Second Edition

HUMAN BIOLOGY

An Evolutionary and Biocultural Perspective

EDITED BY SARA STINSON
BARRY BOGIN
DENNIS O'ROURKE

WILEY-BLACKWELL

HUMAN BIOLOGY ASSOCIATION

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PREFACE

This book is a collaborative effort by members of the Human Biology Association (www.humbio.org) to provide an introduction to the field of human biology. Human biology deals with understanding the extent of human biological variability, with explaining the mechanisms that create and pattern this variability, and with relating it to health, disease, and the social issues that concern all individuals today. Human biology relies heavily on an evolutionary perspective to explain variation through space and time, but also considers the effect that human culture has had on our biology, a biocultural perspective, to be crucial.

This book covers the major areas of human biology: genetic variation, variation related to climate, infectious and noninfectious diseases, stress, growth, aging, and demography. Each chapter is written by an authority in the field in order to provide expert coverage of these topics. Boxed text within the chapters explains the methods that human biologists use. Important terms are defined in the glossary, with each glossary term appearing in bold type the first time it is used in a chapter. Each chapter of this book begins with a list of “big questions” related to the topic of the chapter. It is the hope of all the chapter authors that when readers finish this text, they will be able to add their own lists of “big questions.” Indeed, perhaps readers will be able to make such lists well before completing all chapters. A set of recommended readings at the end of each chapter directs students to sources that will provide a good introduction to the topics covered in the book.

We thank the members of the Human Biology Association for their continuing enthusiastic support of this project and all of the reviewers who so generously gave of their time to review the chapters in this volume. Special thanks are due to Deb Crooks, chair of the Human Biology Association Publications Committee, for her proficient management of

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PART I
INTRODUCTION
CHAPTER 1

Human Biology: An Evolutionary and Biocultural Perspective

SARA STINSON, BARRY BOGIN, DENNIS O'ROURKE, and REBECCA HUSS-ASHMORE

INTRODUCTION

What Are the Big Questions?

What is human biology and what do human biologists study? What constitutes the shared biology of people and other nonhuman species? What are the novel characteristics of the human species, and can the time of origin and the reasons for the evolution of these new and novel features be determined? What biological differences are there among and within living human populations, and how are these differences the product of both evolution over generations and **plasticity** during an individual's lifetime? These are several of the "big questions" in the field of human biology. This book summarizes current research aimed at answering these questions.

The major points of this chapter are the following:

(1) Human biology is a well-defined discipline.

(2) Human biology is founded on an evolutionary perspective.

(3) The recognition of different types of biological adaptation, including processes of plasticity in development and behavior, is at the core of human biology.

(4) A biocultural and cross-cultural perspective is a unifying principle of all human biological research and thinking.

BOX 1.1 DEFINITIONS OF HUMAN BIOLOGY

There is no single, all encompassing definition of "human biology." This is due to the fact that the biology of the human species is studied from a variety of disciplines, each with its own perspective. These disciplines vary from the practical applications of clinical medicine for the treatment of human disease to studies to better understand the basic physiological pathways and mechanisms in the human body to research aimed at understanding the adaptive/evolutionary context of human biology. Here we offer a definition and a mission statement found in university catalogs and descriptions of the topics covered in three journals with human biology in their titles to provide a taste of the diversity of thought about human biology.

1. Loughborough University Human Biology Programme definition: "Human Biology is the study of humans from the cellular and individual level to the population level. Human Biologists study human anatomical structure and function and investigate the determinants of biological and behavioural variability in people, including genetic, environmental and cultural factors. Human Biologists study how the human species evolved, how the species changes over the lifespan, how humans adapt to external stressors, and how human biology and culture influence disease risk. Graduates go on to a diverse range of careers, including research, teaching, medicine or allied professions, laboratory work or graduate training schemes. The degree is unique for its emphasis on applied, individual and population level biology and the international perspective that is generated by staff research interests."

2. Stanford University Program in Human Biology, mission statement: "The Program in Human Biology is an interschool, interdepartmental, undergraduate major. The program's mission is to provide an

interdisciplinary approach to understanding the human being from biological, behavioral, social, and cultural perspectives. The curriculum provides a broad and rigorous introduction to the biological and behavioral sciences and their interrelationships, and explores how this knowledge, in conjunction with studies in other fields, can be applied to formulate and evaluate health, environmental, and other public policies that influence human welfare."

