J. evol. Biol. 4: 167–194 (1991)

1010–061X/91/02167–28 $ 1.50 + 0.20/0

Homology and hierarchies: Problems solved and unresolved

V. Louise Roth

Department of Zoology, Duke University, Durham, North Carolina, 27706 USA

Key words: homology; replicator; polymorphism; developmental constraints; biological hierarchy; cladistics; characters.

Abstract

Homology as a topic in phylogenetic analysis has to do with what is conserved in evolution. The problem of homology in systematics – to find homologues, and in so doing, to identify taxa – is distinct from the problem of identifying what kinds of features tend to be conserved, how and why. The two sets of issues are fundamentally interdependent at the point that one selects the appropriate taxonomic units, identifies the characters one wishes to study, or decides what constitutes a single character.

Homology as a phenomenon is a manifestation of replication and of continuity of biological information. Replication occurs at many levels in the biological hierarchy: from the DNA replication that accompanies cell replication, to the replication of gross phenotypic characteristics within individual organisms that results in iterative homologues, to the replication of individuals to form a population that persists (in replication through successive generations) in evolutionary time. In replication, biological information may persist unchanged, or it may be disrupted or transformed. Different patterns of change may be expressed at different levels of the biological hierarchy. Here a concept developed in arguments on levels of selection becomes useful: change at one level of the hierarchy – e.g., genes or gross phenotype – may be *screened off* from changes at other levels. Understanding the manner in which phenotypic features develop or are replicated, the mechanisms of screening off, and the evolutionary origin and transformation of these mechanisms is a major challenge for understanding the biological basis of homology.

The recognition or coding of characters for a phylogenetic analysis calls for decisions on what level of description and how complex a unit character is to be recognized. Here an additional point of comparison within the biological hierarchy – the relationship between organisms and taxa – becomes important. We use

characters of organisms to trace phylogenies of taxa; yet because traits can arise and subsequently become fixed in different segments of a population lineage, phylogenies of organismal characters can conflict with the phylogeny of the taxa that comprise them.

For these reasons, a full understanding of evolutionary changes undergone in lineages will require us to combine phylogenetic analyses with analyses of development, studies of developmental and population genetics, and comparisons of gross phenotype.

Review and introduction

Phylogeny has elements of both continuity and change. We may identify the elements of continuity as homologies; the change is called evolution (Roth, 1988). In Van Valen's words, "homology is resemblance caused by continuity of information" (Van Valen, 1982; cf. Osch, 1973).

Any comparison can be summarized as a partitioning of similarities from differences. For biological entities, what is considered fundamentally the same and is inferred to have been evolutionarily conserved, is homologous.

Change occurs by degree, and "sameness" is relative. By "fundamentally the same" I refer neither to essentialism nor to phenetic similarity, but rather (in accordance, e.g., with Patterson, 1988:605 and Schoch, 1986:131) to "abstracted identity" or "1:1 correspondence" that may trace through intermediate forms.

To be meaningful, a statement about homology must precisely specify which features or aspects of a character have been conserved; alternatively, it must specify a level in the taxonomic hierarchy (Bock, 1977). For example, bird and bat wings are homologous in their possession of an ulna – which is to say, in the features of that bony element necessary for its identification as an ulna. They are not homologous in their possession of a forelimb surface used in flight, because genealogical continuity is absent for this feature. Alternatively stated, bird and bat wing are homologous as tetrapod limbs. Their homology is manifest at the levels of the clade Tetrapoda and all higher (more inclusive) taxa, by virtue of genealogical continuity at these levels. A statement about homology becomes meaningful when its taxonomic level is specified, or when the particular features that have been conserved are identified (Bock, 1977).

The relationship of homology exists for characters within single individual organisms, as well as between them. In the first case, the phenomenon is called iterative homology (Ghiselin, 1976; Roth, 1984). Here, continuity (i.e., homology) is manifest to the extent that two parts of an individual (e.g., two leaves in a plant, or a leaf and a bract; two segments of an annelid; two chains of a single hemoglobin molecule) share a developmental program. The second phenomenon (e.g., the phasmids of two individual nematodes; the asci of individuals in two fungal taxa) I have called phylogenetic homology (Roth, 1984). Here the continuity must be genealogical. Because iterated characters may evolve separately, both phylogenetic and iterative homology exist where iterative characters are genealogically related.

For example, alpha and beta chains of hemoglobin presumably arose within an ancestral organism through gene duplication.

Haszprunar (1989) suggested, in addition to iterative and phylogenetic homology, two additional categories. One is ontogenetic homology, which I take to be a special case of iterative homology: information is expressed repeatedly within single organisms, but with variation through time rather than space. Another is polymorphic homology: different expressions of the same trait within a single population. Polymorphic homologies are traits that are phylogenetically homologous at the level of character, but differ in their character states: Corresponding parts of male and female urogenital tracts in vertebrates are polymorphic homologues, as are alternative alleles at a single locus. For such traits, the element of continuity (homology) is the "sameness" in the role that the trait plays in development, genetic or physiological processes, or anatomy. "Anatomy" for the urogenital example above refers to gross anatomical topographic position within the body; "anatomy" for an allele is its genetic locus or mapping position on the chromosome. Ontogenetic homology could in one sense be considered a case of polymorphic homology existing through time within a single individual.

By examining patterns of character expression in hybrids, McDade (in press) has provided interesting examples of how the biological basis of polymorphic homologues can be studied, how polymorphic homologues can be discovered, and how they can be used in phylogenetic analysis. In a later section of this paper, I will be discussing *polymorphism* – the existence of more than one form within one population – but not the concept of *polymorphic homology* per se.

Comparative biologists sometimes refer to "homologues," phenotypic features (in practice, often, morphological structures) with a recognizable integrity. A homologue manifest in several taxa may vary in form and function, but in overall pattern it will appear also to resist evolutionary change; to exhibit a cohesiveness and an individuality. The relationship between a homologue and all of its various manifestations is the relationship between a character and all of its character states.

Wagner (1989b) has suggested the following definition of homology: "Characters from two individuals or from the same individual are homologous if they share a set of developmental constraints, caused by locally acting self-regulatory mechanisms of organ differentiation, and are thus developmentally individualized parts of the phenotype." Excluded explicitly from this definition are (a) relatively simple characters (or features or aspects of development) that may be phylogenetically retained in a lineage, or iteratively expressed in various forms within a single individual, but which have not accumulated about them the self-regulatory mechanisms of developmental constraints, and (b) more complex structures consisting of more than one developmentally individuated part, which collectively may not behave as an individual.

I believe it may be more useful to apply Wagner's definition not to the relationship of homology *per se*, but to a special subset of all possible entities that can be homologised – those that are individuated. With slight modification, then, Wagner's definition could be restated, "*Individuated homologues* are characters from

two individuals or from the same individual that share developmental constraints, caused by locally acting self-regulatory mechanisms of organ differentiation and are thus developmentally individualized parts of the phenotype."

In a phylogenetic context, the advantage of retaining *all* expressions of continuity of information within the definition of *homology* is that it allows us to consider the entire spectrum of evolutionary change and conservatism. This spectrum ranges from retention, through brief segments of history, of minor variants that are identical by descent, to the "extraordinary conservatism of [some] morphological patterns" that Wagner argues persuasively are of special interest in the understanding of the biological basis of homology. In an iterative context, pleiotropies may occupy the simplest and most trivial extreme in a spectrum of homologies, whereas repeated organs or identical members of a single clone (which are homotypes sensu Owen, 1848; homonomies sensu Riedl, 1978, and Bronn, 1958 [cited by Schmitt, 1989]; or the individuated homologues of Wagner's definition as modified), exemplify products of more complex developmental systems.

Constraints can be absolute or partial. Developmental constraints, and the establishment for or by characters of a cohesive identity or individuality (see Wagner, 1989a), are matters of degree. The extremes in this spectrum (from the retention of minor variants to the retention of major phenotypic patterns) differ clearly, but I see no sharp, objective point in the continuum at which one is compelled to distinguish between relatively minor characters that happen to be evolutionarily conserved, and conservative systems of greater complexity, greater canalization, and greater resistance to evolutionary modification. A similar argument can be made for iterative characters, which include simple features (pigment spots, for example) that can occur more than once in a single organism, on upward in complexity to entire organs or organisms that are duplicated wholesale within (respectively) an organism or clonal colony. Because this grey zone of transition exists, it would seem preferable to have it encompass the distinction between individuated and other homologues than the definition of the concept of homology itself. The concept of homology has grown to embrace a wide range of phenomena that vary in their complexity from the homology of base pairs and loci, through the homology of developmental processes and the homology of entire organs and organs systems. The conceptual and biological relationships among these organizational grades of homology is something we may profitably explore.

I have just argued that homologies vary along a spectrum of complexity. Yet within this spectrum we are confronted with a major challenge. As Wagner (1989a) and others (e.g. de Beer, 1971; Sander, 1983, 1989; Roth, 1988) point out, our conclusions about homology and the nature of evolutionary change and evolutionary conservatism seem at times paradoxically to differ according to the level of description, be it genetic or gross phenotypic (for examples see below). In this paper, I will first briefly describe two prevailing approaches to the study of homology, and second, explore the influence on each of these views of the hierarchical level of analysis.

Two views of homology

Homology can be viewed as a single concept that has been approached from two directions. The dichotomy of approaches has been observed by many authors and described in various ways. Some differences between the two approaches are listed in Table 1.

To the extent that the main objective of systematics is the reconstruction of phylogeny, we may call one of the columns in Table 1 the approach of the systematist. This approach is "taxic" in Eldredge's (1979) sense, because its focus is on taxonomic groups. A central problem in systematics is to find homologous characters, and in so doing, to identify monophyletic taxa. A character such as, for example, production of flowers, is homologous among the species in which it is found, and diagnoses a monophyletic group known as the angiosperms.

