

CHAPTER 1

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INTRODUCTION

The year 2000 marked what is now being called “the century of the gene” (Keller, 2000), and 2003 will mark the fiftieth anniversary of the “discovery” of the double helix. One hundred years after the so-called rediscovery of Gregor Mendel’s laws of inheritance, the science of genetics has developed to the point where a working draft of the entire sequence of the human genome is now available. These advances have brought with them numerous challenges and opportunities related to our understanding of genetics and its relation to medicine, ethics, and broader social practices and institutions. Scholarly and popular literature addressing these issues has burgeoned, with numerous important contributions being made to the history of the gene (e.g., Keller, 2000; Morange, 2001) and to our philosophical understanding of the gene concept (e.g., Beurton, Falk, and Rheinberger, 2000), as well as to continued exploration of ethical implications of this new genetic age (e.g., to name just two, Buchanan *et al.*, 2000; Andrews, 2001). These dialogues have occurred somewhat in isolation from each other, however, without much explicit effort to juxtapose historical, conceptual, and ethical analyses of the ever-mutating concept of the gene and related topics, or to examine the influence these analyses exert on the disciplines, fields, and sectors of science and society likely to be affected by these changing concepts.

Our intent in crafting this collection was to reflect the spirit of the series in philosophy and medicine to which it belongs; in particular, we have interpreted in a broad way that which counts as a philosophical contribution to the understanding of medicine with the hope of continuing and reinvigorating productive communication among the many constitutive subdisciplines of philosophy and medicine. With examination of the evolution of concepts related to medical genetics as its centerpiece, this volume also reflects our desire, as teacher-scholars working in interdisciplinary settings, to prompt more dialogue between scholars working in the

history and philosophy of biomedical sciences and in bioethics and to provide additional cross-disciplinary teaching resource materials.

The collection's first section, which is primarily historical, focuses on key concepts in human genetics and on their origins; hence it engages deep epistemological issues. Garland Allen's contribution provides the backdrop for many of the subsequent chapters by its exploration of the development of the classical gene—or what he argues should be more appropriately considered to be the 'derived Mendelian' gene—from the rediscovery of Mendel's work in 1900 through the mid-1930s. He denies that this concept of the gene has been abandoned, and argues that today's gene concept is still dominated by these classical concerns, largely as promoted in textbooks and popular literature. He traces the roots of the classical gene concept to the historical development of biology as a discipline seeking to emulate physics and chemistry and to the influence of mechanistic materialism on the development of genetics as a separate discipline. Douglas Allchin provides a more explicitly negative assessment of the impact of supposedly Mendelian concepts on current-day genetics, arguing forcefully in favor of eliminating the concept of dominance. The concept, he claims, is inessential to genetics and misleading in terms of heredity, natural selection, and various molecular and cellular processes. Since clearer language is available to us, we should avail ourselves of it. His examination of key disease concepts in molecular genetics reveals that the concept of dominance has come undone, even at the basic level of classical genetics where it might have been thought to still have force. Allen and Allchin's chapters complement each other strongly, and should be of interest not only to historians of biology, but to anyone who is tempted to continue to use the classical gene concept in teaching or scholarship.

Manfred D. Laubichler and Sahotra Sarkar explore the intellectual history and current social context of the genetic concepts of penetrance and expressivity. In an account with many unexpected historical twists, they argue that the introduction of these terms was motivated by the interaction of research programs in human neuroanatomy and in the evolutionary genetics of fruitflies, together with a project to establish the source of the genius embodied in V.I. Lenin's preserved brain. Their examination reveals that the vision of the discipline of neurobiology endorsed by particular scientists at the time made it inevitable that these two fields would intersect, despite their apparent incongruity. In addition, pursuing the theme introduced by Allen and Allchin—the problematic nature of long-standing concepts in genetics—Laubichler and Sarkar discuss 'penetrance' in contemporary human behavioral genetics, in light of the influence of ideological factors as well as conceptual considerations. As they put it, our post-Human Genome Project era is powered by an atmosphere of 'geneticism,' or what elsewhere has been termed 'geneticization' (Hubbard, 1990; Lippman, 1991, esp. pp. 18-19; Wolf, 1995): there is now a gene for everything. 'Penetrance' serves to mask the difficulties in maintaining this claim, much as Allchin argues the concept of dominance serves to obscure the biological details inherent in current-day medical genetics.

Using the history of genetics and the history of bioethics, Diane Paul's contribution examines the transformation in concepts associated with reproduction from the early 1900s. Focusing on 'reproductive responsibility' and its seeming disappearance from public discourse by the mid-1970s in favor of 'reproductive autonomy,' she argues that although public discourse indeed was affected, the explicit change in concepts masks what was actually continuity in the underlying norms. The changes that did occur through various sociopolitical movements in the 1960s and 1970s did not play out in the same way across different classes and different professions. She concludes that the call for responsibility in reproduction was a central focus throughout the twentieth century, even in the recent era apparently dominated by concern for autonomy. We should be attentive to the intricacies in such historical stories when assessing apparent changes in scientific and bioethical worldviews, particularly with regard to fundamental concepts such as freedom, autonomy, and responsibility.

The second section is explicitly devoted to perspectives from the philosophy of science, but its contributors use interdisciplinary techniques to ground their discussions. Taking up the theme of geneticization introduced in the first chapter, and focusing continued attention on the concepts of penetrance and expressivity, Fred Gifford draws attention to the problems inherent in our current understanding of causation in genetics, and particularly in the genotype-phenotype relation. He argues that formulations of questions and arguments in bioethics and health policy can be criticized because of flaws in the implicit understandings of various conceptual matters being invoked, a premise that is shared by the underlying vision motivating this collection. Pointing to difficulties involved in isolating both particular traits and the genetic contributions to them, as well as to issues raised by the need to take environmental contributions to so-called genetic diseases more seriously, Gifford presents a series of challenges for medical genetics, both current and future, in light of the increasing geneticization of medicine. Rachel A. Ankeny develops the theme of the need to contextualize the implications of advances in medical genetics within the philosophy of medicine to argue that our vision of particular diseases, and of disease in general, will be significantly altered with the availability of genetic sequencing information. As a consequence, the traditional debates in the philosophy of biomedical sciences about reduction of classical genetic concepts to molecular ones must be recast, with more attention being given to holistic views of disease processes involving a variety of biological and conceptual levels. Hence her chapter is a call to resist geneticization rather than to assume its inevitability, given the recent advances in genetics.

Joseph L. Graves, Jr., addresses reductionism and adaptationism as ideologies against the backdrop of traditional medical research into the relationships between the supposed biological category of 'race' and disease prevalence. He argues that historically a variety of conceptual errors have occurred in such research, and that effective and ethical genetic analysis of disease and human genetic variation can only be pursued if sound evolutionary and population genetic thinking is integrated into medicine. At least one concrete outcome of this argument is that the focus of

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