Essay: Homology [1]

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1. Overview

Homology [4] is a central concept of comparative and evolutionary biology, referring to the presence of the same bodily parts (e.g., morphological structures) in different species. The existence of homologies is explained by common ancestry, and according to modern definitions of homology, two structures in different species are homologous if they are derived from the same structure in the common ancestor. Homology [4] has traditionally been contrasted with analogy, the presence of similar traits in different species not necessarily due to common ancestry but due to a similar function or convergent evolution [9], resulting from similar selective pressure in different species. (A more recent contrastive notion is homoplasy, the presence of similar traits in different species without common ancestry, i.e., as an instance of parallel evolution [5]). This sounds straightforward, but in fact the homology concept has a rich history and currently is the subject of extensive theoretical reflection, resulting in different contemporary approaches to homology.

Despite the phylogenetic nature of homology, the homology concept was introduced in early nineteenth century comparative anatomy and embryology [8], and became an influential aspect of comparative practice well before the advent of Darwinian evolutionary theory. In this period and until the first half of the twentieth century, two main criteria were used to establish homologies across species. The positional criterion maintains that homologous structures in different species retain their relative topological positions. For instance, the shape of a certain bone may vary across different species (e.g. if it serves a different function in different species), but this bone will usually be adjacent to or articulate with the same set of other bones across these species. The embryological criterion assumes that homologous structures in different species develop out of the same developmental precursors. This made many cases of homology discovery possible, as the early developmental stages [25] of different species are more similar than the respective adult forms. While the positional criterion primarily relies on comparing the adult morphologies of different species, the embryological criterion involves comparative embryology [20] as an additional source of evidence. However, there are cases where both criteria disagree, which historically led to a conflict between approaches favoring the comparative anatomy of adults and those relying on comparative embryology [9] (Section 3 below).

In addition to its central role for biology and its longstanding history, developments in the second half of the twentieth century strongly enriched and diversified the homology concept. With the advent of phylogenetic systematics [21], homologies came to be consistently assessed by means of the distribution of, homologous traits on phylogenetic trees. The positional and embryological criteria could be and were used independently of any phylogenetic tree, but they are fallible criteria for establishing homologies understood as structures with a common ancestry (Section 4). The advent of molecular evolution [20] and molecular evolution [20] made prominent the idea that molecular structures such as genes [10] and proteins can be homologous across species. In general, many kinds of biological entities are nowadays homologized: molecules, cellular structures, cell types, tissues, developmental modules and processes, gross morphological structures, and behavioral patterns. It is widely recognized that homologues exist on different levels of organismal organization [13]. Furthermore, homologies on different hierarchical levels do not align: there are many cases where developmental processes homologous in two species develop by different developmental processes and/or by the involvement of non-homologous genes [24]. Conversely, the same, homologous gene can be involved in the development of non-homologous structures in different species. As a result, it is nowadays often assumed that homology on one level of organization [13] cannot be reduced to homology on another (e.g. lower) level, supporting a non-reductive and hierarchical view of organisms.

Since homologous structures can develop by different developmental mechanisms, and may develop out of non-homologous developmental precursors, the embryological criterion of homology ultimately fails. However, this does not diminish the role of developmental biology for homology. On the contrary, embryonic structures and developmental processes are an additional and independent level of organization [16] where homologies occur. Furthermore, apart from taxic and transformational approaches to homology (discussed in Section 4), of particular importance are developmental approaches to homology (sometimes called a biological homology concept [12]). The latter attempt to explain why a homologue reappears in different generations and is often present as the same morphological unit across many species, despite undergoing evolutionary change in its internal features. Developmental biology is essential to understand how homologies can function as units of morphological evolution [5], an issue which is central to contemporary evolutionary developmental biology [20].

A detailed survey of the history of the homology concept follows, which pays particular attention to the relation of this concept to development and embryology [8].


Long before the term 'homology' was introduced and the idea of homology clearly spelled out, seventeenth and eighteenth century comparative anatomists and naturalists studied biological characters of known and newly discovered species, recognizing that different species can have the same anatomical structures by giving the same name to them. However, these early naming practices were idiosyncratic and not based on explicit criteria. Usually the same name was applied to characters with a similar shape, internal structure, and function, and only to taxonomically closely related species (e.g. different mammals). The idea of homology originated with the recognition that the same structures exist in less closely related species (mammals and birds [19], or even mammals and fish [20]) and that the sameness of morphological units is independent of their function and form. This idea developed in comparative anatomy independently in Germany and France, though from 1820 onwards both traditions influenced each other as well as British zoology (Appel 1987).