Table 1. Two approaches to the study of homology

	Homology in systematics	The biological basis of homology
objects of focal interest	taxonomic groups & taxonomic characters	transformation or conservatism of characters in lineages
relevant types of homology	phylogenetic	phylogenetic, iterative
objectives	to find homologous characters to identify taxa	to discern what types of phenotypic features of organisms tend to be conserved, why they are, and how
conceptual issues addressed (some examples)	definition of criteria & construction of tests for homology	explication of patterns of conservatism in phenotypic characters
empirical undertakings	application of criteria and tests to identify homologies & taxa	identification and comparison of developmental patterns, processes, interactions, & pathways
methods & tools	cladistic, evolutionary, or phenetic methods and their associated algorithms (for constructing e.g., Wagner networks & trees)	those of comparative and developmental biology and genetics; phylogenetic trees
product	patterns and distributions of characters; arangements of taxa	clarification of similarities & differences in the developmental processes that produce phenotypic features

The focus for systematics is upon phylogenetic homologues[1] (sensu Roth, 1984, 1988; see Patterson, 1982, 1988; Stevens, 1984). Wagner (1989b) has suggested that phylogenetic inference even employs a distinct concept of homology, which he identifies as the "historical homology concept". Some conceptual problems in systematics have been to define criteria or tests for homology, such as those of Remane (1952) or Patterson (1982). The practical problems in systematics, then, are the application of these tests; the methods are those of one's particular school (e.g. evolutionary or cladistic), and the tools are the specific algorithms (including, for example, parsimony, compatibility, and maximum-likelihood methods). The products of systematic work are (1) patterns and distributions of characters (or hypotheses of homology), and (2) arrangements of taxa (branched diagrams or hierarchical listings).

A different approach, which I will call the "biological"[2] approach to homology, is in part[3] "transformationist" in its view (Eldredge, 1979) because it examines change and conservatism. This approach, according to Wagner (1989b), applies a "biological homology concept", and examines what kinds of features are conserved in evolution, and how and why. What causes structures to be preserved through evolutionary time is not only normalizing selection or lack of directional selection, but also what has been identified in other contexts as "developmental constraints". Wagner (1989a) suggested that generative rules of pattern formation and ontogenetic networks (both hierarchical and cyclic) can produce "epigenetic traps", which "restrain what genetic variation can do to the phenotype" (Wagner 1989a). The "burden" (Riedl, 1978) or "generative entrenchment" (Wimsatt and Schank, 1988) of a character is a measure of its developmental, functional, and ultimately evolutionary interdependence on other characters. For example, an organ such as the vertebrate eye will form into a structure of a characteristic pattern under a variety of stimuli, as long as the cyclic feedback between lens and optic cup is preserved; the developing tissues are interdependent, and to the extent that mutations are expressed, the expression tends to be global (e.g., failure to develop at all) rather than localized.

The methods for studying the biological basis of homology are those of comparative and developmental biology, and perhaps genetics. The product of such study is a clarification of the behavior of particular developmental systems, and an enhanced understanding of the interrelationship of variation in genotypes, developmental processes, and phenotypes.

---

[1] To be sure, iterated characters – numbers of petals; types of scutes – often contain phylogenetic information, but they do so as phylogenetic homologues: the number (or form, or what-have-you) is what is compared in different taxa. Iterative homology is a relationship of characters within single organisms.

[2] Obviously, systematics is a branch of biology; I use "biological" here simply as shorthand for "concerning biological processes such as physiology and development".

[3] in part, but not entirely: in Eldredge's (1979) original usage, transformationist approaches were defined as narrower in perspective than, and a subset of, the taxic approach. The distinction I make here is between approaches that are equivalent to each other in status. The two approaches simply choose different facets to focus on as problems, and begin with different sets of assumptions about what can be assumed to be true or sufficiently well understood.

These two approaches to the study of homology are two sides of a coin, and they differ. The objectives are separate, and so is the material that each must take for its assumptions. Recognizing homologous characters, and understanding their biological basis, have long been acknowledged to be distinct issues (e.g. by Simpson, 1975 and Patterson, 1988, but apparently not by Schoch, 1986). One involves application of a concept; the other its explication (and both are explanations, but of different types; Riedl, 1989). We must be reminded, however, that the two approaches intersect and are interdependent. The biological basis of homology can only be examined if we know the phylogenetic pattern on which evolutionary processes can be traced. That is a product of systematics.

The systematist, in turn, whether consciously choosing to or not, always makes an implicit assumption or decision about the biological basis of homology whenever he or she identifies a character. A character is a unit that is counted or listed in deriving a phylogeny. The act of delimiting or delineating characters is automatically and intrinsically a decision of what constitutes an evolutionary step.

A major challenge in systematic work is to specify precisely what has *changed* at particular internodes within a lineage; in other words, to identify the synapomorphies that define each clade[4]. Subdivision of more complex characters into larger numbers of relatively small differences is what can provide the greatest resolution in a phylogeny (as long as characters remain operational and homoplasy is minimal).

By contrast, a problem of major importance for understanding the biological basis of homology is evolutionary *conservatism*. Small variations on major themes are "irrelevant" to this undertaking (Wagner, 1989a), whereas what would be considered massive symplesiomorphy and comparatively uninformative in a cladistic context is here the focus of interest.

Perhaps this difference in emphasis is why the recent conceptual literature tends to treat the biological basis of homology (e.g., de Beer, 1971; Ghiselin, 1976; Van Valen, 1982; Roth, 1982, 1988; Wagner, 1988a, b) separately from homology as a concept used in systematics (e.g., Patterson, 1982, 1988; Stevens, 1984), and why Wagner (1989b) has suggested that what he terms historical and biological concepts of homology are not the same.

If homology is regarded simply as a manifestation of continuity or identity of information, however, the two concepts merge into one – though we may choose to examine it from either of two vantages. The interdependence of biological and systematic approaches to the study of homology suggests that we may profitably explore their common ground.

---

[4] I suggest this is where the emphasis may be in systematics because, as a gross generalization, comparative anatomy has already revealed a large share of the homologies at the most general levels (i.e., symplesiomorphies), and the greatest volume of work remains in resolving relationships within well-established taxonomic groups. Systematics at even the highest levels (Lipscomb, 1985; Field et al., 1988; Sogin et al., 1989) works with very generally-distributed, well-recognized symplesiomorphies, such as components of ribosomes or other cellular organelles, and is a matter of finding character states that diagnose subgroups.

Homology and biological hierarchies: Incongruities

An important complication for both approaches to the study of homology is the fact that there is no simple hierarchical relationship between characteristics of organisms examined at different levels of description, and no one-to-one correspondence between changes (or differences) in genotype and phenotype.

Small genetic changes can result in large phenotypic differences, and constancy in phenotype may belie underlying major diversity of genetic and developmental processes. As de Beer (1971) succinctly remarked, "homology of phenotypes does not imply similarity of genotypes": a structure can retain its integrity and individuality with historical continuity despite fundamental changes in its developmental and genetic basis.

This issue was the subject of an earlier discussion (Roth, 1988), but it may be useful to review briefly some examples here. What I have called "genetic piracy" – the change (at a gross phenotypic level) in the field of expression of particular genes – is nicely illustrated by an example described by de Beer (1971), in which selection for modifier genes can restore eyes in *eyeless* mutants of *Drosophila*. At the level of gross phenotype, the eyes of the wild type and the selection-modified mutant would ordinarily be considered homologous, and yet they differ in the genetic basis of their development. Selection on the mutant has deputized modifier genes to participate in eye formation, compensating for dysfunction at the *eyeless* locus. Different suites of genes are responsible for the appearance of eyes in the differing genetic contexts of the mutant and the wild-type.

Developmental processes and precursors can also change without disrupting phylogenetic continuity at the level of gross phenotype: the intercalar bone of *Amia* and teleosts, though it ossifies in membrane without a cartilage primordium, is nevertheless considered endoskeletal, not dermal, and judged homologous with its namesake in other fishes (Patterson, 1977:87).

Such incongruites arise in part because of feedback loops, many-to-one and one-to-many relationships in the mapping of genotype to phenotype. Genetics, development, and gross phenotype do not, as a result, stand in simple, linearly hierarchical relationship to one another: identity (or phylogenetic continuity) at one level may fail to reflect the changes that occur at another. When the patterns we observe at different levels of analysis conflict, then, where should we focus attention in identifying homologies?

Homology and biology: the role of genetics

The concept of homology originated within the domain of comparative anatomy, and I have argued in the past (Roth, 1984:18) that it should remain there: the concept was in use and useful before biologists had any accurate notion of the nature of genetics. Can we ignore genetics?

I would now argue no. For a full understanding of the biological basis of homology, one needs to understand the genetic basis of homologues, for several reasons.

First, it is not always possible to identify parts of the phenotype that are developmentally *fully* individualized (Wagner, 1989a, b). Selection pressures may at times favor pleiotropies that respect functional and organizational boundaries within organisms (Riedl, 1978, 1982), but numerous conterexamples exist. Neatly nested hierarchies of phenotypic characters within single organisms are not the inviolable rule. "A single structure or set of structures may be affected by several growth fields," (Van Valen, 1962) and not all of these will be congruent. The problem is one of uncorrelated, shifting, and overlapping developmental fields, or fields of genetic influence.

