

POINT OF VIEW

ORIGIN OF EVOLUTIONARY NOVELTIES AND ELIMINATION OF PLESIOMORPHIC ALLELES: SOME COMMENTS ON LIMITATIONS OF THE CONCEPT OF SYNAPOMORPHY

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ABSTRACT

The intuitive concept of synapomorphy involves the idea of modified (apomorphic) features shared by a set of organisms. From the evolutionary point of view, the origin of new features involves at least two processes of completely different nature. One of them is the molecular process of modification of a pre-existing gene at a given locus; the other is the process of plesiomorphic allele elimination at that locus in the population. These two events do not occur at the same time in the history of a "synapomorphy". Actually, they may occur in the history of a phyletic stem separated by one or more cladogenetic events. In these cases, the result is the inheritance of polymorphisms between ancestral-descendent species. The radical differences between these two processes recommend a conceptual discernment between them. Shared evolutionary novelties (apomorphic alleles) and shared absence of plesiomorphic alleles are named herein, respectively, syntrepty and synapousy. Many cases understood as homoplasies - more than one origin of the same evolutionary novelty - may correspond to more than one independent event of elimination of the plesiomorphic allele. One of the most important consequences for the phylogenetic analysis is that syntreptic and synapousic events are not directly comparable as probabilistic entities. This has important effects on the application of the parsimony concept on allele matrix analysis.

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The concept of apomorphy involves the idea that a given characteristic of a biological organisms is a modification of a pre-existing homologous trait. A synapomorphy is an apomorphy shared by two or more groups (Hennig, 1966; Wiley, 1981). These concepts can explain most of the actual situations that the theory of phylogenetic systematics has dealt with until now. Consider, however, the example in Figure 1. Araújo (1990) studied fifteen enzymatic systems (five of which are represented in Figure 1) of a group of five species of the genus *Achirus*, one species of *Trinectes* (Actinopterygii: Soleidae), and five species of two related families (Bothidae and Cynoglossidae) (Araújo, 1990). The distribution of the alleles of each enzyme sampled is at the top of the cladogram. In the way it is presently conceived, the concept of synapomorphy can not handle appropriately with this kind of data.

Since the beginning of the century, it was quite clear that genetic modifications - in the sense of evolutionary novelties - originated in a single gene of a single chromosome of a single gametic cell of a single individual of a population. That is, the genesis of an evolutionary novelty is a very particular event in time and space in the history of a population. After the origin of any new allele, there is a period in the history of a population during which the new allele coexists with the pre-existing homologous allele(s) in the evolutionary stem in which it arose. Later in time the older allele may completely disappear from that genetic system. Hence, the process of generation of a "synapomorphy" involves two evolutionary events: the origin of the apomorphic allele and the elimination of the plesiomorphic allele from the population. These events are entirely different from each other and occur independently in time, the former necessarily preceding the latter. The first is a physico-chemical event occurring at the cytological level and corresponds to errors in the nucleic acid duplicating machinery. The second is a populational event and is generated by sampling errors between generations and/or differentially selective elimination of phenotypes.

Throckmorton (1965), who defended a phylogenetic method in taxonomy and phylogenetic classifications, from his work with *Drosophila* taxonomy perceived very early that inherited polymorphisms with independently fixed alleles could generate false hypotheses of monophyly. However, very few of the strict phylogeneticists have addressed the necessary attention to the fact that two independent processes are involved in the generation of an "apomorphy". Wiley (1981: 125) gave some attention to the problem of the evolution of polymorphisms, without going through its consequences. Saether (1979) has proposed the concept of "underlying synapomorphies", which express the pattern of multiple origin of similar apomorphies in related groups, but his analysis is restricted to the phenotypic expressions of changes and does not approach the genetic basis of the question. Felsenstein (1979) is one of the few who more explicitly approached the problem (see below) and Takahata (1989) also proposed a numerical model of analysis in a context of inherited polymorphic alleles, but important recent papers dealing with

