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Gitte Petersen

*University of Copenhagen*

Ole Seberg

*University of Copenhagen*

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# Genomes, Chromosomes, and Genes and the Concept of Homology

Petersen, G. & Seberg, O.  
Botanical Laboratory  
University of Copenhagen  
Gothersgade 140  
DK-1123 Copenhagen K  
Denmark

## ABSTRACT

The traditional application of genome analysis in phylogenetic inference is questionable. Hypotheses about phylogeny are based upon the analysis of homologous characters, existing as a consequence of common descent. The concept of homology in morphology and molecular biology is well-defined: To count as an homology any character must pass the similarity, congruence, and conjunction tests. In genome analysis homology is related to the behaviour of chromosomes during meiosis: homologous chromosomes pair, nonhomologous chromosomes do not. Thus, in genome analysis homology becomes a purely operational concept. How well does this operational concept work? And what are the relationships, if any, between this operational concept of homology and the homology concept of morphology and molecular biology?

## INTRODUCTION

As a discipline genome analysis was formally founded and outlined by Kihara (1930) and has been applied extensively, not least within the Triticeae, to studies of systematics and evolution ever since. More recently, Dewey (1982: 52) stated that: "*The fundamental premise of genome analysis is that like (homologous) chromosomes pair during meiosis and unlike (nonhomologous) chromosomes do not. The corollary premise is that the level of chromosome pairing in a species-hybrid reflects the degree of relationship between the parental species.*" Thus, genome analysis is strongly dependent upon the homology of chromosomes, and hence upon the concept of homology.

The concept of homology in morphology and molecular biology is well-defined (Patterson 1982, 1988). To qualify as homologous any character must pass the three tests of homology: similarity, conjunction, and

congruence. The test of similarity is intuitively the most obvious, as we would not even consider the possibility of two characters being homologous if we observed no similarity at all between them (Stevens 1984). The test of conjunction deals with the duplication of a character within an organism. e.g., because of the position of the bract scales the female cone of a conifer is considered a condensed shoot and thus homologous to the entire female cone of a cycad (Florin 1944). Because of the presence of bract scales the ovuliferous scales fail the conjunction test. The third test concerns the congruence of one homology with other homologies, and is closely linked to the principle of parsimony. Monophyletic groups are characterized by synapomorphies (=homologies) and the hypothesis of one homology is tested by presence of other synapomorphies. Failing this test may be caused by e.g., parallelism or convergence.

These three tests are equally valid in morphology and molecular biology, the only difference residing in the terminology and in the relative importance attached to the three tests (Patterson 1988). However, in genome analysis homology is being related to chromosome pairing, i.e. to the behaviour of the chromosomes during meiosis. Thus, the concept of homology has been turned into something purely operational. But how then, does this operational concept of homology relate to the homology concept of morphology and molecular biology?

## HOMOLOGY AND CHROMOSOME PAIRING

Since the very early studies of chromosome pairing during meiosis it has been suggested that the pairing chromosomes were homologous (e.g., Sutton 1902, McClung 1908). This was based upon the apparent similarity of the pairing chromosomes and on their assumed descent from a male and female parent. Often it will be so

that two pairing chromosomes are truly homologous. The 4A chromosomes in one plant of hexaploid wheat are most likely homologous to the 4A chromosomes in another wheat plant. We can infer by their apparent similarity, common descent. Thus, chromosomes regarded as entities surely can be homologous. Just as chromosome arms, genes, or other well defined parts of the chromosomes can. We may run into some problems when changes such as translocations, inversions, or substitutions occur, but that will only be a matter of addressing the problem at the "correct" level. The above chromosomes 4A will no longer be homologous if one of them, because of a translocation, carries the short arm of chromosome 4D instead of its own 4AS, but the two 4AL's will still be homologous. Trying to assign a "degree" of homology to the chromosomes 4A and 4AL/DS would be absurd.

As pairing chromosomes within a species usually were homologous, the idea emerged that the degree of pairing could measure the degree of homology and further assess a level of organismal relatedness (e.g., Federley 1914, Kihara 1924, 1930). Thereby the concept of homology was changed into a purely operational one, which is still used in genome analysis. This use of homology raises two major questions.

The first question addresses the relationships between chromosome pairing, chromosome similarity, and DNA similarity. In genome analysis it is assumed that the ability of chromosomes to pair estimates an overall similarity of the total amount of DNA (e.g., Alonso & Kimber 1981, Chapman & Kimber 1992). However, less than 1% of the total amount of DNA is trapped in the synaptonemal complex (Wettstein et al. 1984). As for the similarity of the remaining 99% of DNA we have virtually no knowledge.

