and from direct observations of chromosome breakage in spermatocytes. Woodruff, Henderson, and Thompson (in preparation) and Woodruff and Thompson (1977*b*) found that the male recombination line *OK1* has a high frequency of chromosome breakage and fragmentation at anaphase in spermatocytes. Approximately 20 per cent of anaphases of *OK1* outcrossed males showed chromosome aberrations, whereas the *Canton-S* controls showed none. We believe that this frequent chromosome breakage leads to recombination by rejoining of broken sister chromatids. In addition, the observed chromosome breakage could induce the other genetic events, including mutator activity.

Many outcrossed natural populations have been observed to show mutator activity or the induction of unique chromosome rearrangements (Minamori and Ito, 1971; Slatko and Hiraizumi, 1973; Voelker, 1974; Yamaguchi and Mukai, 1974; Cardellino and Mukai, 1975; Yamaguchi, 1976; Kidwell, Kidwell, and Ives, 1977). When tested, they often show the other events associated with hybrid dysgenesis. The production of new mutations and of unique chromosome rearrangements are common consequences of chromosome breakage.

Distortion of segregation has also been observed in many wild populations (Hiraizumi, 1971; Slatko and Hiraizumi, 1973). There is an unequal recovery of heterozygous (+ +/*ab*) and homozygous (*ab*/*ab*) progeny from crosses of + +/*ab* males with *ab*/*ab* females. As previously mentioned, these lines also show other manifestations of hybrid dysgenesis. Such distortions of segregation may be caused by preferential fragmentation and loss of chromosomes. For example, an excess of *ab*/*ab* progeny can be caused by chromosome fragmentation in + + bearing sperm, leading to inviable zygotes. A similar pattern of preferential chromosome breakage has been observed in *Segregation Distorter* (*SD*) (Crow, Thomas, and Sandler, 1962) and a meiotic drive system (*RD*) (Erickson, 1965) in *D. melanogaster*.

Kearsey *et al.* (1977) have observed that some outcrossed lines show high frequencies of sterility among F_1 female offspring. A number of similar studies have found that the F_1 females or the F_1 males, but not both, show high levels of sterility (Kidwell and Kidwell, 1975*a*; Bucheton, *et al.*, 1976; Picard, 1976; Sved, 1976; Kidwell, Kidwell, and Ives, 1977; Woodruff and Thompson, 1977*a*). Although Kearsey and his colleagues did not assay for other genetic events, other lines from natural populations that have sterile hybrids also undergo male recombination, segregation distortion, mutator activity, and other events attributable to chromosome breakage. One such line, *T-007*, was collected from Texas (Hiraizumi, 1971) as were the lines used by Kearsey.

In summary, we would like to make two points. The first is that evidence is accumulating to support the hypothesis that many apparently independent genetic events may often be due to chromosome breakage, and studies of natural populations should keep this possibility in mind when experiments are designed or results are interpreted. The second comment is that these chromosome breakage events might in fact have even wider interest and importance than might at first be thought. Breakage appears to occur primarily, if not solely, in outcrossed lines (Woodruff and Thompson, 1977*b*). Thus, it may be similar to the chromosome breakage attributed to chromosome-cytoplasm interactions in interspecific hybrids (see, for example, Hennen, 1963) and contributing to genetic isolation between populations and species (Stutz, 1976). Hopefully, detailed studies, like that of Kearsey, *et al.*, but also taking into consideration the other genetic events outlined here, might contribute to a better understanding of the interaction between cytoplasm and chromosomes and its role as a potential isolation barrier.

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