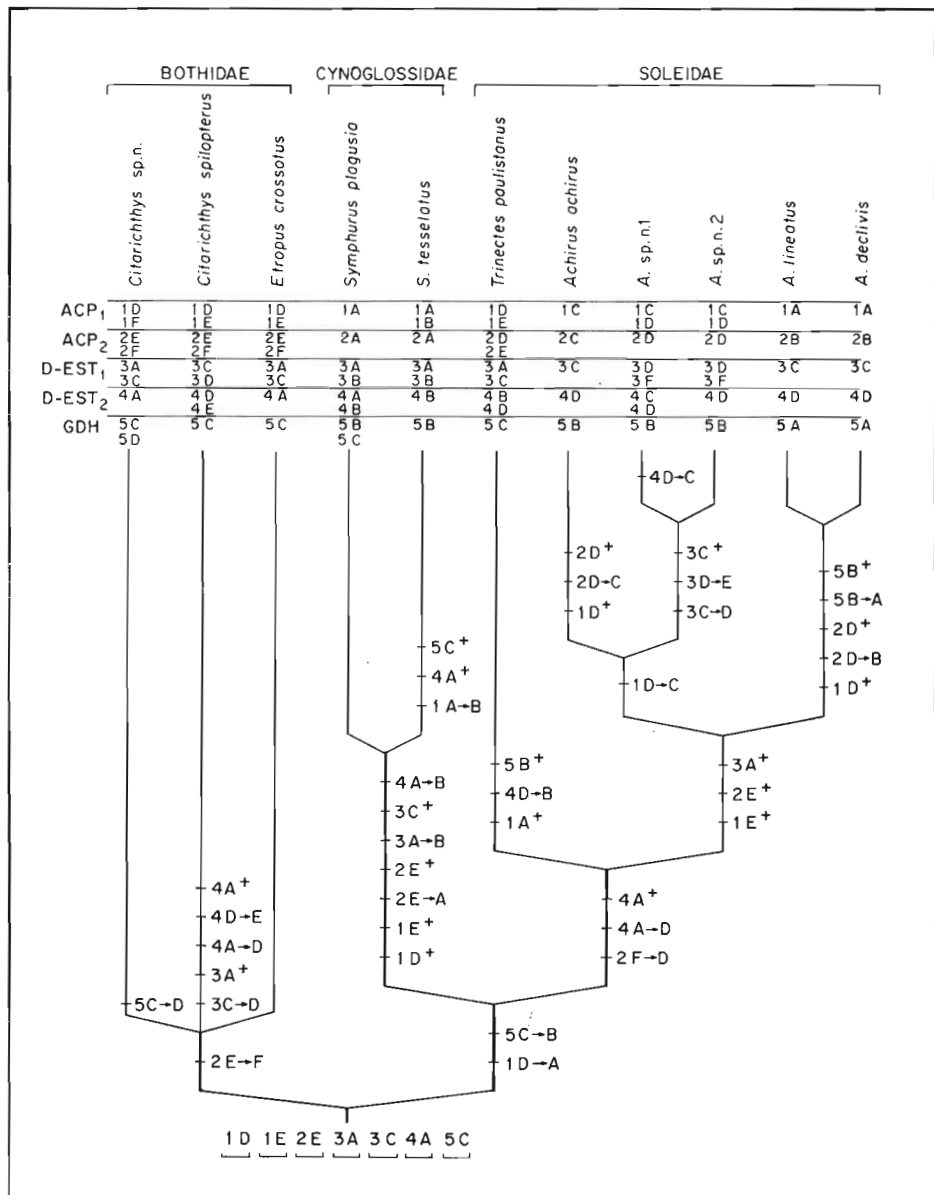


Figure 1 - Evolution of five enzymatic systems in Pleuronectiformes. The alleles found in each enzyme (ACP₁, ACP₂, D-EST₁, D-EST₂, GDH) in the species studied are given above, at the cladogram, below the name of the species. At various levels of the cladogram hypotheses of syntreptics and synapousis are presented. The presence of a pair of alleles with an arrow between them indicates a syntreptic event (the former is the plesiomorphic and the latter is the apomorphic allele). The presence of an allele followed by a cross indicates a synapousis event, in which that allele was lost.

methodological aspects related to molecular data (e.g., Patton and Avise, 1983; Swofford and Berlocher, 1987; Mooi, 1989) have not discerned between these events and, consequently, have not dealt with the question of how they affect the analysis.