The extrapolation from chromosome pairing to DNA similarity is purely *ad hoc*. It is completely unknown to what extent differences in base composition influences pairing ability, both with respect to coding (e.g., genes) and non-coding sequences (e.g., the 70% or so repetitive DNA that occurs in the Triticeae genome). Thus, the invoked relationship between pairing and DNA similarity is more an article of faith than a scientific theory.

The concept of chromosome similarity as viewed macroscopically is equally elusive. Hence it is only very rarely known, whether the chromosomes involved in the pairing in one hybrid combination are the same as observed in another combination.

Further, it is well documented that chromosome pairing is under genetic control, e.g., the Ph-gene of chromosome 5B in *Triticum* L. (e.g., Holm 1986). Functioning/non-functioning of this gene can change pairing from virtually zero to 100%. Thus, a very small change, perhaps just a one-basepair mutation, could make the interpretations from genome analysis change from total similarity to total dissimilarity between two genomes. This of course is the extreme situation, but any genetic or

environmental factor (e.g., temperature [Pickering 1990] or nutrition [Bennett & Rees 1970]) having an influence on chromosome pairing will contribute so that the observed chromosome pairing does not reflect DNA similarity.

The second major question concerns the relationship of homology to phylogeny, and hence the congruence test. Previously both Kellogg (1989) and Seberg (1989) have stated that the ability of chromosomes to pair and hence inferred as homologous as defined by Dewey (1982), is the plesiomorphic character state. The ability to pair tells us only that the chromosomes/genomes have not diverged. Thus, the pairing ability of chromosomes cannot be used in phylogeny reconstruction as only apomorphic character states are informative.

Intermediate levels of chromosome pairing (= the average chiasma frequencies) assessed by genome analysis are not discrete character data but distance data, and thus cannot be transformed into character data. As such they offer no opportunity to examine notions of homology, and in phylogenetic inference they provide very little opportunity for further research (Eernisse & Kluge 1993). Thus, it remains to be proven that homology expressed as pairing ability passes any of Patterson's (1982, 1988) tests, apart perhaps the conjunction test.

## THE OPERATIONAL CONCEPT OF HOMOLOGY - HOW DOES IT WORK?

What is measured in genome analysis is usually the definite number of chiasmata in a definite number of cells. Thereafter an average value of chiasmata per cell is calculated. But what does this average value represent? Assume that we in a diploid hybrid with  $2n=14$  chromosomes, observe cells with every number of chiasmata from one to 13, with an average frequency of 8.6 chiasmata per cell (as in the hybrid *Hordeum brachyantherum* Nevski x *H. muticum* Presl [Bothmer et al. 1986]). Most genome analysts would not put much emphasis on the observed range, but would regard the average value as an indication of a fairly high level of homology between the genomes, and consequently consider the species quite closely related. But what about the chromosome behaviour in the cells with only one chiasma or 13, respectively? One chiasma would indicate a fairly low level of pairing and little homology between the genomes, whereas 13 chiasmata would indicate a high level of pairing and homology. But these two cells nevertheless would (for all practical considerations) contain exactly the same DNA. So we must ask, what is then the biological rule that tells us that the level of "homology" or "relatedness" is given by the average value and not by any of the extremes.

It seems to become even more difficult to interpret the mean values, when looking at the chiasmata distributions in hybrids (Fig. 1, 2). One might have

expected that chiasmata distributions typically would be either binomial with the top-point equalling or close to the mean value (Fig. 2A: *Aegilops geniculata* Roth x *Triticum durum* Desf., 2B: HH 10183-1), or form distributions sloping steeply from either zero in hybrids with virtually no pairing (Fig. 2D: HH 10339-2) or from the absolute maximum number of chiasmata in hybrids or species with normal, full pairing. However, this is rarely the case. The top-point may be strongly skewed (Fig. 1A), the curve may be flattened (Fig. 2D: HH 10339-1), there may be no apparent top at all (Fig. 1B), or there may be more than one top (Fig. 1C: BB 7271a, 1D). If the distribution of chiasmata is skewed, then the average value will be either higher or lower than the most frequently occurring number of chiasmata, and the modal value would better represent the chiasmata distribution than the mean value. If all observed numbers of chiasmata per cell occur with almost the same frequency, the average value seems hardly more representative than any other value. In cases where the distribution is bimodal the average may be closer to the trough between the two peaks than to any of the maximum values (e.g., Fig. 1D: BB 7511b with an average chiasma frequency 17.4). We have most clearly observed bimodal distributions in tetraploid hybrids, and it is possible that such distributions are caused by the combination of two different pairs of genomes having different levels of pairing. If so, combining the distributions into one average chiasma frequency seems absurd.

