教育部高等教育司推荐
国外优秀生命科学教学用书

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ESSENTIALS OF GENETICS

Seventh Edition

遗传学基础

（第7版）（影印版）

William S. Klug
Michael R. Cummings
Charlotte A. Spencer
Michael A. Palladino

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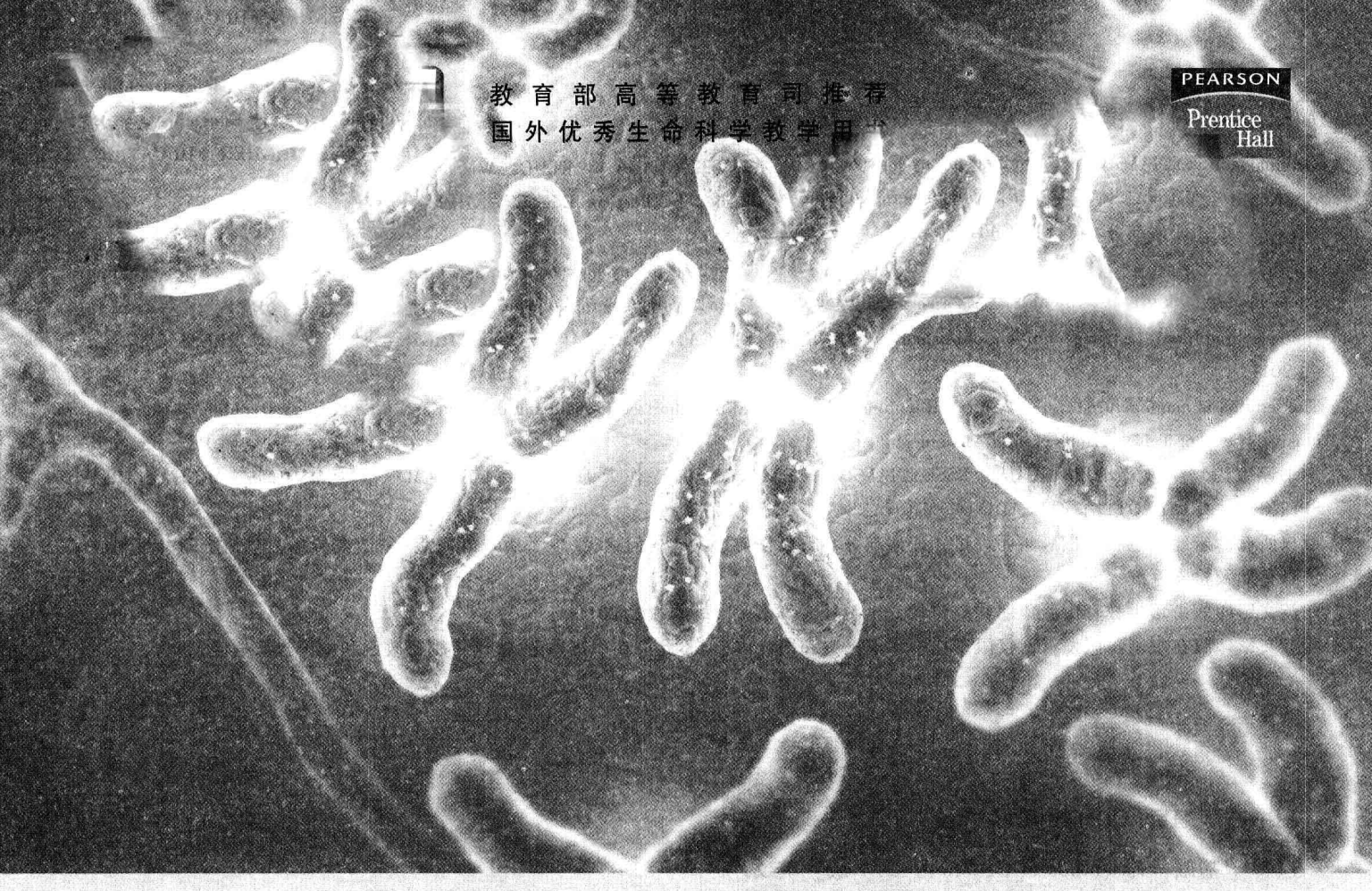

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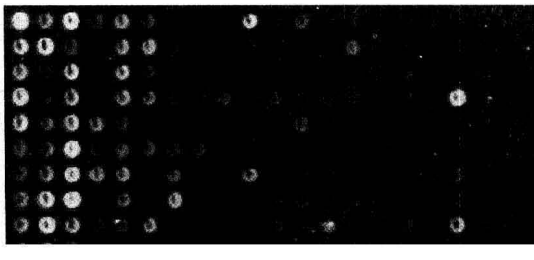