For example, in mammalian skulls, the individuality of single bones is sufficiently clear that phylogenetic homologies with other vertebrates are well-established. Yet intraspecific comparisons of variability in skull measurements (Roth and Thorington, unpublished data) suggest that the shape of the skull as a unit is developmentally constrained to as great or a greater extent than are the size or shape of individual bones – even when allomery (Soulé, 1982) is taken into account. The positioning of boundaries between individual bones (i.e., sutures) can be quite variable at times when the overall shape of the skull (e.g., a breadth measurement that spans portions of two different bones, its endpoints determined by the curvature of the skull and bearing no consistent relationship to any named anatomical landmarks) retains important phylogenetic information. When pieces of bone are excised in a region near a suture, bone growth will restore the shape of the skull, but which of the surrounding bones contributes most can be quite variable, and appears to be a function of minor local growth processes (Watzek et al., 1982). Thus, even at the level of gross phenotype, the individualized units we consider homologues may not be separable, and may overlap or intersect one another (for additional examples, see Olson and Miller, 1958). Self-regulatory units do not necessarily occur as uniquely identifiable, neatly-nested sets.

For this reason, I believe it is of interest not only to identify entire structures (individuated homologues) that exhibit developmental constraints, but also (or alternatively) to homologise phenotypic features or aspects of development, down to and including even those for which single loci are responsible.

A second reason I would suggest that genetics is important is that homology is a manifestation of continuity of information, and this information is largely, though not exclusively, genetic in nature. To put it another way, if we are interested in continuity, we must be concerned about replication. A replicator, in the terms of Richard Dawkins (1978), is "any entity in the universe which interacts with its world, including other replicators, in such a way that copies of itself are made". Genes – genetic material, nucleotides, parts of chromosomes – are ultimately the most direct replicators in biological systems, and this is the source of their importance.

Yet the ability of genes in and of themselves to replicate is limited. Replication (at least within the last several billion or so years, since the time of the primordial soup) can take place only within the context of a cell. Genes require enzymes, cytoplasm, energy: if the cell dies, they cease to replicate (or, if the genes happen to be viral, they must find another cell). For this reason, even what we call genetic

information cannot persist, cannot continue, cannot replicate, except in an *epi*genetic context. This is the context in which genes interact, in which they are involved in building cells and entities of even greater complexity, such as organisms.

We can extend this line of reasoning in observing that, for certain types of living things (what we might call the most highly individuated types, sensu Buss, 1987), cell replication only occurs within organisms (and even then, only within an appropriate external environment). The cells of most metazoans, for example, ordinarily persist and replicate only as parts of an organism. Many cnidarians, flatworms, and sponges are capable of full regeneration and propagation from small bits of tissue, but even here the requisite unit for continued replication consists of more than a single cell. The pervasive exception to this generalization for metazoans is a special type of cell called a zygote: zygotes are capable of producing entire new individual organisms and new cell lineages. Even so, zygotes do not themselves produce new zygotes: they must produce organisms that do that.[5]

Hull (1981 : 151) has pointed out that entities more inclusive than single genes or genomes can function as replicators, and that "organisms themselves might be viewed as replicators, the only difference being in how direct the mechanism of replication happens to be". Regardless of how direct we may perceive *genic* replication to be, however, such replication can be (and I have suggested above that it *is* for many metazoans) highly contingent on the replication of the the more inclusive entities such as cells and organisms. In our consideration of homology and continuity of information (and the embodiment of that information in biological structure), it is therefore essential to construct and consider the entire hierarchy of possible replicators.

But which level (gene, genome, cell, organism, etc.) of replication is most important? If conservatism of gross phenotype can belie large differences in genetic basis or developmental mechanism, and if information is structured and organised in topologically incongruent ways at different levels, as the examples cited above, by de Beer (1971), and by Roth (1988) suggest it often is, then how does one decide at which level homology resides?

An analogous question has arisen for the concept of natural selection, a concept that recent discussions of replicators (e.g. Hull 1980, 1981; Brandon 1990) have been directed at clarifying and extending. Logic similar to that applied to the problem of levels of selection may be applied to the problem of levels of replication (or homology):

In discussing levels (e.g., genic, organismic, group-, or species-, etc.) of selection, it has been useful to introduce the concept of an interactor. Hull (1980 : 318) defined an interactor as "an entity that directly interacts as a cohesive whole with its environment in such a way that replication is differential". A key feature of the selection process, which distinguishes it from the process of simple replication, is its *differential* nature.

---

[5] There are plenty of plants and fungi for which a single somatic cell suffices to reproduce entire organisms; my point here is that there exist some taxa for which the requisite unit for replication is more complex.

Yet interaction might be considered in a more general way. Not only natural selection, but any specified process could involve interaction, including the process of replication itself. Biological replication depends upon a set of physical interactions that are mediated by enzymes, and made possible by cellular sources of materials and energy.

To identify the level (A or B) at which the interactive process of natural selection occurs for any specified example, Brandon (1990) employs Salmon's (1971) notion of *screening off*. If A and B are putative causes, at two different levels, of effect E (or, stated differently, if they are putative participants in an interaction that results in E), we say that A screens off B from E, if A renders B statistically irrelevant with respect to outcome E, but not vice versa. In symbolic terms, $P(E, A^* B) = P(E, A) \neq P(E, B)$, where "$P(E, A^* B)$" is "the probability of E given A and B". A is then considered a better causal explainer of E than is B. Proximate causes (A), for example, screen off remote causes (B) from their effects. Brandon used this idea of screening off to argue that "in standard cases of organismic selection the mechanism of selection, the differential reproduction of organisms, is best explained in terms of differences in organismic phenotypes, because phenotypes screen off both genotypes and genes from the reproductive success of organisms." If, for example, we have a selective scenario in which tall organisms outreproduce shorter ones, the reproductive success of a gene that increases height will be best explained in terms of the height of the organism that houses it: "manipulating the phenotype [e.g., feeding the organism to make it grow taller] without changing any aspect of the genotype can affect reproductive success (castration is only the most obvious example) . . .[but] tampering with the genotype without changing any aspect of the phenotype cannot affect reproductive success." Here, the process of selection is acting at the level of the phenotype, because phenotype screens off genotype.

We can apply similar reasoning to identify levels at which replication occurs in homologues. The examples cited in de Beer (1971) and Roth (1988) indicate that the genes that participate and the processes that are involved in the development of homologues can evolve and change in important ways without affecting gross morphology. Patterns of gross morphology persist and have over evolutionary time been replicated with fidelity, where fidelity may be lacking at the level of genes. In these cases, we infer a continuity of information, homology, or process of replication, at the level of gross phenotype, but observe evolutionary *change* at the other levels. The information in these examples is passed along not in the form of individual alleles, but presumably in the form of entire systems of interacting genes.

Where such examples have arisen, we infer the presence of developmental constraints (Wagner 1989a): systems of interaction have evolved that tend to *screen off* the effects of single alleles, or alterations of developmental processes. The information does not so much reside within individual codons as emerge from the interaction of their products. Wagner (1989a) has identified some types of developmental mechanisms that have this screening-off effect (though he did not refer to them in these terms): they include epigenetic mechanisms such as the generative rules of pattern formation, and cyclic and hierarchical networks. Understanding the origin, establishment, and mechanisms leading to the refinement of developmental

processes at higher levels that can screen off genetic variation is one task that lies before us in studying the biological basis of homology. This understanding has already begun to advance with the work of Riedl (1978), Kauffman (1986), Buss (1987), Bonner (1974), Wagner (1989a, b), Raff and Kaufman (1983), and others. Like selection, which can potentially operate on any of several levels in the hierarchy of interactors (Brandon, 1990), homology may pertain to any of these levels of replication.

In general, the unresolved questions pertaining to the biological basis of homology include: what are the properties of some developmental systems that allow their preservation or replication; what are the rules – if any general ones exist – of mapping from genotype to phenotype, and how common are different types of evolutionary change in developmental processes. An understanding of iterative homologues (as natural experiments in development) may prove especially revealing.

Genetics and homology in systematics

What problems does the incongruence between genotype and phenotype cause for systematics?.

Delineation of characters

Systematic methods require work with characters and character states. Yet evolutionary change does not *occur* in discrete evolutionary events. Evolutionary events are defined by the systematist, in the context of a particular analysis. For the purpose of reconstructing phylogeny, partitioning the phenotype of an organism into characters and partitioning evolutionary history into a series of events are useful (probably essential) exercises. For a given example, some partitionings may be inappropriate, in that they yield incorrect inferences about phylogeny. But there is generally not a unique solution to the problem of delineating appropriate characters and events.

Imagine, for example, an evolutionary transformation of a structure from "small and round" to "large and square". Whether we consider this transformation as one, two (e.g., small → large; round → square), four (e.g., small → intermediate → large; round → intermediate → square), or any other number of events is of importance *only* in the following instances: (a) if we are interested in history, or change through time, and what happened during the time interval between two steps is of interest; (b) if the lineage has undergone a split between two events, so that the evolutionary change exists in some taxon in only partial form; or (c) on practical grounds, if calling the change two, as opposed to one event, happens, by chance, to add some weight to the true phylogeny, when we otherwise are at risk of being misled by homoplasy. (Clearly, this last case has important consequences, but we are in general practice not in a position to evaluate them, since true phylogenies are never directly revealed to us.)

One difficulty for the identification of unit characters is that it depends upon the level of description. Genealogical continuity (or accurate replication of a pheno-

typic feature) can exist, despite changes in the number and identities of genetic loci that participate in development. In other circumstances, small changes in genotype can precipitate major phenotypic discontinuities. A unitary evolutionary change at one level of description does not necessarily correspond to a unit measurable at any others.

