DO SATELLITE DNA'S FUNCTION AS STERILITY BARRIERS IN EUKARYOTES?

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ABSTRACT: Satellite DNAs located in the constitutive heterochromatin in the chromosomes may function as sterility barriers between diverging incipient species in eukaryotes. Satellite DNAs seem not to be adaptive and to be a means of speciation independent of phylogenetic evolution.

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Eukaryote genomes contain satellite DNAs, which are so named because they form satellite bands when the whole DNA of a given species is centrifuged to equilibrium in a CsCl or Ag^{+}-Cs_2SO_4 gradient in the analytical ultracentrifuge (Kit, 1961; Corneo, Ginelli, Soave and Bernardi, 1968).

Satellite DNAs differ in density from the main band DNA because they have a different base composition and/or a peculiar and very highly repeated base sequence (Britten and Kohne, 1968; Southern, 1970). They constitute the most highly repeated fraction of the eukaryote genome. Satellite DNAs account for a variable percentage of the total DNA in different species, but the base sequence and the relative percentage of each satellite appear to be constant in different individuals and in general also in different tissues within a single species. Notable exception are some Diptera (Endow and Gall, 1975) and plants (Pearson, Timmis and Ingle, 1974) in which the relative percentage of each satellite is higher in germ cells and in diploid cells, such as nervous tissue, and is lower in tissues containing polyploid nuclei, because of under-replication of satellite DNA. This is particularly evident in polytene nuclei of larval salivary glands in some Diptera (Schweber, 1974).

Any species may contain one, two, three and exceptionally more satellite DNAs. They are present only in eukaryotes. The satellite-like DNAs found in bacteria are due to episomes.

A few nuclear satellite DNAs have been shown to contain repeated genes, coding for ribosomal RNA (Birnstiel and Grunstein, 1972), transfer RNA (Clarkson, Birnstiel and Purdom, 1973), 5 S RNA (Brown, Wensink and Jordan, 1971) and histones (Birnstiel, Telford, Weinberg and Stafford, 1974). The majority of the nuclear satellite DNAs in eukaryotes, however, do not appear to code for any protein and are probably not even transcribed 'in vivo' (Melli, Ginelli, Corneo and Di Lernia, 1975). They are made up of very short nucleotide sequences, repeated hundreds of thousands or a million times in the genome, where they are clustered mainly (Pardue and Gall, 1970; Jones, 1970; Jones and Corneo, 1971) but not exclusively (Miklos and Nankivell, 1976) in the pericentromeric heterochromatin in the chromosomes.

Another property of nuclear satellite DNAs is that their short highly repeated nucleotide units often contain a high number of base mutations. This is shown by carrying out melting profiles of reassociated satellite DNAs (Corneo, Ginelli and Polli, 1970), or by direct analysis of their base sequence (Southern, 1970).

Satellite DNAs in general appear to be species restricted; however, related species may have very similar satellite DNAs (Jones, Prosser, Corneo, Ginelli and Bobrow, 1972; Gall and Atherton, 1974). This is probably due to the fact

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that these satellite DNAs appeared during evolution before the separation of the two species.

Several functions have been ascribed to satellite DNAs. It has been suggested that they give a selective advantage to the chromosomes that carry them because they would protect the chromosomes during meiosis. Satellite DNAs might be a means of recognition of homologous chromosomes in meiosis (Walker, 1971). How this could happen is unknown. A possible hypothesis is that highly repeated sequences such as satellite DNAs have slightly different molecular conformations according to their peculiar base sequence.

Pairing of homologous chromosomes might occur by chromosomal proteins which bind to specific DNA sequences, and repeated DNA sequences are the most likely candidates to have this function (Mayfield and Ellison, 1975).

If satellite DNAs have such a mechanical function of favouring the pairing of homologous chromosomes in meiosis it is likely that their structural and molecular conformation related to their sequence repetition is more relevant to their function than is their base sequence. Base sequence in the very highly repeated sequence of satellite DNAs is obviously also very important because its specificity may condition the peculiar molecular conformation of each satellite DNA. This occurs because the specific sequence unit of a satellite DNA is repeated many times.

However, the base sequence of a single unit is not so important with regard to the function of satellite DNAs, within certain limits, and one can presume that it is not submitted to a strong pressure by natural selection. Satellite sequences would therefore accumulate base mutations which could be multiplied many times by saltatory replication (Britten and Kohne, 1968). By this mechanism a short sequence is supposed to be multiplied many times to form the very highly repeated satellite sequences.