3. Three journals: (1) *American Journal of Human Biology*: The transdisciplinary areas covered in the journal include, but are not limited to, epidemiology, genetic variation, population biology and demography, physiology, anatomy, nutrition, growth and aging, physical performance, physical activity and fitness, ecology, and evolution, along with their interactions. (2) *Annals of Human Biology*: A journal of human population biology, reporting investigations on the nature, development, and causes of human variation, embracing the disciplines of human genetics, auxology, environmental physiology, ecology, and epidemiology. (3) *Human Biology*: A worldwide forum for state-of-the-art ideas, methods, and techniques in the field, *Human Biology* focuses on genetics in its broadest sense. Included under this rubric are human population genetics, evolutionary and genetic demography, quantitative genetics, evolutionary biology, ancient DNA studies, biological diversity interpreted in terms of adaptation (biometry, physical anthropology), and interdisciplinary research linking biological and cultural diversity (inferred from linguistic variability, ethnological diversity, archaeological evidence, etc.).

In this chapter, we introduce the subject of this book, human biology, and the evolutionary and biocultural perspective that human biologists use in their work. While there are a number of disciplines that could (and some do) call themselves human biology because they deal with human biological characteristics, the human biology covered in this book is the discipline concerned with variation in biological traits both among and within living human populations and understanding the origin, maintenance, and implications of this variation. Human biologists investigate the genetic, environmental, and cultural determinants of biological variability in living people. They study how the human species evolved, how individual humans change over the lifespan, how humans

adapt to external **stressors**, and how human biology and culture interact to shape disease risk.

Human biologists' primary interest is in biological, as opposed to behavioral, characteristics. Among the main topics that human biologists study are variation in genetic traits, disease, health, nutrition, climate responses, growth, aging, and **demography**. One important feature of human biology is its interest in *all* human populations. This interest reflects the fact that most human biologists are trained as anthropologists (especially in the United States), and like anthropologists, human biologists often study remote groups whose lives are very different from those of most of the readers of this book. For example, the authors of the chapters in this book have conducted fieldwork in Alaska, Dominica, Mexico, Guatemala, Ecuador, Peru, Bolivia, Kenya, Zaire, Egypt, Tibet, Siberia, China, The Philippines, and Samoa. But human biologists also study populations in industrialized countries, and you will see many examples of this research in this book. Human biologists study populations around the world because they are interested in understanding the effects of the many different environments with which humans must cope, and are often particularly interested in responses to severe environmental stressors such as the extreme cold in Alaska and Siberia or the very high altitudes in Peru and Bolivia.

Because human biologists frequently collect data in the field, meaning outside the laboratory or hospital setting, some traits are more feasible for them to study than others. It would be very difficult (and expensive) to conduct research using CT scans on a large portion of the world's populations. On the other hand, the instruments for measuring height and weight can be transported relatively easily to even the most remote location. As you read about human biology research, you will see the emphasis we place on developing methods that can easily be used in the field.

Over the last several decades, new data collection and analysis methods have greatly increased the questions that human biologists can answer. There are now smaller instruments such as portable heart rate monitors and accelerometers to measure energy expenditure; collection methods that do not require access to electricity, such as measuring **hormones** from saliva rather than from whole blood; and techniques that reduce the burden on the research participants, such as analysis of blood **proteins** from spots of blood, from a finger prick, dried on filter paper, rather than from blood drawn from a vein.

Human biologists study individuals, but their primary interest is in the characteristics of groups of individuals, called populations; in fact, the discipline is sometimes called human population biology (Baker 1982; Little and Haas 1989). The importance of populations to the human biologist is illustrated by comparing human biology with Western medicine (frequently called biomedicine), another discipline that is concerned with human biological traits. Both biomedical doctors and human biologists are interested in the biological characteristics of groups, but the main reason for this interest is different. In biomedicine, knowing the blood pressure of an *individual* is important mainly because it can be used to determine if the value is outside the normal clinical range, and thus if the patient is ill and in need of medical treatment. The “normal clinical range” is that found for people *within* the clinical population. In the industrialized Western nations of the United States, Canada, the European Union, Australia, and Japan, the clinical population is usually comprised of men of middle and upper **socioeconomic status**. Women, children, and ethnic minority groups are often not well represented in clinical reference values, even though there has been an effort to increase the participation of women and minorities in recent years (Department of Health and Human Services

1994; http://grants.nih.gov/grants/funding/women_min/guidelines_amended_10_2001.htm; Murthy et al. 2004). In the United States, the National Institutes of Health has explicit guidelines about including women and minorities in studies. Despite these efforts, there has been only limited improvement in the representativeness of biomedical research. Human biologists, on the other hand, are interested in knowing the average blood pressure and range of variation of *populations* to be able to compare values among and within groups, and to use these comparisons to make statements about population variation. Groups of nonindustrialized people, the hunter–gatherers, horticulturalists, pastoralists, and traditional agriculturalists studied by anthropologists/human biologists often have lower blood pressure than people in Western, industrialized nations (see Chapter 13).