But let us examine the opposite point of view: let us try to atomize. Because phenotypic variation is continuous for some phylogenetically interesting characters, and because different phenotypic characters must be measured in qualitatively different units (e.g., dimensionless counts, units of length, units of complexity, units of frequency, etc.), and because phenotypically disparate characteristics can be pleiotropically linked, it is difficult *a priori* to define what constitutes a discrete evolutionary event in phenotypic terms.

Perhaps, then, the fundamental level of analysis should be genetic. If unitary evolutionary events exist, mutations are logical candidates. Are mutations the units of evolutionary change a systematist might seek?

I should stress that this question is conceptual and ontological, not practical. The issue is not, for example, whether molecular systematics is possible, or useful; clearly it can be. But the act of counting mutations in and of itself tells us nothing more than how many mutations have occurred. It is not the same as determining when particular taxa diverged (though it may provide clues to it). It is not the same as tracing the evolution of phenotypic characters (though it may correlate with that as well). Without making important additional suppositions and inferences about accumulation of change that is measured in other types of units – units of time, or units of phenotypic difference – counting of mutations becomes an empty exercise, or simply an act of bookkeeping, until it can be related to more interesting issues. These issues, timing in evolution and the nature of phenotypic characters, are what place organisms in an interactive context in which we can think about processes such as selection and constraint.

Ultimately, a central objective in systematics is to reconstruct phylogeny. In identifying differences and counting characters shared by taxa, one is addressing the question of the probability of an interconnected, non-quantized mass of differences arising by one phylogenetic pattern or another. Converting this question to an exercise in *counting* simply provides a way of ranking probable alternatives. To do this, we think in terms of *numbers* of evolutionary events. But evolution itself is not quantized.

Unit characters may have no ontological reality, but in practice, as systematists, we define and work with characters, and on the whole the practice has yielded useful and consistent results in the form of impressive congruence in many phylogenetic hypotheses, despite the differences in the techniques applied and the researchers who apply them (see Nijhout et al., 1987; Archie, 1989). However, considering mutations as the ultimate unit characters magnifies another problem.

Phylogenies of characters vs phylogeny of taxa

Mutations are characters of single organisms and their descendants, and not necessarily of all individuals within entire populations. The problem polymorphism

presents for systematics is extreme for mutations: whose genomes are to be compared?

All mutations – virtually all characters – arise within individual organisms. *A major challenge that analyses at the level of genetics present for systematics is to consider at what point a new characteristic comes to characterize an entire taxonomic group – and what groups are relevant.* This is not just the empirical problem of the study of the process of speciation; it is also the problem of translating population concepts into species concepts. And these issues, well apart from discussions of homology, have a philosophy, a literature, and arguments of their own (Mishler and Brandon, 1987; Otte and Endler, 1989).

Potentially most troublesome for the reconstruction of phylogeny, however, are those features (characters, at any level of description) of individual organism that do *not* come to characterize entire taxa: situations in which phylogenies of characters differ from the phylogeny of taxa (e.g. Throckmorton, 1962; Tajima, 1983; Pamilo and Nei, 1988; Bonhomme et al., 1989). The problem is especially acute where polymorphisms arising in an ancestral population persist and are retained by daughter species.

Imagine an apomorphic character x′ arising in some member of a sexual population before a speciation event (Fig. 1). Imagine that the character arises sufficiently near the geographic boundary between the daughter species that descendants of the original x′ individual are separated into both of the new daughter species: both daughter species comprise some individuals with x (the primitive character state) and some individuals with x′. Imagine then that subsequent speciation (involving certain patterns of selection or drift) in both lineages separates off distinct species consisting only of x′ individuals. The result is four "granddaughter species": two pairs of sister taxa, each pair comprising one species fixed for x′, and one polymorphic or fixed for x. A phylogenetic analysis (using additional characters) of such a system could lead us to one of three conclusions: (1) We could derive a phylogenetic tree that was congruent with the distribution of character x′. Such a tree would be at odds with the phylogenetic history of the populations as we have described them. (2) On the basis of other characters, we could reconstruct the true phylogeny of the populations. In this case we would call x′ a homoplasy – *despite the fact that for all individuals in which it ever occurs it is identical by descent.*

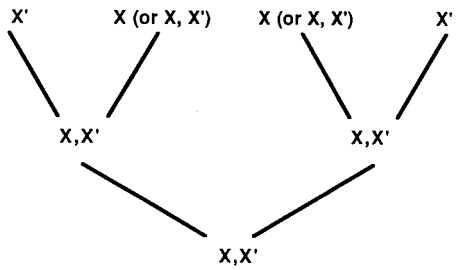


Fig. 1. Hypothetical phylogeny of a bifurcating polymorphic population.

(3) We could derive a phylogenetic tree that was not fully consistent with either scenario 1 or scenario 2.

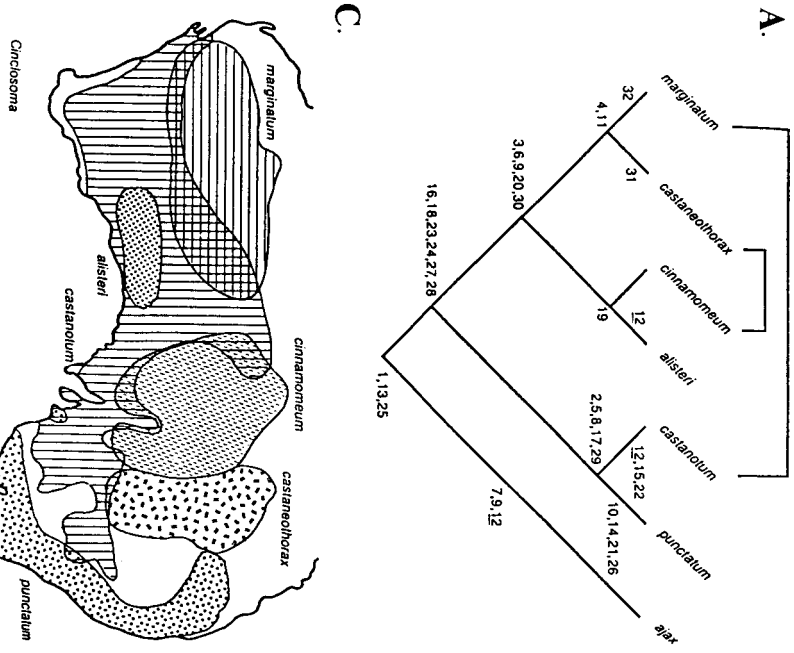
Clearly, then, where ancestral polymorphisms can persist, the true phylogeny of a character will not necessarily match the true phylogeny of the populations in which it resides. If we imagine such a situation (the retention of ancestral polymorphisms) arising not just in one character, but in several, at various times and in various parts of an ancestral population's geographic range, the possibility of reconstructing phylogeny might be seriously compromised.

A key point in the example described above is that the *origin* of character x′ occurs in a segment of the lineage distinct from its *fixation*. In contrast (especially with morphological data), cladistic methods usually represent origin and fixation as a single event. Our error in coding character x′ stems from failure to recognize that, despite its unique origin, *fixation* of x′ within the granddaughter species occurred in parallel. *The phylogenies we trace are those of taxa, but the characters we work with are generally those of individual organisms*: x′ may be a character of an organism, but "fixation of x′" is a character at the level of a population. Systematists working with molecular data, confronted frequently with polymorphism in extant populations, have discussed its implications for phylogenetic reconstruction (e.g., Felsenstein, 1981; Buth, 1984; Mickevich and Mitter, 1981, 1983; Avise, 1989; Swofford and Berlocher, 1987), but the problem may be more pervasive than is generally acknowledged (Arnold, 1982: 17): Regardless of the type of character (morphological or chemical), or whether polymorphisms are evident in today's populations or not, *all* characters *originate* as polymorphisms within populations, except where populations are founded by single organisms. We will in general have no information about the point in history at which fixation occurs – only that it has (in a currently monomorphic species)or has not (in a species that is polymorphic at the time it is sampled).

A cladogram recently presented by Cracraft (1989) can serve as an example of how considering the possibility of retention of ancestral polymorphisms through more than one bifurcation of a lineage can dramatically alter our view of phylogeny. I present this example not as a challenge to Cracraft's analysis, but rather to highlight the distinction between phylogenies of characters and phylogenies of populations, and as an illustration of how more than one reasonable scenario can be invoked to explain the same data.

Cracraft presented the cladogram shown in Fig. 2A as the most parsimonious interpretation of the data matrix in Fig. 2B for Australian birds (genus *Cinclosoma*) with the geographic distribution shown in Fig. 2C. A scenario (omitting the outgroup *C. ajax*) that includes the possibility of polymorphic lineages and that would produce the same distribution of characters as Fig. 2B is shown in Fig. 3A (which will be explained in some detail below). The genealogy represented here is summarized in Fig. 3B. The diagram in Fig. 3B is like a cladogram in the sense that it depicts the temporal sequence of bifurcations in the lineage, but is not a cladogram in that it was not generated using cladistic methods.

(Interestingly, but perhaps only coincidentally, this population phylogeny is more consistant with the geographical distributions shown in Fig. 2C than is the



Distribution and phylogenetic hypothesis for the phylogenetic species of Australian quail-thrushes (*Cinclosoma*). An analysis of 32 ordered characters (Table 2) produced a best-fit tree of 35 steps (consistency index = 0.914). The tree was rooted using the outgroup taxa, *Ptilorrhoa castanota* and *P. leucosticta*. Underlined characters identify parallelisms and brackets link taxa that are known to hybridize. Distributions are after Ford (1983).