In the German context, the relevance of Johann Wolfgang von Goethe (1749–1832) and other morphologists such as Lorenz Oken (1779–1851) is well known. For the tradition of Naturphilosophie [18], homology was one of the manifestations of the unity in nature that it emphasized. Of particular concern was what is nowadays called serial homology, i.e., the repeated occurrence of the same morphological unit in one and the same individual. For instance, the famous vertebral theory of Oken, which established that the different skull bones are in fact transformed vertebrae (Nyhart 1995, Rupke 1994, Russell 1916).

In France, of pivotal importance was the work of Etienne Geoffroy (1772–1844). While previous anatomists, including Georges Cuvier [16] (1769–1832), had assumed that many vertebrate structures were present in only one of the four vertebrate classes (fishes, reptiles, birds [19], mammals), Geoffroy found homologies across these classes. His 'philosophical anatomy' [2] posited the unity of organic composition. In particular, his 'principle of homologous structures' (the name homologue) is based on the fact that all vertebrates consist of the same number of basic building elements (Geoffroy Saint-Hilaire 1811). Later he even attempted to homologize structures from different Cuvierian embranchements [6] (vertebrates, molluscs, articulates, radiates; which unlike contemporary phyla were defined by Cuvier in terms of functional organ systems). This triggered the famous public dispute between Geoffroy and Cuvier in 1830, but the disagreement was rooted in Cuvier’s emphasis on functional considerations in anatomy, whereas for Geoffroy homology was subordinated to structure/homology as the same structure could fulfill different functions (Appel 1987). For instance, Geoffroy showed that the scutula, the wishbone used to exist only in birds [19], is present in fishes as well, and he homologized structures of normally developed animals with malformed structures and tetany, which have an altered or no function. Geoffroy is so important to this discussion because he introduced a major criterion of homology. His principe de connexions stated that what matters is the relative position of structures and in which the anatomical relations connected to and combined with each other in an organism. For instance, the same muscles are innervated by the same nerves in different species. Homologues in different species may vary in shape and function, but the different homologues composing a body maintain their relative position (Le Guyader 2004, 30–32). This yields a way of recognizing homologies (even across unrelated species where a structure differs substantially in its form and function), nowadays called the positional criterion.

The French tradition of 'philosophical anatomy' was continued and developed by Geoffroy’s disciple Etienne Renaud Augustin Serres (1786–1868), among others. It established an important point of contact between embryology and the concept of homology, due to the influential idea of recapitulation, i.e., the view that the development of higher animals recapitulates the adult forms of animals lower on the organic scale. Recipitulation in the pre-Darwinian context was dubbed the Meckel-Serres law (by Russell 1916), as already the German anatomist Johann Friedrich Meckel (1781–1833) had defended the idea in his 1811 essay 'Sketch of a Portrayal of the Parallel that Obtains Between the Embryonic Condition of Higher Animals and the Permanent Condition of Lower Animals' [23]. Meckel 1811; Lorenz Oken (1816) expressed similar views at the same time). Serres used this idea to defend the general validity of Geoffroy’s unity of organic composition tenet, by arguing that comparing adult forms only may be the wrong point of reference. Instead, one has to compare the adult form of lower animals with the embryonic stage of higher organisms, thereby recovering otherwise unnoticed homologies. Furthermore, Serres and Geoffroy explained development with reference to the idea of a nius formativus (formative drive [20]), which is a force guiding the development to the final adult stage. The fact that the development of a lower animal is only an initial segment of the developmental sequence of a higher animal was misled by the claim that lower animals have less of this formative drive [20]. Serres’s theory of the arrest of development [19] for recapitulation was closely tied to his study of teratology [16]. Malformed organs in higher animals were viewed as structures from an organism on a lower level of the scale of being. This yielded an embryological theory that explained homologies across species and included an account of morphological relations between normally and abnormally developed organisms. (Geoffroy used these ideas on tetany development for a proto-evolutionary theory, by viewing malformations as being due to external influences on the embryo and assuming that the environment was capable of directly acting on the developing fetus [24] so that a sort of evolutionary change resulted.)

