Evolution, Development, & The Predictable Genome

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This book is dedicated to

Paul Sherman
Charles Aquadro
Carlos Martinez del Rio
&
Michael Akam

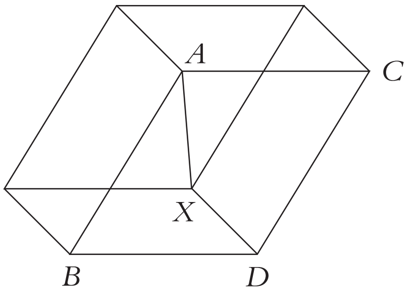
each of whom, in his own way, transformed my understanding of biological diversity

The only objections that have occurred to me are 1st that you have loaded yourself with an unnecessary difficulty in adopting 'Natura non facit saltum' so unreservedly. I believe she does make *small* jumps—and 2nd it is not clear to me why if external physical conditions are of so little moment as you suppose variation should occur at all.

—Thomas H. Huxley, in letter to Charles Darwin, upon finishing *The Origin of Species*, November 23, 1859

You have most cleverly hit on one point, which has greatly troubled me; if, as I must think external conditions produce little *direct* effect, what the devil determines each particular variation. What makes a tuft of feathers come on a Cock's head; or moss on a moss-rose?—I shall much like to talk over this with you.

—Darwin's reply to Huxley, November 25, 1859



The rhomboid *AX* is drawn so that the solid angle *A* should be seen the nearest to the spectator, and the solid angle *X* the furthest from him, and that the face *ACBD* should be the foremost, while the face *XDC* is behind. But in looking repeatedly at the same figure, you will perceive that at times the apparent position of the rhomboid is so changed that the solid angle *X* will appear the nearest, and the solid angle *A* the furthest; and that the face *ACDB* will recede behind the face *XDC*, which will come forward; which effect gives to the whole solid a quite contrary apparent inclination. . . . After . . . attentive analysis of the fact, it occurred to me, that it was owing to an involuntary change in the adjustment of the eye for obtaining distinct vision.

—Prof. L. A. Necker, "Observations on some remarkable Optical Phaenomena seen in Switzerland; and on an optical Phaenomenon which occurs on viewing a Figure of a Crystal or geometrical Solid."

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PREFACE

When a population evolves in response to natural selection, organisms change—in appearance, physiology, or behavior—over time. Colder temperatures select for thicker fur. A new pathogen selects for pathogen resistance. Faster predators select for swifter prey. Since individuals vary for many characteristics—fur thickness, pathogen resistance, running speed—some will survive and reproduce better than others in the face of ecological challenges. These "fitter" individuals contribute disproportionately to the next generation. If the differences between individuals have a genetic basis, then more individuals of the next generation will share the characteristics—thick fur, robust immune systems, fleet feet—that helped their parents to survive and to reproduce. The population has evolved.

Differences between individuals—the raw material of evolution—are caused by variation in genes and by variation in the environment. The genetic component of the variation starts as mutations. Since each aspect of an organism—fur, microbial defenses, limbs—is constructed by multiple genes, and since mutations occur randomly in the genome, we might expect that a diverse set of mutations, occurring in many of these genes, would be selected for during evolution, but this is not the case. Natural selection favors some kinds of mutations and disfavors others. This is because mutations alter organisms by changing the way that genes act during development. Some mutations may cause a single specific change in development, whereas others may cause multifarious changes. Natural selection tends to favor mutations that result in limited, specific changes. Thus, development will bias which mutations are selected.

But development is only half the story. All evolution occurs within populations, and different kinds of mutations will be selected in different kinds of populations. For example, a mutation causing a small change will be more efficiently selected in a large population than in a small population. Other aspects of population biology also influence how selection acts. Evolutionary mechanisms, like developmental mechanisms, bias which mutations are selected. Therefore, in seeking to understand how genomes evolve to generate biological diversity, we should consider both development and mechanisms of evolution.

This book presents one way to integrate evolutionary biology with developmental biology. Many other attempts at integrating evolution and development have been made, particularly in the past few decades. In contrast to most of these previous efforts, I do not discuss broad-scale patterns across large spans of evolutionary history. Instead, I focus on the individual steps of evolutionary change, as revealed by examination of variation within species and between closely related species. As William Bateson wrote in 1900, in an attempt to promote the new discipline he later called genetics: "The essential problem of evolution is how any one given step in evolution was accomplished." I explore how the mechanisms that generate each step in evolution—the process that ultimately generates differences between species—interact with development. I focus on the genetic causes of evolution and therefore rely heavily on insights from population genetics. Currently, population genetics and developmental biology are about as far from integrated as any two biological disciplines could be. This intellectual chasm probably has many causes, and it is worth comparing several salient differences between the disciplines.