Character-state data for the genus *Cinclosoma*.[a]

	Characters																													
Taxa	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25	26	27	28	29	30	31	32
P. leucosticta	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
P. castanota	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
C. ajax	1	0	0	0	0	0	1	0	1	0	0	1	1	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0
C. punctatum	1	1	0	0	1	0	0	1	0	1	0	0	1	1	0	1	1	1	0	0	1	0	1	1	1	1	1	1	1	0	0	0
C. castanotum	1	1	0	0	1	0	0	1	0	0	0	1	1	0	1	1	1	1	0	0	0	0	1	1	1	0	1	1	1	0	0	0
C. castaneothorax	1	0	1	1	0	1	0	0	1	0	1	0	1	0	0	1	0	1	0	1	0	1	1	1	1	1	1	1	0	1	1	0
C. cinnamomeum	1	0	1	0	0	1	0	0	1	0	0	0	1	0	0	1	0	1	1	1	0	0	1	1	1	0	1	1	0	1	0	0
C. marginatum	1	0	1	1	0	1	0	0	1	0	1	0	1	0	0	1	0	1	0	1	0	0	1	1	1	0	1	1	0	1	0	1
C. alisteri	1	0	1	0	0	1	0	0	1	0	0	1	1	0	0	1	0	1	1	1	0	0	1	1	1	0	1	1	0	1	0	0

[a]Outgroups include *Ptilorrhoa castanota*, *P. leucosticta*, and C. ajax, all distributed in New Guinea. Key for characters (0, primitive and absent; 1, derived and present, in all cases): 1, male throat blue-black; 2, female throat gray; 3, female throat buff or cream; 5, female breast solid gray; 6, female breast light brown to brown; 7, female breast reddish chestnut; 8, feathers at sides of breast and upper throat solid gray; 9, feathers at sides of breast brown to rufous; 10, male breast solid gray; 11 male breast with extensive chestnut or rust-red patch; 12, male breast extensively blue-black; 13, male flanks with spotting; 14, female flanks with spotting; 15, male flanks gray-brown; 16, male with white spots on lesser primary coverts; 17, crown and forehead gray; 18, crown light brown; 19, crown light cinnamon or rufous cinnamon; 20 upperparts cinnamon to light rufous cinnamon; 21, upperparts heavily streaked; 22, male back and rump deep rufous; 23, primaries light brown; 24, tertials rufous to cinnamon with dark central streak; 25, male white malar streak not extending onto throat; 26, male malar streak reduced anteriorly; 27, male light eye stripe; 28, relative bill size decidedly reduced; 29, ear coverts gray-brown; 30, ear coverts rufous or cinnamon; 31, male upper breast rich rust-red; 32, male upper breast pale chestnut.

Fig. 2. Figure and table reprinted (with permission) from Cracraft (1989).

cladogram in Fig. 2A, in that sister species often have adjacent, contiguous geographic ranges. It may also be more consistent with the pattern of interspecific hybridization.)

In rendering Fig. 3A, to improve the clarity of an already-complex diagram, I employed a few conventions. (1) For simplicity of representation, groups of characters that travel as a block are represented by single marks. Superficially, this simplification may seem extreme. The difference, for example, between the white and filled circles of Fig. 3A – which can be identified in Fig. 2A as the combination

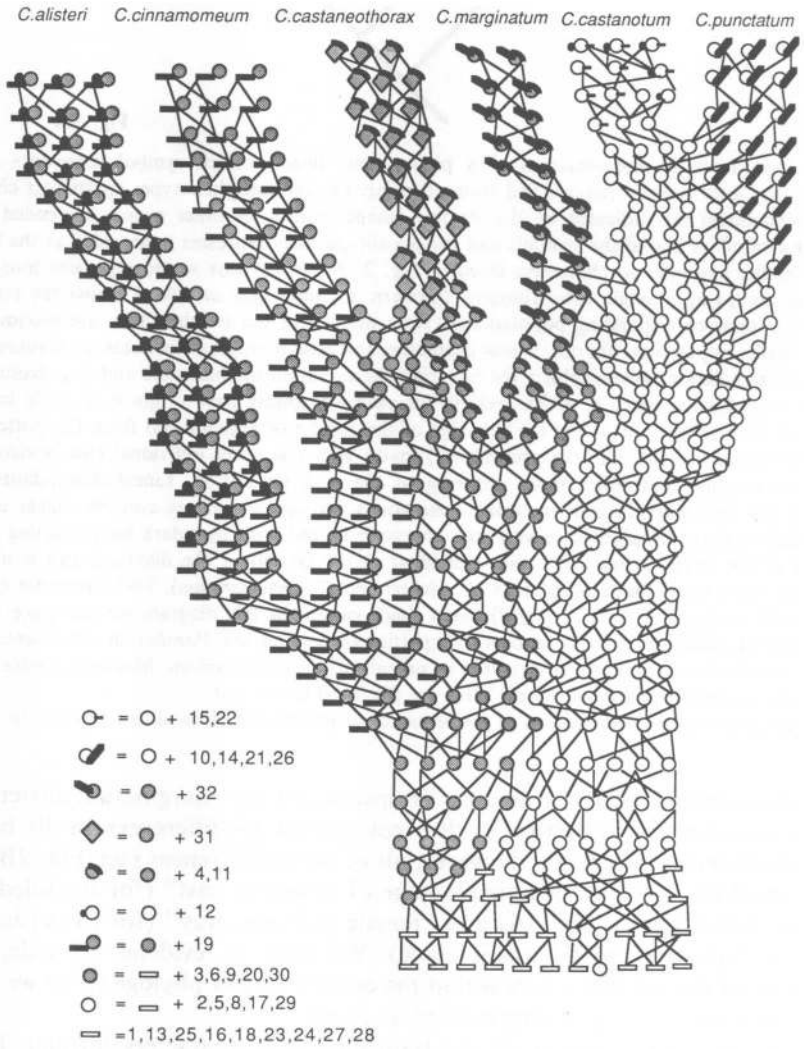


Fig. 3A.

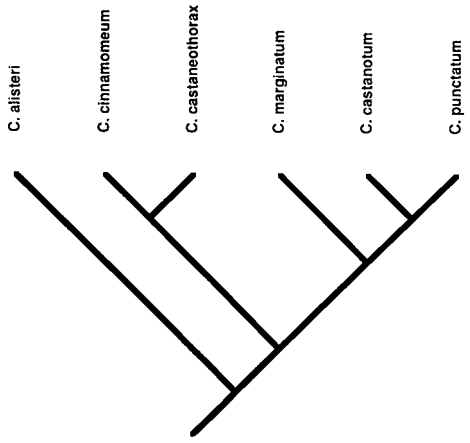


Fig. 3B.

Fig. 3A. Diagrammatic representation of a phylogenetic history. Each symbol represents a single organism, and lines connect parents and their offspring. Organismal phenotypes (particular character-state combinations) are indicated by the shading, shape, marks, or other gizmos appended to that particular symbol. A key to the symbols and the phenotypes they represent is provided at the left, and numbers in the key refer to characters listed in Fig. 2. A vertical axis would represent time, with a generation occupying a single row. Terminal (modern, existing) taxa are listed across the top of the figure above their corresponding populations. (Note that all of the terminal taxa are monomorphic, although earlier populations are not. If new characters first arise in single individuals, they automatically render their population polymorphic.) We can trace the evolution of characters and populations in the following way. Characters 3, 6, 9, 20, and 30, for example, jointly have origin in a single individual represented by a gray circle at the far left edge of the third row (generation) from the bottom. This individual (which we will call "the progenitor") mates with one other individual (the horizontal bar adjacent to it) and contributes three offspring to the next generation. Lineal descendants of the progenitor can be traced through successive generations and appear as grey symbols higher up in the figure. Additional apomorphies arise with time: character 19, indicated by a dark bar projecting from the lower left of the circle, arises in another individual at the far left of the diagram, and is ultimately transmitted into two of the terminal taxa (*C. alisteri* and *C. cinnamomeum*). Each character except 12 (which arises twice, as it does in Fig. 2) has a single origin. In this diagram we can trace both the transmission of characters and the sequence of splitting of populations. Population cohesiveness due to gene flow is reflected in the meshwork created by parent-offspring connections. Making a legible diagram required certain simplifying conventions; these are discussed in the text.

Fig. 3B. Diagrammatic representation of the sequence of population bifurcations depicted in Fig. 3A.

of synapomorphies that characterize (respectively) the marginatum-alisteri clade and the castanotum-punctatum clade – correspond to differences in 10 binarily-coded characters. Collectively, however, all of these differences (see Fig. 2B) could be expressed concisely as "female plumage of brownish cast" (for the filled circles and their derivatives in Fig. 3A) and "female plumage grey" (for the open circles and their derivatives in the other clade). We have no evidence of independent assortment of the ten characters within the context of this phylogeny, so we may as readily represent them as a single event as as ten.

(2) To make visual tracing of the transmission of characters through Fig. 3A relatively easy, I assumed full penetrance of all apomorphies (so, the offspring of a

cross between an individual with any given apomorphy and one without will always have the apomorphy). Males and females alternate across the page, so crosses are possible only between adjacent individuals or individuals with an even number intervening. Although the diagram could not be realistic in all details, the meshwork conveys a subjective impression of the coherence (or "integration" in the terms of Mishler and Brandon, 1987) that a lineage manifests through its genealogical interrelationships.