The original concept of synapomorphy generally applies to the process of generation of new characters in monophyletic groups, but does not refer directly to events below the population level. If in the history of a phyletic stem the origin of a novelty and the respective elimination of the plesiomorphic allele occur at the same level, that is, between two known cladogenetic event - a "single" synapomorphy may represent it in a cladogram. This is the way it has traditionally been represented, although both evolutionary, independent events have occurred. However, if there is at least one known cladogenetic event between the origin of a novelty and the elimination of its plesiomorphic allele, the cladogram can not adequately represent the event as one step. It is possible that many cases of "homoplastic origin" of similar characters among very related groups may not be due to an independent origin, but simply to independent fixations of the same apomorphic allele, as suggested Throckmorton (1965).

As these events are of different natures, we believe that they should be named differently. It would be better to restrict the name synapomorphy to the cases in which there is no allele information available. We will refer, then, to a syntrepty (from the Greek $\tau\rho\epsilon\pi\tau\omicron\varsigma$ = changeable, variable) for the shared evolutionary novelty, whether its plesiomorphic allele is present or not. On the other hand, we will refer to a synapousy (from the Greek $\alpha\pi\omicron\upsilon\sigma\iota\alpha$ = absence) for the "shared absence" of a plesiomorphic allele, regardless of the level from which its homologous apomorphic allele arose. These terms will be more rigidly defined elsewhere using the formal mereological language applied to comparative biology, presently being developed by Dr. Nelson Papavero and by Prof. Jair M. Abe. Syntrepties always imply modifications, producing new, derived alleles, which will only rarely be perfect reversions. However, synapousies act on two or more coexisting alleles, any of which can be lost, including the apomorphic ones. When a phylogenetic analysis focuses on very high levels of taxonomic hierarchy, the extinction of intermediate stems makes it more likely to find corresponding syntrepties and synapousies occurring at the same level. However, we certainly can not expect the same at the lower levels of analyses.

In this conceptual discernment is important for the theory, its implications for phylogenetic analysis are also considerable. The method of dealing individually with syntrepties or synapousies is identical to the phylogenetic reasoning used for synapomorphies, since they are events "inherited" through generations of a population. All descendants of an ancestral species share evolutionary novelties (trepties) arisen in that species, except if secondarily eliminated or modified. Lost plesiomorphic alleles (apousies) will be absent in all descendants of an ancestral species in which it has been eliminated, except if secondarily generated. However, a "synapomorphy", from the point

of view of the new concepts, may involve either: (1) a single event of syntrepty, without further fixation - the stem or its descendant stems, if divisions occur, retain the polymorphism; (2) a single syntrepty and a single correspondent synapousy, both occurring at the same level; (3) one syntrepty and one or more synapousies occurring in different descendent stems in which the plesiomorphic or the apomorphic allele is lost. When no information is available about shared alleles (i.e., if we deal only with phenotype information), we must use the traditional concept of synapomorphy. However, if allele data is at hand, this kind of analysis may improve much more information about the history of the taxa studied and furnishes additional corroboration to hypothesis of phylogenetic relationships.

If no incongruence occurs between synapousies and syntrepties, the analysis of allele data is made as usual in phylogenetic systematics (Figure 2). However, some serious problems arise if there is incongruence between syntrepties and synapousies (Figure 3). Analyses, at least for the time being, can not use parsimony to analyze matrices with both, syntreptic and synapousic characters presenting incongruence: syntrepties and synapousies are events of a different nature and can not be directly comparable as probabilistic events.