Population genetics provides a framework for attempting to understand how, looking forward, the mechanisms of evolution will influence the genetic fate of populations and how, looking back, patterns observed in modern populations are best explained by the action of evolutionary mechanisms in the past. Population genetics is a bit like weather prediction. Sometimes the starting conditions can be defined precisely enough to allow accurate short-term prediction, but long-term prediction is complicated by the intercession of random events. Nonetheless, population genetics provides a guide to likely evolutionary outcomes. Similarly, meteorologists cannot say for certain where every hurricane will make landfall, but they can predict when during the year hurricanes will be prevalent and the general geographic areas the hurricanes will hit. But population geneticists rarely spend much time trying to predict the future. Population genetics is mainly about the past: What historical forces have generated current patterns of genetic variation? Are current patterns best explained by the deterministic force of natural selection, or by random processes?

Development, by contrast, is only weakly influenced by random events. Of course, random events cause some variation in development, but the scale of this variation pales in comparison to the variation caused by unpredictable events in population genetics.

Population genetics and developmental biology employ dramatically different explanatory frameworks. Explanation in population genetics proceeds from indirect inference of the causes of evolution through statistical analysis of data collected from populations. Explanation in developmental biology usually involves experiments that provide reasonably direct evidence for the causes of development.

At first glance, then, attempts to integrate development and population genetics would seem to make little sense. What benefit could possibly come from trying to integrate a very noisy process, genetic evolution in populations, with a rather predictable process, development? Will any new insights emerge when we consider simultaneously the mechanisms operating at the population level and those acting within individuals? Patterns in the currently available data suggest that genetic evolution is, to some extent, predictable and that this predictability emerges from the interplay of developmental mechanisms with the mechanisms operating within populations. It seems to me that the current data are insufficient to rigorously test this hypothesis. But relevant new data are emerging rapidly, and they will allow robust tests of the combined roles of evolution and development in generating patterns of biological diversity.

This book is written for a reader, perhaps an advanced undergraduate or a beginning graduate student, who may have little or no formal training in either evolutionary biology or developmental biology. I assume that the reader has a basic understanding of undergraduate biology, including a basic understanding of DNA, genes, and proteins. Gene names are italicized and gene products are capitalized and set in roman type. I explicitly indicate whether I am referring to a gene (the DNA sequence in the genome: "the *Frigida* gene"), a gene product (either the RNA or protein encoded by the gene: "the

Frigida gene product"), or the protein product of the gene ("the Frigida protein"). I avoid abbreviations and acronyms (with a few exceptions: DNA, RNA) in the hope of clearing away trivial roadblocks to understanding.

Chapter 1 sets out the essential questions to be addressed, and Chapter 2 explores how long-term evolutionary patterns can be explained by principles from population genetics. Each of Chapters 3 through 5 starts out by exploring a different basic genetic concept that is fundamental both to developmental biology and to population genetics. I explore how these concepts allow us to draw connections between the disciplines and how a developmental perspective and a population-genetics perspective can be mutually informative. In Chapter 6, I briefly explore the profound consequences of two seemingly banal facts. First, all populations contain a finite number of individuals; and, second, in many species, individuals tend to stay in the same general locality generation after generation. In Chapter 7, I challenge the reader to think about development in a new way, backwards, because I think this approach to development clarifies why some genes contribute disproportionately to evolution. Finally, in Chapter 8, I review observations that suggest that genetic evolution is predictable and I discuss reasons why genetic evolution might be predictable.

While the early chapters provide brief sketches of some elementary concepts from evolutionary genetics and developmental biology, it is not possible to provide a complete introduction to either discipline in a book of this length. Thus, at the end of the book, I provide a list of recommended readings that expand on topics covered in each chapter. More advanced readers may find some of the early chapters elementary, but, even in these chapters, I have attempted to illustrate connections between evolution and development that have not been discussed widely in the literature. In addition to citations, the Notes at the end of the book provide expanded discussion of some topics mentioned only briefly in the main text.