(Fig. 3A is a heuristic device for showing some key features of the phylogeny. In a real situation, crosses between heterozygotes for the character would eventually, through sexual recombination, reconstitute the primitive condition in some descendants, so regions of the diagram shown here as filled purely with apomorphs would not be so uniform. But this simplified representation does not affect the key features this diagram is intended to represent, and should not bias interpretation. A mechanism like drift or selection must be assumed to eliminate descendants that retain primitive states. But such a mechanism must be assumed to exist regardless of the pathway to fixation; invoking it here does not constitute special pleading. My objective is not to depict accurately all features of an evolving diploid population – that would require larger population sizes, more generations, explicit assumptions about selective value and gene flow, and a closer match to the actual breeding systems of these birds. That could be worth doing as a computer simulation [see, e.g., Neigel and Avise, 1986 for models of maternally-transmitted characters].)

In producing this diagram, I assumed that each apomorphic character except #12 had a single origin within the clade. In this sense, Fig. 3 requires no more ad hoc hypotheses than the analysis that produced Fig. 2. Apomorphies originate in Fig. 3A in the same temporal sequence as, and with a topology congruent to, that in Fig. 2A; the genealogies of characters illustrated in these two figures are identical. However, the genealogy of *populations* represented by Figs. 3A and B is different, and topologically incongruent with the genealogy of characters. Where situations like that shown in Fig. 3 arise in real life, character data will not reflect the history of populations.

It might be argued that the pattern in Fig. 3 would only arise given certain temporal relationships between the bifurcation of lineages, the origination of characters, and their fixation, and that this scenario may be too constrained in its incorporation of assumptions to arouse any general concern about the use of character data in reconstructing the histories of populations. Granted, other patterns could easily be generated, by assuming other sets of temporal relationships. Yet if one does use character data to reconstruct population phylogenies, one must implicitly incorporate similar types of assumptions: namely, that in sexual taxa, new characters, which arise initially as polymorphisms, only reach fixation in single lineages (despite the mechanistic simplicity – illustrated several times within Fig. 3A – of other patterns); or, at least, that misleading patterns of fixation will be few relative to "well-behaved" ones, and will not together cohesively argue for the wrong phylogenetic hypothesis. A paraphyletic pattern (of which there are several shown in Fig. 3) does require us to imagine fixation occurring in parallel in more than one lineage, and hence parsimony might incline us to favor hypotheses

requiring fewer fixations. Yet if we think of fixation as simply a change in character frequency, differing in its consequences, but not causally or qualitatively different from the fluctuations ordinarily occurring from generation to generation in any polymorphic character, I cannot see any particular economy in assuming the occurrence of two fixations to be very different in probability from the occurrence of one. The spread of characters within a population is ultimately limited by the factors that limit gene flow. However, there is no reason a priori to expect polymorphisms to assort in such a way as to cause characters otherwise to respect cladistic boundaries.

The implications for phylogeny reconstruction may not be particularly discouraging, however. If an ancestral polymorphism is retained across a bifurcation in a lineage, how likely is it to produce character distributions in descendant taxa that conflict (i.e., are cladistically incongruent) with the true genealogy of the taxa?

Tajima (1983) examined a model of genetic divergence under conditions of drift, with neutral mutations accumulating at a locus at a constant rate, and observed that when the time since divergence between two populations is relatively short, a nucleotide sequence sampled from one population will often be more similar to sequences from the other population than to another sequence sampled from the same population. This situation has an especially high probability when effective population sizes are large. In a similar model, Pamilo and Nei (1988) considered genetic divergence in populations undergoing a sequence of bifurcations. Here, too, the probability that the topologies of the "gene tree" and "species tree" were the same became small when the time between the successive speciation events was short, and when the effective population size was large. Increasing the number of alleles sampled per locus within each species improved the probabilities only slightly, and with increasing numbers of terminal taxa (i.e., additional bifurcation events within the lineage) the probability of obtaining congruent trees declined. The effect of different modes of choosing founders for daughter species was examined by Neigel and Avise (1986) in a series of computer simulations. When founders of daughter species were only distantly related (as might be the case, for example, when the daughter species arise from geographically distinct portions of the parental species range), the probability of population and gene lineages coinciding was enhanced relative to the situation in which founders were chosen at random.

These studies provide important guidance for determining sampling regimes for molecular phylogenetics, but the expected magnitude of effects on – and the relevance of any particular parameter values in the models to – phylogenetic analysis of morphological characters may be difficult to assess. The example I will present does not necessarily embody qualitatively different types of assumptions, but it frames questions in different and somewhat simpler terms, without explicit reference to neutrality and drift.

Imagine a phylogeny of three taxa as shown above the arrow in Fig. 4A. (The two lineages marked "E" are monomorphic outgroups.) At the arrow, an apomorphy arises, rendering the population polymorphic. The population remains polymorphic through an initial split, and at least until the first dot encountered on each branch. In each daughter lineage, for each subsequent time interval demar-

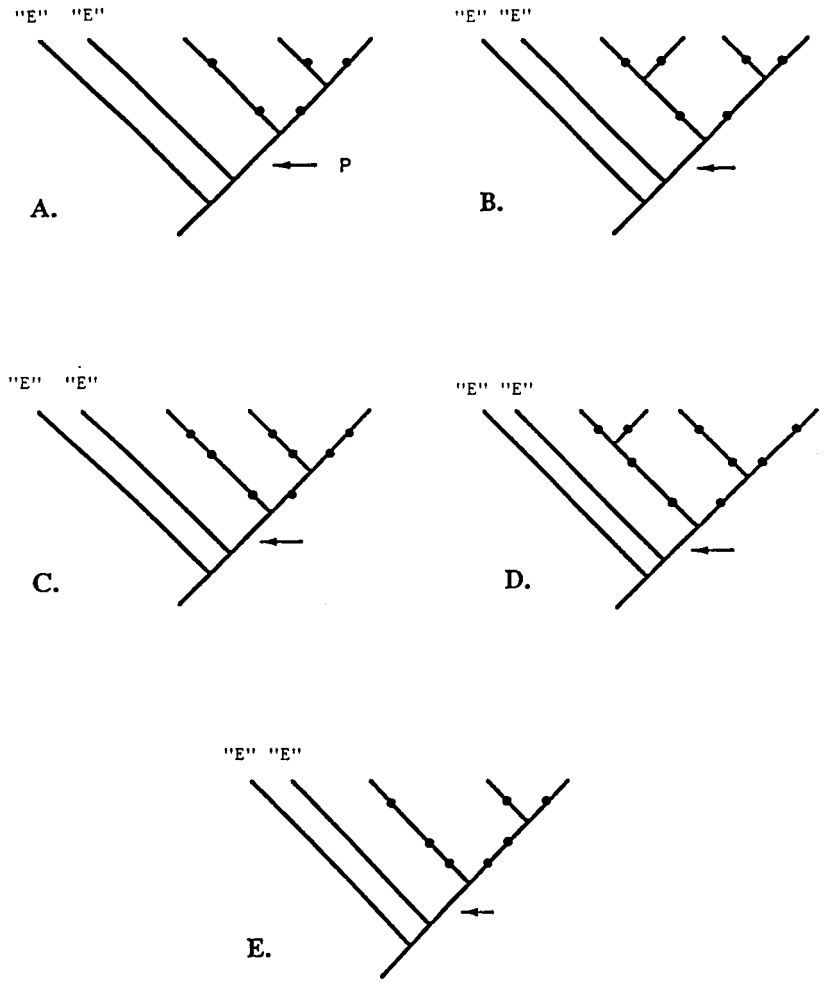


Fig. 4. Topologies of population phylogenies. Populations are assumed to be monomorphic up to the point indicated by an arrow, when polymorphism arises. Dots on branches indicate time intervals, at each of which a polymorphic population has a specified probability of remaining polymorphic [P(P)], becoming fixed for the apomorphy [P(F)], or becoming fixed for the primitive condition [P(E)].

cated by dots on the branch segments, we assign finite probabilities (say, 1/3 : 1/3:1/3, for $P(E) : P(P) : P(F)$) for each polymorphic population remaining polymorphic ($= P(P)$), going to fixation in the apomorphic state ($= P(F)$), or going to fixation in the primitive state ($= P(E)$), (i.e., the apomorphy going extinct). Once the population goes to fixation in either direction, it and its descendants remain that way. Using these probabilities to generate all possible trees in their proper frequencies, we then consider the distribution of characters (F, P, or E) in the terminal taxa

of each tree, and compute the ultimate probability of obtaining character distributions that conflict with the phylogeny.

Results for the three-taxon situation just described show congruent distributions 642 times out of 729, although congruence in fact represents many different possible evolutionary histories, and the most parsimonious scenarios are often incorrect. For example, given the polymorphism assumed in the ancestral population, a pattern such as {P{E, E}} among three terminal taxa will be generated in two ways: through two independent extinction events (i.e., in each of two terminal taxa), or by one (i.e., in their common ancestor). Yet the *reason* such a pattern would be considered a congruent one is different. The reason such a pattern would be considered congruent is that (given the "E" condition in the two outgroups) we could reconstruct it as arising in one step (origin of "P" in one terminal taxon), with no parallelisms or reversals.

Table 2 shows the consequences of (1) reducing the probabilities of remaining polymorphic from 1/3 to 1/10; (2) allowing *both* of the initially polymorphic daughter lineages to bifurcate after one time interval, to produce four, rather than three, terminal taxa (Fig. 4B); (3) allowing the system to proceed for an additional time interval without further bifurcation (Fig. 4C); (4) allowing the system to proceed for an additional time interval and imposing a bifurcation on the unbranched lineage (Fig. 4D); and (5) increasing the time interval between bifurcations (Fig. 4E).

