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Origination of Organismal Form: The Forgotten Cause in Evolutionary Theory

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Evolutionary biology arose from the age-old desire to understand the origin and the diversification of organismal forms. During the past 150 years, the question of how these two aspects of evolution are causally realized has become a field of scientific inquiry, and the standard answer, encapsulated in a central tenet of Darwinism, is by "variation of traits" and "natural selection." The modern version of this tenet holds that the continued modification and inheritance of a basic genetic tool kit for the regulation of developmental processes, directed by mechanisms acting at the population level, has generated the panoply of organismal body plans encountered in nature. This notion is superimposed on a sophisticated, mathematically based population genetics, which became the dominant mode of evolutionary biology in the second half of the twentieth century. As a consequence, much of present-day evolutionary theory is concerned with formal accounts of quantitative variation and diversification. Other major branches of evolutionary biology have concentrated on patterns of evolution, ecological factors, and, increasingly, on the associated molecular changes. Indeed, the concern with the "gene" has overwhelmed all other aspects, and evolutionary biology today has become almost synonymous with evolutionary genetics.

These developments have edged the field farther and farther away from the second initial theme: the origin of organismal form and structure. The question of why and how certain forms appear in organismal evolution addresses not what is being maintained (and quantitatively varied) but rather what is being generated in a qualitative sense. This causal question concerning the specific generative mechanisms that underlie the origin and innovation of phenotypic characters is probably best embodied in the term *origination,* which will be used in this sense throughout this volume. That this causal question has largely disappeared from evolutionary biology is partly hidden by the semantics of modern genetics, which purports to provide answers to the question of causation, but these answers turn out to be largely restricted to the proximate causes of local form generation in individual development. The molecular mechanisms that bring about biological form in modern-day embryos, however, should not be confused with the causes that led to the appearance of these forms in the first place. Although the forces driving morphological evolution certainly include natural selection, the appearance of specific, phenotypic elements of construction must not be taken as being caused by natural selection; selection can only work on what already exists. Darwin acknowledges this point in the first edition of *The Origin of Species,* where he states that certain characters may have "originated from quite secondary causes, independently from natural selection" (Darwin, 1859, 196), although he attributes "little importance" to such effects. In a modified version of the same paragraph in the sixth edition (Darwin, 1872, 157), he concedes that "we may easily err in attributing importance to characters, and in believing that they have been developed through natural selection."

It is the aim of the present volume to elaborate on this distinction between the origination (innovation) and the diversification (variation) of form by focusing on the plurality of causal factors responsible for the former, relatively neglected aspect, the origination of organismal form. Failure to incorporate this aspect represents one of the major gaps in the canonical theory of evolution, it being quite distinct from the topics with which population genetics or developmental genetics is primarily concerned. As a starting point, we will briefly outline the central questions that arise in the context of origination. We have identified four areas (represented by parts II–V of the book) from which the most important open questions arise: (1) the phenomenology of organismal evolution (phylogenetics); (2) genotype-phenotype relationships; (3) physical determinants of morphogenesis; and (4) the structure of the evolutionary paradigm. It will be noted that the questions that arise in each of these areas are often similar or overlapping. Indeed, the presence of recurrent themes across quite disparate subdisciplines is one important indication of the lacuna with regard to origination in the field as a whole.

Questions Arising from Phylogenetics

The evolution of organismal forms—morphological evolution—consists of the generation, fixation, and variation of structural building elements. Cell masses form microscopic structures such as spheres, cones, tubes, rods, plates, and coils. These are often branched and connected by attachments, fusions, or articulations. Such units assemble to form higher-level, macroscopic building elements, again connected to one another, resulting in the body plans of organisms that evolve further by progressive modification. This scenario raises a number of questions that relate specifically to the macroscopic features of morphological evolution. Why, for instance, did the basic body plans of nearly all metazoans arise within a relatively short time span, soon after the origin of multicellularity? Assuming that evolution is driven by incremental genetic change, should it not be moving at a slow, steady, and gradual pace? And why do similar morphological design solutions arise repeatedly in phylogenetically independent lineages that do not share the same molecular mechanisms and developmental systems? And why do building elements fixate into body plans that remain largely unchanged within a given phylogenetic lineage? And why and how are new elements occasionally introduced into an existing body plan?