Here we shall not attempt to answer in depth what it signifies that the average number of chiasmata deviates from the most frequently occurring number(s), but merely ask what biological relevance the average value has over any other value.

One further, serious problem in the use of average chiasma frequencies to assess phylogenetic relatedness is the variation between values that can be obtained from reciprocal hybridization and between progeny from hybrid combinations involving the same parental species. Few, if any studies, since Kihara (1929) have focused on these problems, though the observed discrepancies ought to be most alarming to any genome analyst.

In reciprocal hybrids involving *Triticum* and *Aegilops* L. (Fig. 2A) Kihara (1929) observed quite deviating patterns of chromosome pairing. In one hybrid virtually no pairing occurred, whereas in the reciprocal an average of approx. 4 chiasmata per cell were observed. Lu & Bothmer (1993b) observed significantly different pairing in reciprocal hybrids between *Elymus caucasicus* (C. Koch) Tzvelev and *E. tibeticus* (Melderis) G. Singh, and the difference would place the hybrids in each of two groups, defined by Lu (1993) to distinguish five different levels of chromosome pairing. As these levels are being interpreted as a measure of phylogenetic relatedness, it must be disturbing that reciprocal combinations give different measures of distance between the same parental species.

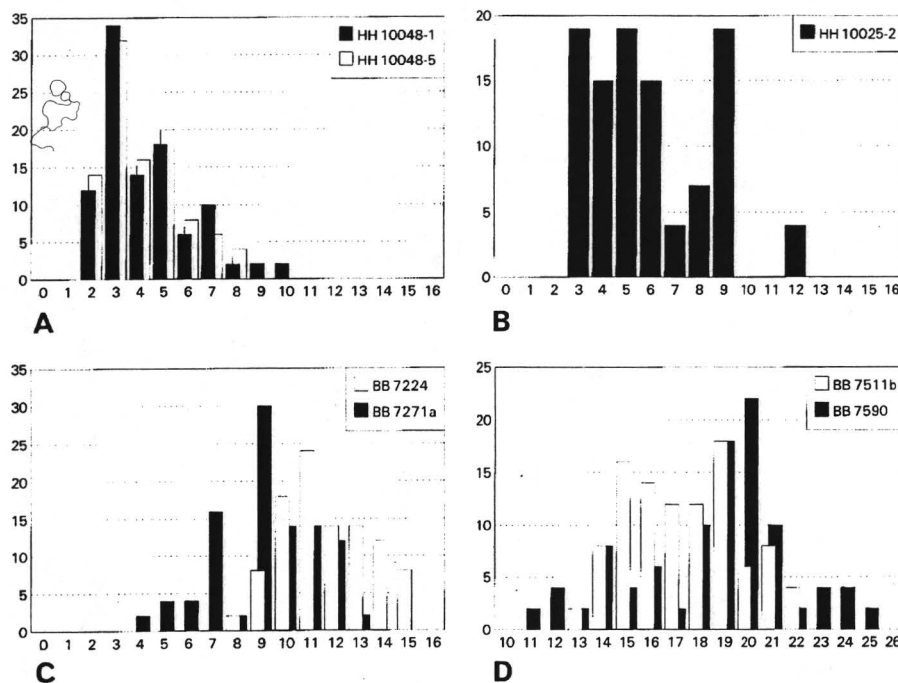


Figure 1 Distribution of chiasmata in hybrids. A, B: *Hordeum brachyantherum* (4x) x *Secale cereale*, two crosses involving different parental accessions. C: *Elymus tschimganicus* (Drob.) Tzvelev x *E. caninus* (L.) L., two crosses involving different parental accessions. D: *Elymus tschimganicus* x *E. caucasicus*, two crosses involving different parental