As we might predict, reducing the probability of remaining polymorphic, allowing populations a longer common history before bifurcation events, and sampling the populations at a later time since the most recent bifurcation, all reduce the likelihood of producing a tree that conflicts with the population phylogeny (Table 2). Long time intervals (like a simple reduction in the probability of remaining polymorphic) increase the opportunity for a population to become fixed for a character, creating distributions of characters defining monophyletic groups of

Table 2. Probability of obtaining character distributions in terminal taxa (descendants of a polymorphic population) that conflict with the population genealogy, given specified probabilities of fixation of primitive or derived states (P(E), P(F)), or of remaining polymorphic (P(P)), and given particular patterns and timing of population bifurcations (illustrated in Fig. 4). It is assumed that "E" is known to be primitive, "P" intermediate, and "F" derived, so that in a three-taxon situation, for example, F{P, F} or P{E, P} would represent conflicting character distributions, but P{F, P} would not.

Topology in Fig. 3	Reference # in Text	P(E) : P(P) : P(F)	Probability of Conflict
A	(original scenario)	1/3 : 1/3 : 1/3	0.12
A	(1)	0.45 : 0.10 : 0.45	0.03
B	(2)	1/3 : 1/3 : 1/3	0.22
C	(3)	1/3 : 1/3 : 1/3	0.10
D	(4)	1/3 : 1/3 : 1/3	0.13
E	(5)	1/3 : 1/3 : 1/3	0.04

descendants. However, fixation or extinction of the apormorphy can also contribute conflict, if they happen to result in a paraphyletic pattern.

By contrast, introducing additional bifurcations (even with the addition of times as in Fig. 4D, situation # 4 described above) enhances the probability of error, but still favors reconstruction of the true phylogeny. For the worst case examined here, the ratio congruence to conflict is 3.5 : 1. The additional conflicts (beyond those counted for the original three-taxon case) result when polymorphism persists across a bifurcation, enhancing the probability of a paraphyletic distribution of the apomorphy. Moreover, for a four- (or more) taxon problem, a greater variety of incongruent patterns are inherently possible.

In any event, depending upon the relative probabilities of maintenance of polymorphism, fixation, and extinction, and the timing and pattern of population bifurcations, the potential loss of cladistic information is real, but not necessarily overwhelming.

Characters of organisms, characters of taxa, and character complexity

These examples highlight the importance of the choice of characters used in a phylogenetic analysis. It is customary to regard homoplasy in a cladogram as reflecting either mistakes (including inevitable ones) made by the systematist, or loss of phylogenetic information: characters that are not identical by descent but rather caused by evolutionary reversal or convergence. Yet as I discussed above (and as has been suggested by various molecular studies), the cause of homoplasy can also be the true *homology* of characters whose phylogeny differs from that of the population as a whole.

If we wish to trace the phylogeny of population events, perhaps we should use population-level, not individual organism-level, characters. In the situation described above, the character of individual organisms, the apomorphy x′, is homologous. The relevant character *at the level of the population*, "*fixation* of x′" is not. But independent fixation events may be more difficult to detect than independent origins of an apomorphy: characters of independent origin often look different; independent fixation events do not. We rightly assume the likelihood of a character arising more than once (alternatively stated, the likelihood of independently-originating characters being indistinguishable), to be low; the likelihood that the same character could reach fixation in more than one population is not. Fortunately, as the models described earlier in this section indicate, our chances of obtaining character distributions that are congruent with the population phylogeny remain high, even in the face of multiple fixation and extinction events.

Two points of comparison within the biological hierarchy are relevant here: the relationship between a population of individual organisms and the species, on the one hand, and (again) the relationship between genotype and phenotype.

If a phylogenetic analysis is to reflect the pattern of population divergence, it will be best to choose characters that reach fixation within the population relatively rapidly with respect to the frequency of lineage bifurcation. A particularly interesting class of such characters are those that are complex in such a way that they cannot be said to arise within single individuals, but rather characterize many

individuals from their time of origin: they become recognizable as replicating characters only very near the time that they reach fixation. An example of this type of trait is a polygenic character with a threshold of expression – that is, a character that is not recognized as present unless all its genetic components are assembled. The *components* may arise in various individuals before they spread throughout the population, but for the *trait* to be recognized as present, *all* of its components must be found together within single individuals. If the components are not genetically linked, such a character will not "breed true" (i.e., it will repeatedly be broken up by genetic recombination) until the components have reached high frequency within the population. Only at this point, close to the time at which the components reach fixation (and *after* a period in which the trait appears and disappears sporadically), will the character be observed to replicate – to be handed down consistently from generation to generation.

Characters (or homologies), both simple and complex, contain phylogenetic information, of differing sorts. An organism can be partitioned into larger numbers of unit characters if the unit characters are small (i.e., relatively simple). Greater numbers of characters can provide good resolution in clades with numerous branches; small and simple unit characters allow us to describe character evolution with greater precision. Yet, as discussed above, the phylogenies of simple characters may not be congruent with the phylogenies of population lineages, especially when the lineages branch frequently. Retention of ancestral polymorphisms through the bifurcation of a lineage may be a less frequent problem with complex (polygenic) characters, but such characters require more calls of judgement in their delimitation (how complex can a character be and still be recognized as a single unit?) and coding in the face of variability (what variation can a character sustain and still be considered the same character?) (vide Dohle, 1989).

It remains a challenge for systematists to identify and interpret characters of all organizational grades. Further understanding of the biological basis of homology, and further modelling of character evolution within populations, can only improve the accuracy with which we reconstruct phylogeny; improving our knowledge of phylogeny in turn can only improve our understanding of the biological basis of homology.

Summary and conclusion

Homology as a topic has to do with what is conserved in evolution. The problem of systematics – to find homologues, in so doing, to identify taxa – is distinct from the problem of identifying what kinds of features tend to be conserved, how, and why. The two sets of issues intersect (and are fundamentally interdependent) at the point that one (a) selects the appropriate taxonomic units, (b) identifies the characters one wishes to study, or (c) decides what constitutes a single character.

Homology is a manifestation of continuity of information (Van Valen, 1982). The complexity of development defies simplistic attempts to locate that information within the genome. Interesting difficulties (apparent paradoxes) arise because evolutionary change in genetic or developmental mechanisms may or may not be manifest at the level of gross phenotypes: change at one level of the biological hierarchy (genes, cell or tissue interactions, gross phenotype) may be screened off from variation occurring at others. Replication (which sustains continuity, the basis of homology) occurs at several levels within an individual, in biological entities of varying complexity. For this reason, a full understanding of evolutionary changes undergone in phylogeny will require knowledge of genetic processes and analyses of development as well as comparisons of gross phenotype.

The problem of understanding why some features rather than others are conserved in evolution becomes one of identifying and understanding what have been called developmental constraints (Wagner, 1989a, b). Constraints vary in strength, and can apply not only to the generation of morphological structure, but also to characters that lie closer (in level of description) to simple gene products. Gowan (1989), for example, illustrates how lichen chemosyndromes, which are sets of compounds that regularly appear in various species in particular relative concentrations, may reflect chemical pathways and products that have become individuated in much the same way as morphological structures. Used as characters, they contain phylogenetic information that correlates with phylogenetic patterns inferred from morphotypes (Gowan, 1989).

The recognition or coding of characters for a phylogenetic analysis calls for decisions on what level of description (genetic, gross phenotypic) and how complex a unit character are to be recognized. Subdividing the phenotype into large numbers of relatively simple characters might be expected to produce the greatest discrimination, yet the simpler a trait is, the more likely its phylogeny is to differ from the phylogeny of a lineage as a whole. Traits can arise, and subsequently become fixed, in different segments of a lineage, and the consequence (not widely appreciated in phylogenetic reconstruction based on morphological characters or characters that are fixed in modern populations) is conflict in the cladistic topologies suggested by different characters, or, perhaps worse, inaccuracy, with little or no apparent conflicts (compare Figs 1A and 2B). Sensitivity to the sources of error in phylogenetic analysis, and better phylogenetic hypotheses with which to analyse character evolution, will arise with reciprocal illumination among the fields of population genetics, systematics, and comparative developmental biology.

Acknowledgements

I thank Robert Brandon, John Lundberg, Lucinda McDade, Brent Mishler, Michael Schmitt, Leigh Van Valen, and Günter Wagner for valuable discussion and constructive comments on the manuscript. Günter Wagner and the European Society of Evolutionary Biology kindly gave me the opportunity to discuss some of this material during the most recent congress. I gratefully acknowledge the financial support of NSF grant DEB 85-16818 for my systematic work.

References

Archie, J. W. 1989. A randomization test for phylogenetic information in systematic data. Syst. Zool. 38: 239–252.

Arnold, E. N. 1982. Estimating phylogenies at low taxonomic levels. Z. zool. Syst. Evolut.-forsch. 19: 1–35.

Avise, J. C. 1989. Gene trees and organismal histories: a phylogenetic approach to population biology. Evolution 43: 1192–1208.

Bock, W. J. 1977. Foundations and methods of evolutionary classification, pp. 851–895. *In* M. K. Hecht, P. C. Goody and B. M. Hecht (eds.), Major Patterns in Vertebrate Evolution. NATO Advanced Study Institute, Ser. A, Vol. 14. Plenum Press, New York.

Bonhomme, F., J. X. She, E. Desmarais, F. Catzeflis and P. Boursot. 1989. Gene phylogenies and species phylogenies: reticulation and mosaicism in the genus *Mus*. Oral presentation and abstract in Second Congress of the European Society for Evolutionary biology, Rome, Italy.

Bonner, J. T. 1974. On Development. Harvard University Press, Cambridge, Massachusetts.

Brandon, R. N. 1990. Adaptation and Environment. Princeton University Press, Princeton, New Jersey.

Buss, L. W. 1987. The Evolution of Individuality. Princeton University Press, Princeton, New Jersey.