Here it is suggested that satellite DNAs not only allow the pairing of homologous chromosomes in meiosis, but also hinder the pairing of homologous chromosomes in hybrids of species having differing satellite sequences. The appearance of new satellite DNAs in evolution by saltatory replication could be an important genetic mechanism in speciation independent of natural selection. Satellite DNAs could function as sterility barriers in hybrids of even closely related species.

The evolution of satellite DNAs might occur at a rate independent of natural selection, being controlled by mutation, while phenotypes evolve at an extremely variable rate by natural selection.

If satellite DNAs constitute the major sterility barrier in hybrids of different eukaryote species, this could explain some situations occurring in nature. The presence of widely differing varieties within the same species could be due to high selective pressure on phenotypes of different populations of the same species, while sterility barriers have not yet been formed because the evolution of satellite DNAs is relatively independent of natural selection. On the other hand, so-called sib species could be due to a strong unifying selective pressure for small phenotypic differences between the two species while the presence of differences between the satellite sequences, which appeared independently of natural selection, could form the sterility barrier responsible for the separation of the two species.

However, it is known that sterility barriers and other post-mating mechanisms are less important than pre-mating mechanisms in determining reproductive isolation at least in animals. Therefore new species might evolve without sterility barriers and have the same satellite DNAs.

Mechanisms of meiosis have been recently reviewed by John (1976), who stated that heterochromatin association does not have an essential role in pairing of homologous chromosomes. My proposed hypothesis that satellite DNAs hinder the

pairing of homologous chromosomes in meiosis of sterile hybrids of related species is not necessarily in contrast to this statement, nor is it in contrast to the observations that euchromatin synapses first and there is not early association of heterochromatin in homologous pairs (John, 1976) and that in some species the same satellite is found on many or all of the chromosomes (Jones, 1970). In fact the presence of different satellite DNAs on homologous chromosomes in hybrids may prevent the completion of the homologous pairing already begun in other sectors of the genome. The different amounts of difference between satellite DNAs in the parental species may explain the different amounts of hybrid sterility occurring in nature.

Satellite DNAs may have appeared through saltatory replication or by multiple subsequent crossing over (Southern, 1975; Smith, 1976). This latter hypothesis, recently proposed, is in agreement with my hypothesis that repeated DNAs are not controlled by natural selection, and it gives a mechanism for the maintenance of satellite DNAs during evolution.

The new satellite sequence is likely to appear in a geographically isolated population, and to spread to all the individuals of this population. It is likely that there is a considerable polymorphism in the satellite sequences of a population. However, only in few cases could extreme variations determine pathological anomalies in meiosis and sterility. This could be a means of eliminating too wide deviations from the average in a single population and could explain the relative stability of satellite DNAs in a single species. Perhaps some sterile crosses within species and even within populations have this cause. On the other hand, in reproductively isolated populations different satellite sequences could evolve, leading to the establishment of sterility barriers between two new incipient species.

The study of satellite DNAs in sib species, in species which form hybrids and in species closely related phenotypically (Mazrimas and Hatch, 1972; Gall and Atherton, 1974) may open new perspectives on molecular evolution.

It may be suggested that multiple chromosomal rearrangements are enough to explain interspecies sterility. However, there are cases, like some Hawaiian Drosophila (Carson et al., 1967; Ahearn et al., 1974), in which there is an extraordinary karyotypic stability even when related species are isolated by hybrid sterility. Karyotypic rearrangements may be only incidental accompaniments of speciation (Carson et al., 1967) and may be a consequence of changes in repeated DNA sequences if these are involved in chromosome organization. One could postulate that chromosomal rearrangements are cytological aspects of a molecular phenomenon of which changes in satellite DNA sequence and structure may be a major aspect.

In conclusion, it seems relevant to emphasize that in eukaryote genomes one can make a definite distinction between genes (DNA sequences most of which are unique, which are transcribed and translated, and the function of which is to control the phenotype) and very highly repeated DNA sequences such as satellite DNAs, which are not transcribed, which are genetically inert, and which probably have a role in chromosome organization. Genes are controlled by natural selection, while satellite DNAs are relatively independent of natural selection.

I propose that while pre-mating mechanisms of reproductive isolation, such as ethological mechanisms, are controlled by natural selection, such a post-mating reproductive isolation mechanism as hybrid sterility, based on differences in satellite DNA sequences, is independent of natural selection. This implies that events of speciation which are due to this second mechanism are independent of natural selection.

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