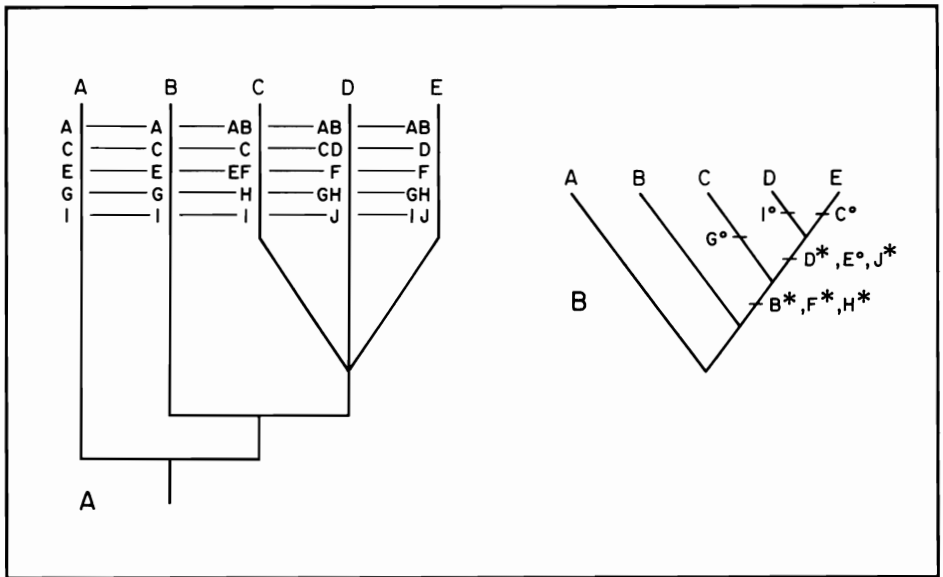


Figure 2 - Hypothetical case of distribution of alleles in which there is no incongruence between the data on the acquisition of new alleles and losses of alleles. A. Distribution of alleles of five enzymes in five species, three of which compose a monophyletic group with undetermined internal phylogenetic relationships; B. Proposed reconstruction of the relationships among the species C, D, and E based on the allelic data available, with the attribution of generality of the events of syntrepties (e.g. B^*) and synapousies (e.g., G°) in the group C + D + E.