Human biologists' interest in comparison leads naturally to the questions of how variation arises, why characteristics do or do not persist in given situations, and what are the larger implications of human biological diversity. To understand how human biologists go about answering these questions, we need to look at the explanatory framework that human biologists use: an evolutionary and biocultural perspective. As Peter B. Medawar (1964), who was awarded the Nobel Prize for his work on tissue grafting and was an important theorist of aging, wrote, "Human Biology is not so much a discipline as a certain attitude of mind… ."

EVOLUTIONARY PERSPECTIVE

Human biologists need to account for biological change over time as well as the distribution of traits in space. As a result, human biologists—like most other biological scientists—use

the synthetic theory of evolution as their primary explanatory framework.

What do we mean by the “synthetic” theory of evolution? This term refers simply to the fact that the modern theory of evolution is a synthesis of Darwinian theory (Charles Darwin, 1809–1882, published *Origin of Species* in 1859) and the science of genetics. Darwinian theory revolves around the principles of **natural selection**. At its simplest, Darwin’s theory has four basic tenets: (1) More organisms are produced than can survive; (2) organisms within a species vary in their traits; (3) some of this variation is heritable; and (4) variants best suited to the environment survive to be represented in the next generation. Mendelian genetics (Gregor Mendel, 1822–1884, proposed that inherited traits are discrete particles) provided a plausible explanation of how variation is inherited. Molecular biology and **cytogenetics** of the 20th century clarified how variation arises at the level of the deoxyribonucleic acid (**DNA**) molecule (genetics is explained in more detail in Chapter 3). Another way to explain natural selection using the language of genetics would be (1) changes in DNA can produce phenotypic changes that are subject to natural selection; (2) **phenotypes** best suited to the environment are most likely to survive and reproduce; (3) phenotypes with greater reproductive success leave more of their **genes** to the next generation; and (4) a change in **allele** frequencies from one generation to the next is defined as evolution.

Natural selection is central to human biology because changing environments over time, as well as the diversity of environments experienced by contemporaneous populations, challenge phenotypes leading to differential reproductive success of their **genotypes**. The classic example of natural selection leading to human variation is that of falciparum malaria and hemoglobin S, the protein

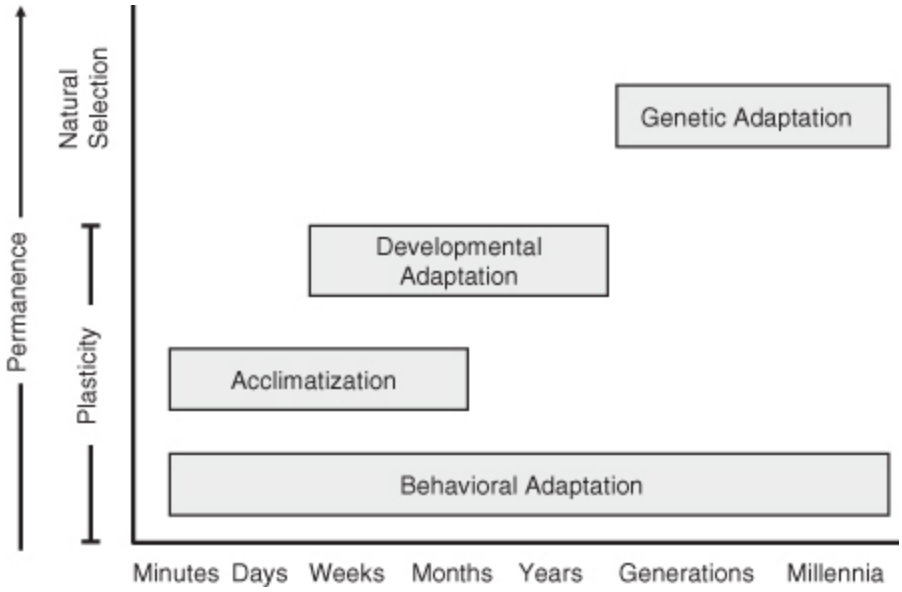
responsible for sickle-cell disease. Populations experiencing high levels of falciparum malaria (the most deadly form of malaria) have high frequencies of the hemoglobin S allele because **heterozygotes** have an advantage in **endemic** malarial environments. Individuals with a genotype of one hemoglobin A (the most common or “normal” hemoglobin allele) and one hemoglobin S allele (AS genotype) have increased resistance to falciparum malaria and do not suffer the often fatal effects of the sickle-cell disease that occur in **homozygotes** for hemoglobin S. In populations without falciparum malaria, there is no advantage to the hemoglobin S allele, which, without medical treatment, is usually lethal to children with the homozygous SS genotype. If these children die before reproducing, then both S alleles are lost to the population and the frequency of the S allele declines over time in the population. Diversity in responses to climate extremes (see Chapter 6) and diet (see Chapter 7) provide other well-supported examples of the action of natural selection in producing human variation.