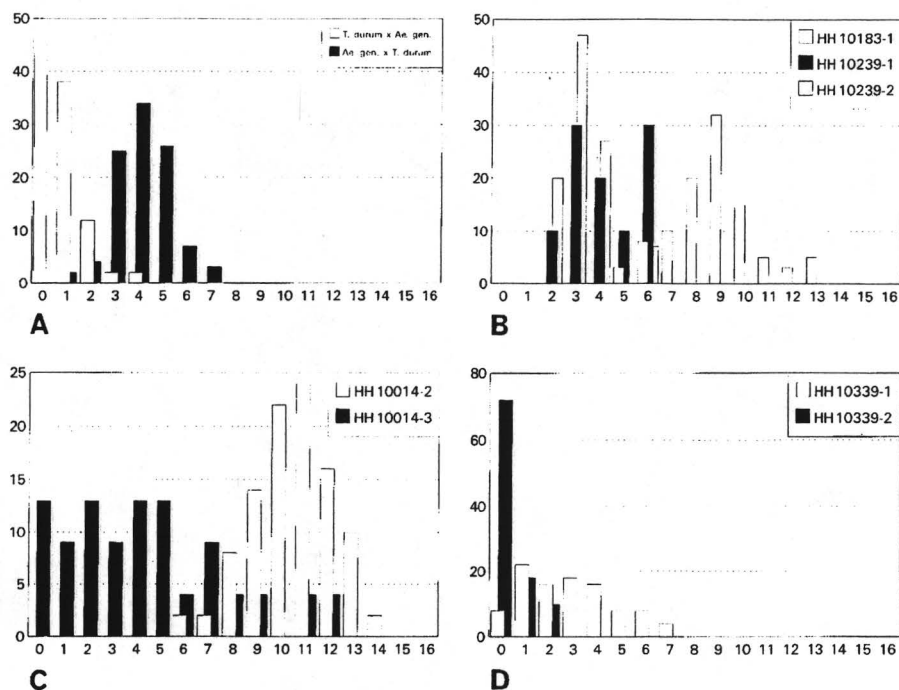


Figure 2 Distribution of chiasmata in hybrids. A: Reciprocal hybrids between *Triticum durum* and *Aegilops geniculata* (Data from Kihara 1929). B: *Hordeum procerum* x *Secale cereale*, two crosses involving different parental accessions. C: *Hordeum brachyantherum* (4x) x *Secale cereale*, two plants from the same cross. D: *Hordeum capense* Thunb. x *Secale montanum* Guss., two plants from the same cross. X-axis: Number of chiasmata; Y-axis: Percentage of cells.

Similar differences in chromosome pairing can be observed when comparing offspring from different crosses of the same hybrid combination. e.g., hybrids from two different crosses between *Elymus brevipes* (Keng) Löve and *E. tsukushiensis* Honda had average chiasma frequencies of 20.66 and 8.19, respectively (Lu & Bothmer 1993a), and offspring from one cross between *Hordeum procerum* Nevski and *Secale cereale* L. (Fig. 2B: HH 10183-1) had more than twice the high average chiasma frequency than offspring from a second cross (Fig. 2B: HH 10239) (Petersen 1991). Even individual plants from the same cross may exhibit strongly deviating patterns of chromosome pairing (Fig. 2C, D). In two hybrids between *Hordeum brachyantherum* (4x) and *Secale cereale* the average chiasma frequencies were 10.50 and 4.39, respectively (Fig. 2C) (Petersen 1991). As in these hybrids only *Hordeum* L. chromosomes take part in the pairing, one hybrid shows almost complete pairing of the *Hordeum* chromosomes (average 6.42 bivalents per cell) whereas in the other, only half of the chromosomes are paired (average 3.85 bivalents per cell) (Petersen 1991). Thus, following genome analysis the two genomes of *Hordeum brachyantherum* should in one hybrid be interpreted as almost fully homologous and in the other as only partly homologous.

If the average chiasma frequency is accepted as a measure of relatedness (= "overall similarity") between

two species, this may only be extended to more inclusive groups of species by using phenetic clustering methods (e.g., UPGMA [Lu 1993]). However, that the overall similarity (and phenetic clustering) is an unsatisfactory measure of phylogenetic relationships is beyond debate.

## CONCLUSION

It seems a paradox that it was Kihara (1930), who immediately after having observed great differences in chromosome pairing between reciprocal hybrids, within offspring from crosses, and even within florets from just one spike (Kihara 1929), formulated the theories and practices of genome analysis. Kihara (1929) assumed that most of the variation could be explained by the influence of environmental factors. Though this may to some extent be true, other factors, not least genetic, may be strongly influential, too. Both factors make reproducibility and comparisons a difficult matter. However, we do not here aim at speculating about possible ways in which chromosome pairing may be affected, but merely wish to demonstrate some of the patterns and magnitude of the variation that are not addressed or deliberately neglected in genome analysis. Variation obscures the biological relevance of the mean values, which are the underlying basis for genome analysis.

The conversion of chromosome pairing data into a measure of homology and phylogenetic distance is questionable on the basis of the conceptual discrepancy alone. There is no known relationship between the theoretically formulated definitions of homology in classical morphology and molecular systematics and homology defined as chromosome pairing (Moritz & Hillis 1990). As previously stressed, by e.g., Kellogg (1989) and Seberg (1989), the presence or absence of pairing may to the extent it represents states of the same character, be used in phylogenetic reconstruction. The degree of pairing,

though being mathematically well-defined, can only be used in phenetics and hence it is phylogenetically incomprehensible.

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