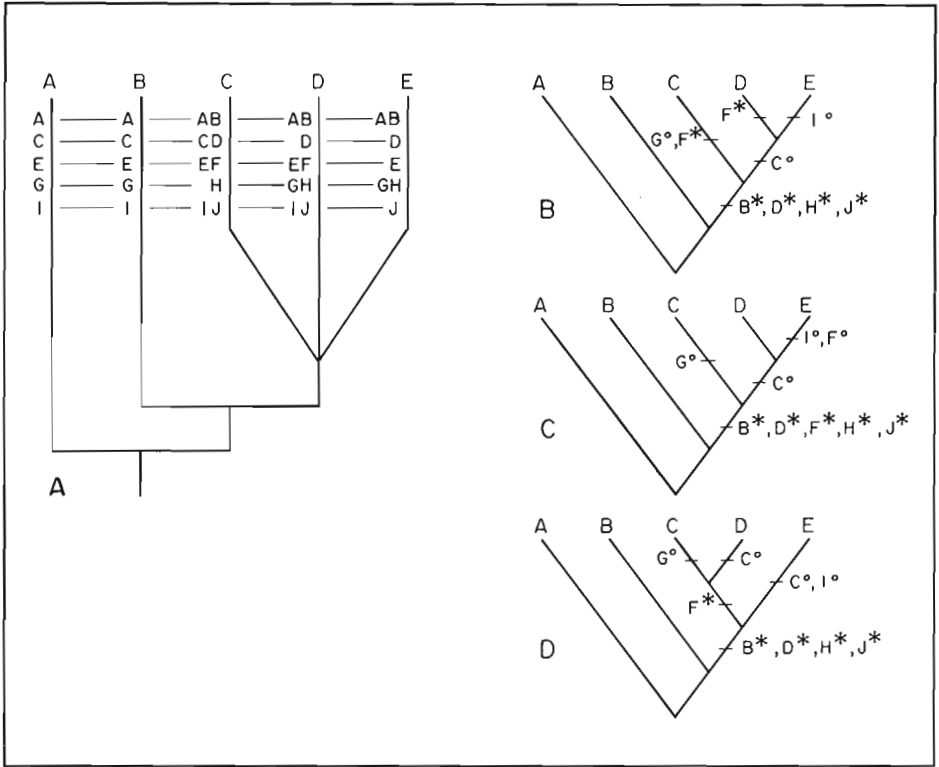


Figure 3 - Hypothetical case of distribution of alleles in which there is incongruence between syntrepties and synapousies. A. Distribution of alleles of five enzymes in five species, three of which compose a monophyletic group with undetermined internal phylogenetic relationships; B. One of the possible solutions for the relationships among species C, D, and E based on the allele data available. A single loss of the allele C and two independent origins of the allele F is seen as more parsimonious than two independent losses of allele C and a single origin of allele F; C. Same as B, but with a different interpretation for the evolution of the allele F; D. Alternative solution for the relationship among the species C, D, and E. A single origin of allele F and two independent losses of allele C is seen as more parsimonious than two independent origins of allele F and a single loss of allele C.

Felsenstein's (1979) polymorphism method is a first step towards dealing with the methodological questions involved, although he did not address the conceptual nor the ontological basis of the problem. Felsenstein (1979; see also 1983) compared different numerical methods of phylogenetic analysis, considering a model in which a character could be 0, 01 or 1 in a population, with probabilities attributed for shifts of the character from one condition to another. The change from 0 to 01, in the terminology proposed here, is a syntrepty, and from 01 to 1 or to 0 a synapousy. In the case of Figure 3, if we consider synapousic events as more reliable indicators of phylogenetic relationship than

syntrepties (in Felsenstein polymorphism model, $b/2 dt \ll a dt$), then options 3B and 3C would be more acceptable. If syntrepties are more reliable ($b/2 dt \gg a dt$), then Figure 3D would be the best option. Another point is that Felsenstein (1979) considered the probabilities of a polymorphic feature to lose either the apomorphic or plesiomorphic alleles to be the same, what may prove to be a false premise of the method. The understanding of the evolution of alleles finds a number of other problems, as the reconstruction of the transformation series of alleles. In Figure 1, for example, one may propose many different possible histories for the alleles in the same cladogram. The scope of this paper is mainly conceptual and these points are left to be worked out elsewhere.

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RESUMO

O conceito intuitivo de sinapomorfia envolve a idéia de características modificadas (apomórficas) compartilhadas por um conjunto de organismos. Do ponto de vista evolutivo, a origem de novas características inclui dois processos de natureza completamente distinta. Um deles é o processo molecular de modificação de um gene pré-existente em um dado loco; o outro é o processo de eliminação de um alelo plesiomórfico daquele loco na população. Esses dois eventos não ocorrem ao mesmo tempo na história de uma "sinapomorfia". Na verdade, eles podem ocorrer na história de um ramo filético separados por um ou mais eventos cladogenéticos. Nesses casos, o resultado é a herança de polimorfismo entre espécies ancestrais-descendentes. Novidades evolutivas (alelos apomórficos) compartilhadas e ausência de alelos plesiomórficos são aqui denominados, respectivamente, syntreptias e sinapousias. Muitos casos na literatura compreendidos como homoplasias - mais de um evento de origem de uma novidade evolutiva - podem corresponder, na verdade, a mais de um evento de eliminação do alelo plesiomórfico de um loco. Uma das conseqüências mais importantes para a análise filogenética é que eventos de syntreptia e sinapousia não são diretamente comparáveis como eventos probabilísticos. Isto tem efeitos importantes na aplicação do conceito de parcimônia a análises de matrizes de caracteres em que estão apresentados dados de distribuição de alelos.

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