Apart from homology’s role in comparative anatomy, comparative embryology [8] led to a major theoretical enrichment of the concept of homology and the practice in which it figured. In particular, what is now called the embryological criterion of homology found its first clear expression in the work of the Estonian comparative embryologist Karl Ernst von Baer (1792–1876). Von Baer’s embryological theory was in fact part of a critique of recapitulation, as endorsed by Meckel and Serres. Apart from counterexamples to recapitulation, von Baer defended an alternative account of comparative development, which he summarized in four laws, later referred to as von Baer’s laws [17] (1828–1832). On this theory, while the early embryos of different vertebrates cannot be distinguished from each other, later in development successive differentiation [20] takes place in that an embryo acquires the features that characterize its order, family, and finally its species. The view is not that the human embryo’s development recapitulates
the adult forms of lower animals. Instead, the human and the insects
insects
3. Homology 4) after the advent of evolutionary theory

With the advent of evolutionary theory, previous morphological and taxonomic notions became reintegrated in the light of phylogeny 4). Homologies came to be viewed as being due to common ancestry, morphological types were interpreted as shared body plans inherited from an ancestral type, and taxa came to be conceived as branches of the tree of life. This interpretation of previous ideas was already advocated by Charles Darwin 5) (1809–1882) in Chapter 13 of the Origin of Species (1859), but it received its most complete implementation by the discipline of evolutionary morphology 4), as promoted by Carl Gegenbaur 6) (1825–1903) and Ernst Haeckel 7) (1834–1919) in Germany and Edwin Ray Lankester 8) (1847–1929) in Great Britain (Lankester 1870). As one of the most thriving disciplines within all of biology in the second half of the nineteenth century, evolutionary morphology 9) was less concerned with the study of the mechanisms of evolutionary change (such as natural selection 10), but consisted in a thoroughly phylogenetic approach in morphology and taxonomy, including the establishment of phylogenetic trees and patterns of morphological evolution 4).

It is often (falsely) assumed that evolutionary theory introduced a novel homology concept: an ‘evolutionary’ homology concept defined in terms of common ancestry, to be distinguished from the pre-Darwinian ‘idealistic’ homology concept defined in terms of morphological types. However, this interpretation ignores the continuity that existed in the practice of nineteenth century morphology 4), including the actual use of the homology concept, and is based on the flawed essentialism story about pre-Darwinian biology (Amundson 2005). Even though homology came often to be defined in terms of common ancestry, evolutionary morphologists did not read off homologies from phylogenetic trees (showing common ancestry), but instead established homologies using the positional and embryological criterion, just like morphologists in the first half of the nineteenth century (Russell 1916). Known homologies formed the basis for detailed morphological comparison, and only in the last step were phylogenetic trees set up. In addition to not introducing any novel criterion of homology, the advent of evolutionary theory did not change what researchers attempted to achieve by the use of the homology concept. Throughout the nineteenth century, this concept was used for the purpose of morphological comparison and the classification of species. Overall, rather than overturning previous practice, evolutionary morphologists made existing practice more sophisticated by interpreting traditional notions in the light of common ancestry (Coleman 1976).

An important element of theoretical continuity across pre- and post-Darwinian biology was given by development. In the first half of the nineteenth century, embryological ideas had been used to see the nature of homology and the shared morphological organization 11) of different species. After the advent of evolutionary theory, definitions of homology as the common development of structures in different species—in line with the embryological tradition—were commonly seen (e.g. Darwin 1859, p. 512). More generally, biologists in the second part of the nineteenth century conceived of orthography 4) and phylogeny 4) as two parts of one coin, by viewing phylogenetic patterns in analogy to patterns of development and sometimes using assumptions about developmental processes to theorize about the mechanisms of evolutionary change, in particular in the models of Lamarckism 12) and orthogenesis 13) (Bowler 1988). A clear-cut connection between evolution 4) and development was given by Haeckel’s biogenetic law 14). This law states that every organ is mechanically caused by phylogeny 4), which is in turn causally determined by the recapitulationist pattern postulated by the biogenetic law 15). Known homologies formed the basis for detailed morphological comparison, and only in the last step were phylogenetic trees set up. In addition to not introducing any novel criterion of homology, the advent of evolutionary theory did not change what researchers attempted to achieve by the use of the homology concept. Throughout the nineteenth century, this concept was used for the purpose of morphological comparison and the classification of species. Overall, rather than overturning previous practice, evolutionary morphologists made existing practice more sophisticated by interpreting traditional notions in the light of common ancestry (Coleman 1976).

Around the turn of the century, however, evolutionary morphology 4) lost in significance, largely due to experimental embryology 4) (developmental mechanics) as an approach concerned with the experimental study of model organisms but not with comparative or phylogenetic questions. One factor fueling this development was a clash between comparative anatomy (e.g., the use of the positional criterion to compare structures across species) and phylogenetics, so that it originally seemed reasonable to assume that homologous anatomical structures develop by means of the recapitulationist pattern postulated by the biogenetic law 15), which establishes phylogenetic trees based on comparisons of adult forms of different species. However, this overall feature distribution across species is best explained by the assumption that a particular feature originated in an ancestral species and has been inherited to all its descendants, a situation called synapomorphy. (The phylogenetic tree is obtained by the study of many different characters, where the most likely tree is the one that shows the shallowest branching and the fewest homoplasious 4) features.) While in the nineteenth century homologies were established using the positional and embryological criterion (and some additional criteria) that do not presuppose a phylogenetic tree, the advent of embryology 4) for morphological studies and theorizing about the structural relations of different species (Lenor 1982). A variant of the embryological concept could also be used by proponents of recapitulationism. After the later more formal formulation of the germ layer theory, the embryological concept came to imply that homologous structures always develop from the same germ layer.