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Michael R. Cummings is currently Research Professor in the Department of Biological, Chemical and Physical Sciences at Illinois Institute of Technology, Chicago, Illinois. For more than 25 years, he was a faculty member in the Department of Biological Sciences and in the Department of Molecular Genetics at the University of Illinois at Chicago. He has also served on the faculties of Northwestern University and Florida State University. He received his B.A. from St. Mary's College in Winona, Minnesota, and his M.S. and Ph.D. from Northwestern University in Evanston, Illinois. In addition to ***Essentials of Genetics*** and its companion volumes, he has also authored several texts in human genetics and general biology for nonmajors. His research interests center on the molecular organization and physical mapping of the heterochromatic regions of human acrocentric chromosomes. At the undergraduate level, he teaches courses in Mendelian and molecular genetics, human genetics, and general biology, and has received numerous awards for teaching excellence given by university faculty, student organizations, and graduating seniors.

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Michael A. Palladino is Dean of the School of Science and Associate Professor of Biology at Monmouth University in West Long Branch, New Jersey. He received his B.S. degree in Biology from Trenton State College (now known as The College of New Jersey) and his Ph.D. in Anatomy and Cell Biology from the University of Virginia. He directs an active laboratory of undergraduate student researchers studying molecular mechanisms involved in innate immunity of mammalian male reproductive organs and genes involved in oxygen homeostasis and ischemic injury of the testis. He has taught a wide range of courses for both majors and nonmajors and currently teaches genetics, biotechnology, endocrinology, and laboratory in cell and molecular biology. He has received several awards for research and teaching, including the 2009 Young Investigator Award from the American Society of Andrology, the 2005 Distinguished Teacher Award from Monmouth University, and the 2005 Caring Heart Award from the New Jersey Association for Biomedical Research. In addition to *Essentials of Genetics* and its companion volumes, he is coauthor of the undergraduate textbook *Introduction to Biotechnology,* Series Editor for the Benjamin Cummings Special Topics in Biology booklet series, and author of the first booklet in the series, *Understanding the Human Genome Project.*

前　言

《遗传学基础》是应课程设置的需要而编写的，与内容更为广泛的姊妹篇《遗传学概念》相比，它更为短小，也更为基础，但仍然覆盖全面，内容新颖，尤其适合于低年级生物专业的学生以及农业、化学、工程、林业、心理和野生生物保护等专业的学生。由于篇幅比其他很多书都短，本书更适合于单季课程和一学期课程。

目标

本书的目标与前几版相同，明确地说，就是：

- 强调概念，而不是过多的细节；
- 通过清楚、直接的描述，提供给学生关于复杂主题的易于理解的诠释；
- 在章节内部和章节之间建立细致的组织架构；
- 传播遗传学的丰富历史，完美阐述在学科形成和发展过程中，信息如何获得和扩充；
- 强调问题的解决，引导学生分析思考，应用和扩充他们的遗传学知识；
- 提供本领域最新的、最前沿的知识；
- 绘制引人入胜的，同时有助于教学的全彩色图，再加上同样有益的照片，为概念的发展提供支持；
- 提供出色的在线资源指南，引导学生通过动画、指导练习及自测工具来理解重要的概念。

这些目标是《遗传学基础》的基石，为达成这些目标，就要求教科书适应不同形式的课程安排。尽管本书提供了一个联成一体的内容列表，它代表了一种遗传学教学安排，但各章的编写是相互独立的，允许教员打乱原来的顺序自主安排。

第7版的新特点

第7版有很多新的重要特征，连同其新颖的内容，使得本书更加适于学习遗传学的学生。本书的这些改进，建立了一个促进学生更加深入思考的平台，使得他们对刚刚学过的知识有更加全面深入的理解。

探索基因组　第12章的“探索基因组”是第7版的新内容，它对遗传学的大多数领域都持续产生影响。每一条目都要求学生进入一个或多个公众资源库和数据库中与基因组相关的网站，通过交互演练，以确保熟悉可用的基因组和蛋白组信息类型。“练习”指导学生如何搜索特定的题目以及如何进入重要的数据库；“问题”引导学生做进一步探索，学着自己应用那些网站。尤其重要的是，“探索基因组”部分将基因组的信息融合到整个书中，为学生在课上或课下的学习提供了基础。

案例分析　每章都有一个新栏目，叫做“案例分析”，作为课堂交流的提高内容。每一栏目都提供了一个与本章某一项内容相联系的案例，并就案例提出问题。这就要求学生把新获得的知识应用于实际生活中。在教学中可以采取小组讨论或个人作业的方式。

主要知识点　在概括每章所有主要内容的总结中，我们新增加了一项内容，叫做“主要知识点”。这些知识点都在各章中频繁出现，言简意赅地概括了各章的主旨。把这些知识点整合到各章，它们就可以作为学生阅读或复习各章内容的“路标”。