Natural selection results in adaptation. The type of adaptation that results from natural selection is called a **genetic adaptation** because the differential reproductive success that is the basis of natural selection causes changes in the frequency of alleles. As we will see below, there are other types of adaptations as well (see Fig. 1.1). By adaptation we mean a beneficial adjustment to the environment. To be considered adaptive, the benefits of the trait must be greater than its costs, but that does not mean that adaptations are always totally without cost. Hemoglobin S in malarial environments is an excellent example of this. While having the hemoglobin S allele is beneficial overall because heterozygotes have a reproductive advantage, hemoglobin S also has a cost as a result of early **mortality** from sickle-cell disease in those who inherit an S allele from each parent (SS homozygotes).

Sickle cell also illustrates that most traits are only adaptive in particular environments: Available evidence suggests that the S allele is advantageous only where there is falciparum malaria.

Figure 1.1 Modes of adaptation showing the different timescales at which different types of adaptations occur. Acclimatizations and developmental adaptations occur during the lifetime as a result of plasticity, and can take from minutes to years to occur. Biological plasticity can involve physiological, morphological, and epigenetic changes. Humans also exhibit a high degree of behavioral plasticity, which can lead to adaptations over the short or long term. Genetic adaptations occur over generations as a result of natural selection.

Adapted from a figure created by Jodi Lyons and Cynthia Beall, which was adapted from Gluckman et al. (2009b) and Thomas (1975).



With a few exceptions, it has been difficult to demonstrate unequivocally the operation of natural selection in humans. At the molecular level, there are now techniques for inferring whether selection has acted on a DNA sequence (Harris and Meyer 2006), and as discussed in Chapter 6, evidence for selection has been found for a number of genes affecting high-altitude adaptation. But for most of our

phenotypic traits, genes interact with each other and with the environment, so the correspondence between genotype and phenotype is not one to one (Kimura 1979). In addition, often no easily measurable relationship exists between either genotype or phenotype and reproductive success. The long generation time of humans makes us a difficult species in which to document differential survival and reproduction (or differential contributions of genes to the next generation). The difference in the number of offspring between high-altitude Tibetan women estimated to have different oxygen saturation genes is one of the rare cases in which we have come close to measuring differences in reproductive success (see Chapter 6), but much of human biological research uses other, more **proximate** indicators of probable adaptive success (health, growth, work capacity, etc.).

Natural selection is frequently considered the most important mechanism of evolution, although there is no universal agreement on this point. (Sewall Wright [1982], for example, argued for a prominent role for **stochastic** gene changes, meaning random changes in allele frequencies in populations of small **effective** size.) Regardless of which side of this debate one favors, it is important to remember that there are forces other than natural selection that lead to allele frequency change over time. It is generally accepted that four basic mechanisms can change the frequency of alleles and genotypes within a population: natural selection, **mutation**, **genetic drift**, and **gene flow**. (These forces of evolution are described in more detail in Chapter 4.)

Mutation is the ultimate source of genetic variation, through the alteration of **bases** in the DNA molecule. Mutation provides the raw material on which natural selection can operate. Because mutations arise by chance, different mutations are likely to occur in different

populations, and this can be a cause of population variation. Random occurrence of different mutations is one possible explanation for why the genes conferring the ability to digest the milk sugar **lactose** in adulthood are not the same in all populations (see Chapter 7) or why the mechanisms of adaptation to high altitude are not identical in Himalayan and Andean populations (see Chapter 6).

Genetic drift refers to stochastic changes in allele frequencies, such as the one person with a particular allele being eaten by a predator or killed in a motor accident. Random change is likely to have larger effects in small isolated populations, where a given allele may be introduced and retained (or eliminated) by chance. In small populations, the loss of an individual and his or her genes could significantly reduce the overall genetic variability for the next generation. Genetic drift also operates in large populations, but its effects are so small as to be effectively unnoticeable. Because humans lived in small populations for most of our evolutionary history, genetic drift was likely a much more important cause of evolution in the past than it is today.

Gene flow is the exchange of genetic material between populations through the processes of **migration** and mating. In human populations, mobility and intermarriage have probably always been important means of maintaining genetic diversity. Historical forces such as droughts, wars, economic alliances, international trade, and colonialism have influenced the rate and location of gene exchange. Since the 19th century, global travel and population contact have undoubtedly increased rates of gene flow and thus are important mechanisms of evolution.

Mutations alone are too rare to cause perceptible changes in allele frequencies, but the frequencies of new mutations can be greatly affected by stochastic processes such as genetic drift and, as noted above, enhanced or eliminated