Many of the phenomena on which these questions are based bear classical names (table 1.1; most "why" questions are also "how" questions here and in table 1.2), giving the issues a seemingly old-fashioned aura. But hardly any of the problems specified by this traditional terminology are explained in the modern theory of evolution. Whereas the

Table 1.1
Open questions concerning morphological evolution

Burgess shale effect	Why did metazoan body plans arise in a burst?
Homoplasy	Why do similar morphologies arise independently and repeatedly?
Convergence	Why do distantly related lineages produce similar designs?
Homology	Why do building elements organize as fixed body plans and organ forms?
Novelty	How are new elements introduced into existing body plans?
Modularity	Why are design units reused repeatedly?
Constraint	Why are not all design options of a phenotype space realized?
Atavisms	Why do characters long absent in a lineage reappear?
Tempo	Why are the rates of morphological change unequal?

classical questions refer to phenomena at the organismal level, most can also be applied to the microscopic and even to the molecular level. All are linked by one common, underlying theme: the origin of organization. The nature of the determinants and rules for the organization of design elements constitutes one of the major unsolved problems in the scientific account of organismal form. The chapters of part II explore some of the most important aspects of this problem.

Questions Arising from Genetics

A second set of open questions relates to the role of genes in the origination of biological form (table 1.2). Organismal evolution is nowadays almost exclusively discussed in terms of genetics. But are genes the determinants of form? Is it true that complete knowledge of the genetic-molecular machinery of an organism also explains how it was brought into being? That is, if we were to know all components and functions of an anonymous genome, would we be able to compute the form of its organism? And is it correct to assume that morphological evolution is driven solely by molecular evolution? Comparative evidence indicates substantial incongruences between genetic and morphological evolution, and the same genotypes do not necessarily correspond with identical phenotypes (Lowe and Wray, 1997). On the one hand, genetic and developmental pathways can change over evolutionary time even when morphology remains constant (Felix et al., 2000); on the other, similar gene expression patterns can be associated with different morphologies.

These questions converge in the second major unsolved problem of organismal form: the genotype-phenotype relationship. Now that entire genomes are mapped out and the genomic approach is seen to be unable to explain biological complexity, this problem will be a central concern of future research. Recognizing that the origination of biological form

Table 1.2
Open questions concerning the genotype-phenotype relationship in development and evolution

Jurassic Park scenario	Does the genetic code contain the complete information of organismal form?
Novelty	Do new structural elements arise from mutations?
Polyphenism	Why can identical genetic content be associated with very different morphological phenotypes?
Redundancy and overdetermination	Why are there multiple genetic and biochemical pathways to the realization of biological forms?
Discordance	Why do morphological and genetic evolution proceed at different paces?
Epigenesis	How is the genotype-phenotype relationship mediated in development?

cannot be understood solely from genetic analysis will necessarily stimulate investigation of the processes that actually construct the phenotype from materials provided, in part, by the genotype. Also, to analyze, interpret, and predict the genotype-phenotype relationship, mathematical model building and computer simulation will be essential, representing a new research approach that has been called "phenomics" (Palsson, 2000). The chapters of part III provide viewpoints on several of the problems that will have to be taken into account in future modelling approaches.

Questions Arising from Development

Two causal processes interact in the generation of organismal form: development and evolution. The new field of evolutionary developmental biology acknowledges this fact, but much work in this area proceeds under the assumption that the only important link between the two processes lies in genetics—as if the individual generation of form were merely a reading out of evolved genetic programs. However, development does not appear to behave like any program known to computer science—phenotypic outcomes persist despite extensive derangement in lines of "program code" (i.e., gene expression levels and interactions) induced by such evolutionarily unprecedented manipulations as experimental "knockouts" (Shastry, 1995) and nuclear transfer (Humpherys et al., 2001). Moreover, that genetic circuitry involved in development can undergo evolutionary "rewiring" without overt changes in the phenotype (Szathmary, 2001) suggests that phenotypes have autonomy that can trump that of the programs they supposedly express.

Epigenesis, the sum of processes that determine the transformation of a zygote into an adult phenotype poses a number of unanswered questions regarding the generation of individual forms (table 1.3). Among the most fundamental but least understood class of epigenetic factors are the physical properties of biological materials that participate in

Table 1.3
Open questions concerning epigenesis and its role in morphological evolution

Programs	Does the developmental generation of organismal form result from deterministic programs?
Context	How are developmental processes modulated by epigenetic context?
Generic Properties	What is the role of the physicochemical properties of biological materials?
Environment	What is the role of the external environment in development?

morphogenesis. How do the generic, physical properties of cell aggregates and tissues shape the constructional outcomes of development (segmentation, multilayering, body cavity formation, and so forth), and, equally important, to what extent are these same properties relevant to the origin of these forms in evolution? Although the properties are paradigmatic of the determinants that generate form, these determinants may take on different importance at different stages of evolution. The chapters of part IV deal with them individually and collectively.

Questions Arising from Evolutionary Theory

The neo-Darwinian paradigm still represents the central explanatory framework of evolution, as exemplified by recent textbooks (e.g., Mayr, 1998; Futuyma, 1998; Stearns and Hoekstra, 2000). This refined and canonical theory concerns the variational dynamics and adaptation of existing forms. It is a gene-centered, gradualistic, and externalistic theory, according to which all evolutionary modification is a result of external selection acting on incremental genetic variation. The resulting adaptations lead to successive replacement of phenotypes and hence to evolution.