Before the advent of Darwinian evolutionary theory, the pinnacle of the biological practice based on the homology concept was the work of the British anatomist Richard Owen 16) (1804–1892). By coining the distinction between homology and analogy, Owen 16) laid the basis for the practice of evolutionary morphology 4) and embryology 4) independently of the outcomes of embryology 4) and developmental processes, and developmental modules are often viewed as homologous characters 4). In this context, homology is contrasted with homoplasy, which is the occurrence of similar character states in two species not due to common ancestry (but parallel evolution 4).

4. Development and hierarchy. Approaches to homology

Over the past few decades there has been an intensification of interest in the concept of homology. Its scope of application has increased and new theoretical interpretations of homology have been proposed (Donoghue 1992, Brignall and Griffiths 2007). In addition to traditional anatomical studies, anatomy 4) introduced the idea that behavioral patterns in different species can be homologous. With the rise of molecular biology, genes 4), proteins, and other molecular structures came to be homologized, making possible the fields of molecular phylogeny 4) and evolution 4), which establish phylogenetic trees based on molecular data. Since the distinction between homology and analogy is based on the function of structures and the concepts of homoplasy 4) and synapomorphy 4), the relationship of homology and analogy to other biological concepts is dependent upon the definition of homology and the criteria used to establish homologies 4).
The evolutionary approach to homology focuses on how a certain character state can evolve independently of each other, even though an anatomical structure develops based on developmental processes and the action of evolutionary forces [49]. So that in development there are close causal-functional relations among characters on different levels, there are apparently partial developmental dissociations among these characters and levels (yet to be understood), which permit the independent evolution of characters. Thus, developmental approaches to homology are germane to the explanation of evolvability as well as modularity in evolution [50] and development. Since homologies have to be explained in the first place, a connection to explanations of evolutionary novelties exists. Traditional morphology [51], including pre-evolutionary and pre-embryological comparative approaches to homology, is epistemologically fundamental in understanding morphological organization [52]. Modern evolutionary developmental biology [53] focuses on how a certain character state is shared by a whole. Here situations are relevant where the character's state in ancestral species is significantly different from its state in the species of any form of another organism or character state. Whether the character is understood as a homology, as a traditional perspective in evolutionary biology, focuses on how a character is inherited in a single evolutionary lineage and undergoes gradual modification in its state from ancestor to descendant. The homology as the dominant perspective on homology in phylogenetic systematics [54] focuses on how a certain character state is shared by a wide range of species. However, the relationship between the character's state in ancestral species A is significantly different from its state in the species of B the character's state is not 'plesiomorphic' condition in A vs. the 'apomorphic' condition in B while all species descending from B still possess the state found in B so that this state is a novel condition that originated in B and characterizes the taxon of which B is the most recent common ancestor.

The advent of evolutionary developmental biology [55], recently developed approaches to homology have been introduced (Donoghue 1992). Typically, different biological disciplines dealing with evolutionary issues have a different perspective on homology (Brigandt 2003). While these are sometimes viewed as different interpretations that are hard to reconcile, they may very well be compatible accounts that focus on different aspects of an overall phenomenon (Brigandt 2007). To explain the main approaches, it is useful to recall the distinction between a character and a character state. A character (properly speaking) is a homologue, i.e., a morphological unit present in many individuals and which can take on a different form and function in different individuals or species. A character state is the particular condition (e.g. form) a character can take on with respect to an overall phenomenon (Brigandt 2007).

In the last few decades, novel and different theoretical approaches to homology have been proposed (Donoghue 1992). Typically, different biological disciplines dealing with evolutionary issues have a different perspective on homology (Brigandt 2003). While these are sometimes viewed as different interpretations that are hard to reconcile, they may very well be compatible accounts that focus on different aspects of an overall phenomenon (Brigandt 2007). To explain the main approaches, it is useful to recall the distinction between a character and a character state. A character (properly speaking) is a homologue, i.e., a morphological unit present in many individuals and which can take on a different form and function in different individuals or species. A character state is the particular condition (e.g. form) a character can take on with respect to an overall phenomenon (Brigandt 2007).

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