Buth, D. G. 1984. The application of electrophoretic data in systematic studies. Ann. Rev. Ecol. Syst. 15: 501–522.

Cracraft, J. 1989. Speciation and its ontology: The empirical consequences of alternative species concepts for understanding patterns and processes of differentiation, pp. 28–59. *In* D. Otte and J. A. Endler (eds.), Speciation and its Consequences. Sinauer, Sunderland, Massachusetts.

Dawkins, R. 1978. Replicator selection and the extended phenotype. Z. Tierpsychol. 47: 61–76.

De Beer, G. R. 1971. Homology, an Unsolved Problem. Oxford University Press, London , UK.

Dohle, W. 1989. Zur Frage der Homologie ontogenetischer Muster. Zool. Beitr. N. F. 32: 355–389.

Eldredge, N. 1979. Alternative approaches to evolutionary theory. Bull. Carnegie Mus. Nat. Hist. 13: 7–19.

Felsenstein, J. 1981. Evolutionary trees from gene frequencies and quantitative characters: Finding maximum likelihood estimates. Evolution 35: 1229–1242.

Field, K. G., F. J. Olsen, D. J. Lane, S. J. Giovannoni, M. T. Ghiselin, E. C. Raff, N. R. Pace and R. A. Raff. 1988. Molecular phylogeny of the animal kingdom. Science 239: 748–753.

Ghiselin, M. T. 1976. The nomenclature of correspondence: a new look at "Homology" and "Analogy", pp. 129–142. *In* R. B. Masterton, W. Hodos, and H. Jerison (eds.), Evolution, Brain, and Behavior: Persistent Problems. Lawrence Erlbaum, Hillsdale, New Jersey.

Gowan, S. P. 1989. A character analysis of the secondary products of the Porpidiaceae (lichenized Ascomycotina). Syst. Bot. 14: 77–90.

Haszprunar, G. 1989. Estimation of homology: Implications for the methodologies of classification. Oral presentation and abstract in Second Congress of the European Society for Evolutionary Biology, Rome, Italy.

Hull, D. L. 1980. Individuality and selection. Ann. Rev. Ecol. Syst. 11: 311–332.

Hull, D. 1981. Units of evolution: A metaphysical essay, pp. 23–44. *In* U. L. Jensen and R. Harré (eds.), The Philosophy of Evolution. Harvester Press, Brighton, UK.

Kauffman, S. A. 1986. A framework to think about evolving genetic regulatory systems, pp. 165–184. *In* W. Bechtel (ed.), Integrating Scientific Disciplines. Martinus-Nijhoff, Dordrecht.

Lipscomb, D. L. 1985. The eukaryotic kingdoms. Cladistics 1: 127–140.

Olson, E. C. and R. L. Miller. 1958. Morphological Integration. University of Chicago Press, Chicago, Illinois.

Osche, G. 1973. Das Homologisieren als eine grundlegende Methode der Phylogenetic. Aufsätze u. Reden senckenb. naturf. Ges. 24: 155–165.

McDade, L. Hybrids and phylogenetic systematics I. Patterns of character expression in hybrids and their implications for cladistic analysis. (in press) Evolution.

Mickevich, M. F. and C. Mitter. 1981. Treating polymorphic characters in systematics: a phylogenetic

treatment of electrophoretic data. pp. 45–58. *In* N. I. Platnick and V. A. Funk (eds.), Advances in Cladistics: Proceedings of the First Meeting of the Willi Hennig Society. New York Botanical Garden, Bronx, New York.

Mickevich, M. F. and C. Mitter. 1983. Evolutionary patterns in allozyme data: a systematic approach, pp. 169–176. *In* N. I. Platnick and V. A. Funk (eds.), Advances in Cladistics, Volume 2. Columbia University Press, New York, New York.

Mishler, B. D. and R. N. Brandon. 1987. Individuality, pluralism, and the phylogenetic species concept. Biology and Philosophy 2: 37–54.

Neigel, J. E. and J. C. Avise. 1986. Phylogenetic relationships of mitochondrial DNA under various demographic models of speciation, pp. 515–534. *In* E. Nevo and S. Karlin (eds.), Evolutionary Processes and Theory. Academic Press, New York, New York.

Nijhout, H. F., G. A. Wray, C. Kremen and C. K. Teragawa. 1987. Ontogeny, phylogeny, and the evolution of form: an algorithmic approach. Syst. Zool. 35: 445–457.

Otte, D. and J. A. Endler (eds.) 1989. Speciation and its Consequences. Sinauer, Sunderland, Massachusetts.

Owen, R. 1848. On the Archetype and Homologies of the Vertebrate Skeleton. R. and J. E. Taylor, London, UK.

Pamilo, P. and M. Nei. 1988. Relationships between gene trees and species trees. Molec. Biol. Evol. 5: 568–583.

Patterson, C. 1977. Cartilage bones, dermal bones and membrane bones, or the exoskeleton versus the endoskeleton, pp. 77–121. *In* S. M. Andrews, R. S. Miles and A. D. Walker (eds.), Problems in Vertebrate Evolution. Linnean Society Symposium Series # 4. Academic Press, London, UK.

Patterson, C. 1982. Morphological characters and homology, pp. 21–74. *In* K. A. Joysey and A. E. Friday (eds.), Problems of Phylogenetic Reconstruction. Academic Press, London, UK.

Patterson, C. 1988. Homology in classical and molecular biology. Mol. Biol. Evol. 5: 603–625.

Raff, R. A. and T. C. Kaufman. 1983. Embryos, Genes, and Evolution. Macmillan, New York.

Remane, A. 1952. Die Grundlagen des natürlichen Systems, der vergleichenden Anatomie und der Phylogenetik. Geest and Portig, Leipzig, East Germany.

Riedl, R. 1977. A systems-analytical approach to macroevolutionary phenomena. Quart. Rev. Biol. 52: 351–370.

Riedl, R. 1978. Order in Living Organisms: a Systems Analysis of Evolution. Wiley, New York.

Riedl, R. 1982. A dialectic approach to epigenetics and macroevolution, pp. 41–50. *In* V. J. A. Novak and J. Mlikovsky (eds.), Evolution and Environment. Academid, Prague, Czechoslovakia.

Riedl, R. 1989. Homology and the pattern of morphological variation. Oral presentation and abstract in Second Congress of the European Society for Evolutionary Biology, Rome, Italy.

Roth, V. L. 1982. On homology. Biol. J. Linn. Soc. 22: 13–29.

Roth, V. L. 1988. The biological basis of homology, pp. 1–26. *In* C. J. Humphries (ed.), Ontogeny and Systematics. Columbia University Press, New York.

Salmon, W. C. 1971. Statistical Explanation and Statistical Relevance. University of Pittsburgh Press, Pittsburgh, Pennsylvania.

Sander, K. 1983. The evolution of patterning mechanisms: gleanings from insect embryogenesis and spermatogenesis, pp. 137–160. *In* B. C. Goodwin, N. Holder and C. C. Wylie (eds.), Development and Evolution, Cambridge University Press, Cambridge, UK.

Sander, K. 1989. Genes and homology. Oral presentation and abstract in Second Congress of the European Society for Evolutionary Biology, Rome, Italy.

Schmitt, M. 1989. Das Homologie-Konzept in Morphologie und Phylogenetik. Zool. Beitr. N. F. 32: 505–512.

Schoch, R. M. 1986. Phylogeny Reconstruction in Paleontology. Van Nostrand-Reinhold, New York.

Simpson, G. G. 1975. Recent advances in methods of phylogenetic inference, pp. 3–19. *In* W. P. Luckett and F. S. Szalay (eds.), Phylogeny of the Primates. Plenum Press, New York.

Sogin, M. L., J. H. Gunderson, H. J. Elwood, R. A. Alonso and D. A. Peattie. Phylogenetic meaning of the kingdom concept: an unusual ribosomal RNA from *Giardia lamblia*. Science 243: 75–77.

Soulé, M. E. 1982. Allomeric variation. I. The theory and some consequences. Amer. Nat. 120: 751–764.
Stevens, P. 1984. Homology and phylogeny: morphology and systematics. Syst. Bot. 9: 395–409.
Swofford, D. L. and S. H. Berlocher. 1987. Inferring evolutionary trees from gene frequency data under the principle of maximum parsimony. Syst. Zool. 36: 293–325.
Tajima, F. 1983. Evolutionary relationships of DNA sequences in finite populations. Genetics 105: 437–460.
Throckmorton, L. H. 1962. The problem of phylogeny in the genus *Drosophila*. Studies in Genetics 2: 207–343.
Van Valen, L. 1962. Growth fields in the dentition of *Peromyscus*. Evolution 16: 272–277.
Van Valen, L. 1982. Homology and causes. J. Morphol. 173: 305–312.
Wagner, G. 1989a. The origin of morphological characters and the biological basis of homology. Evolution 43: 1157–1171.
Wagner, G. P. 1989b. The biological homology concept. Ann. Rev. Ecol. Syst. 20: 51–69.
Watzek, G., F. Frundschober, II. Plenk Jr. and J. Eschberger. 1982. The influence of various surgical procedures upon bone growth at viscerocranial sutures, pp. 347–364. *In* A. D. Dixon and B. G. Sarnat (eds.), Factors and Mechanisms Influencing Bone Growth. Alan R. Liss, New York.
Wimsatt, W. C. and J. C. Schank. 1988. Two constraints on the evolution of complex adaptations and the means for their avoidance, pp. 231–273. *In* M. H. Nitecki (ed.), Evolutionary Progress. University of Chicago Press, Chicago, Illinois.

Received 27 December 1989;
accepted 11 May 1990.
Corresponding Editor: G. Wagner.