Although this theory can account for the phenomena it concentrates on, namely, variation of traits in populations, it leaves aside a number of other aspects of evolution, such as the roles of developmental plasticity and epigenesis or of nonstandard mechanisms such as assimilation (table 1.4). Most important, it completely avoids the origination of phenotypic traits and of organismal form. In other words, neo-Darwinism has no theory of the generative. As a consequence, current evolutionary theory can predict what will be maintained, but not what will appear. Although recent years have seen attempts to extend evolutionary theory to organism-environment interactions (Oyama, 2000; Johnston and Gottlieb, 1990; Sober and Wilson, 1998) and self-organizing processes (Kauffman, 1993), what is still lacking is an evolutionary theory that specifically addresses the morphological aspects of evolution and integrates the interactional-epigenetic aspects with the genetic. The missing generative dimension in evolutionary theory is the subject of part V, whose

Table 1.4
Open questions concerning the theory of morphological evolution

Origination	What generative mechanisms are responsible for the origin and innovation of phenotypic characters?
Plasticity	Are developmental response capacities specifically evolved, or is plasticity a primitive property?
Epigenesis	Do the rules of developmental transformation shape evolution?
Evolvability	Is the evolutionary potential of a lineage associated with the capacity of its developmental system to respond to the environment?
Assimilation	What is the role of genetic co-optation and assimilation in the evolution of organismal form?

chapters illustrate, with specific examples across a range of morphogenetic systems, the ways in which epigenetic processes are beginning to take their place in a more complete and comprehensive evolutionary theory.

Elements of a Postgenomic Synthesis

If, as we suggest, the failure of the current theory of evolution to deal with the problem of origination is the major obstacle to a scientific understanding of organismal form, it is incumbent on us to provide at least a sketch of an alternative view. In fact, it is our contention that a synthetic, causal understanding of both the development and the evolution of morphology can be achieved only by relinquishing a gene-centered view of these processes (Newman and Müller, 2000).

Processes of natural selection can lead to morphological novelty by unleashing new epigenetic relationships (Müller, 1990; Müller and Wagner, 1996). Alternatively, they can consolidate the expression of a morphological phenotype that was previously dependent on developmental or environmental conditionalities (Johnston, Barnett, and Sharpe, 1995). In neither case does an understanding of changes in gene frequencies shed light on the evolution of forms—only on the evolution of genes. And even though hierarchical programs of gene expression often govern the sequential mobilization of morphogenetic processes in modern-day organisms, the mobilized processes are distinct from these triggering events. Again, detailed information at the level of the gene does not serve to explain form.

In the framework we propose, epigenetic processes—first, the physics of condensed, excitable media represented by primitive cell aggregates and, later, the conditional responses of tissues to each other and to external forces—replace gene sequence variation and gene expression as the primary causal agents in morphological origination. These determinants and their outcomes are considered to have set out the original, morphological templates during the evolution of bodies and organs, and to have remained, to varying extents, effective causal factors in the development of all modern, multicellular organisms (Newman and Müller, 2000).

Genetic evolution is highly suited for enhancing the reliability and inheritance of forms originally brought about by conditional processes: promotor duplication and diversification, metabolic integration, and functional redundancy can all add parallel routes to the same endpoint (Newman, 1994). By such means, the morphogenetic outcomes originated by epigenetic propensities become captured and routinized, "assimilated" (Waddington, 1961), by genetic circuitry over the course of evolution. In this view, morphological plasticity, and much of evolvability are primitive properties—the phylogenetic retention of the conditionality of the originating, epigenetic processes. At the end of long evolutionary trajectories, organisms come to embody a species-characteristic mix of conditional and programmed modes of development. Finally, in any given species the ratio of conditional to programmed determinants of morphogenesis may vary at different stages and developmental subsystems.

The view described here emphasizes the distinction between the mechanisms underlying origination and those underlying variation in morphological evolution and hence the necessity to account for that distinction in evolutionary theory. It clearly suggests that the relationship between genotype and phenotype in the earliest metazoans was different from that in their modern counterparts and that the present relationship between genes and form is a derived condition, a product of evolution rather than its precondition.

Although not all contributors to this volume would accept the most radical implications of this view, which challenges major tenets of neo-Darwinism, including its incrementalism, uniformitarianism, and genocentricity, all were invited to participate in this project because their work explicitly influenced the development of the ideas behind it. Readers will evaluate each chapter on its own terms; we hope they will also recognize a coherence that transcends the disciplinary boundaries of the contributors.

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