遗传学、技术和社会短文　第7版中我们继续提供一些针对性很强的短文，但是以一种新的交互方式呈现。在每一篇短文后，有一项新内容叫做“轮到你了”，其中向学生提出一些问题，并同时提供各种资源以帮助他们解答问题。这种改进提供了另外一种加强课堂交流的方式。所有以前的短文主题都经过了修订和更新，并加入了两篇新短文：“唐氏综合征，产前诊断和新优生学”（第6章）和“1000美元的基因组——个人基因组计划和种族”（第19章）。

主题现代化　如同每次修订一样，我们的主要努力都放在书中各方面内容的更新上，尤其是基因组学和蛋白组学方面（第18章），以及分子遗传学的各领域，包括基因表达调控（第15章）和癌的遗传学（第16章）。而Sarah Ward又一次加入了保护遗传学（第24章）的内容。

新章节　我们时刻关注遗传学中新近出现的相应领域，由此我们增添了新的一章——“遗传学与行为”（第21章）。这一章反映了我们对基因在生物体行为的方方面面所起作用的不断增加的认识。学生会对这一章很感兴趣，因为围绕这个问题的发现与我们对自身的认识有关。

更新的教师和学生资源　教师和学生资源都已更

新，以反映新版的变化。对讲授和其他教学活动的支持包括：书中图片和表格的电子版、教学资源 DVD 中大量的各种 ppt。经过更新后的配套网站上的资源，反映了正在形成的一个共识，即今天的学生应该尽可能明智地利用他们有限的学习时间。学生网站也提供了引导学生去做概括、比较和评估的一些问题。

重视概念和问题的解决

《遗传学基础》关注于遗传学的概念以及通过解决问题加深对这些概念的理解。我们的经验显示，首先关注于基本概念的那些学生，更容易领会和掌握随后课程中遗传学的重要思想。最重要的，将概念阐述和解决问题联系起来，得以增强生物学的解析观念。为帮助学生确定重要主题所涉及的概念，每一章均以一个称作“本章概念”的小节开始，列出了即将出现的最重要的概念。

另外一个有价值的教学特色就是在第 6 版首次引入每一章中的“我们如何得知”。这些条目要求学生通晓每章中出现的最重要生物学概念的实验基础。“科学是求知之路”作为生物学学习的延伸，加强学生对每一章许多关键主题的理解。

“现在来解决问题”这个条目在每章都数次出现，它要求学生把理解概念和解决问题更及时地联系起来。每一个都将学生引向本章结尾的一个问题，每个问题都与课文中的讨论密切相关。

此外，每一章都以“观察与解答”这个最受欢迎又行之有效的小节作为结语。本小节提供例题和解析，对如何做遗传分析进行示范，帮助学生形成分析的思维以及实验推理能力。消化“观察与解答”中的信息，为学生进入每章最后那较难的“问题和讨论议题”一节做了先期准备。在最后这节，我们给出了复习本章各个主题的议题和要求学生进行分析思考与遗传概念应用的问题。问题的难度逐级增加，越往后的问题要求越高。

致谢

审稿人员

所有的综合性教科书，都有赖于许多同仁的倾力投入。以下人员给予本书第 7 版宝贵的建议和建设性批评或/和意见：

Althea K. Alton, *Western Illinois University*

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Tatiana Tatum, *Saint Xavier University*

Paul Wilson, *Trent University*

对书中的任何错误，我们负全部责任。我们衷心感谢以上人员的帮助。

贡献人员

我们首先特别感谢为本书作出直接贡献的同仁。特别感谢科罗拉多州立大学的 Sarah Ward 写了保护遗传学一章，新泽西大学的 Sudhir Nayak 写了基因组的内容，东密歇根大学的 David Kass、耶鲁医学院的

Katherine Uyhazi 和 North Hennepin Community 大学的 Tamara Mans 对遗传学、技术、社会短文部分的诸多贡献；亚利桑那州立大学的 Elliott Goldstein 对分子遗传学方面的投入，LightCone Interactive 的 Mike Guidry 以及田纳西大学的 Karen Hughes 在媒体程序方面贡献颇多。我们也特别感谢最近刚从 Creighton 大学退休的 Harry Nickla，在他所作的《学生手册》和《教员指南》中，写了很多新题目，并给出了选定题目的答案（见附录 A）。

我们衷心感谢上述同仁，不仅在于分享他们的遗传学专业知识，而且在于他们对本书的贡献以及与他们互动的美好经历。

编辑和出版人员

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校对长达 600 页的教科书，对其贡献的感激之情不是言语所能表达的。我们最深的感谢献给 4 位人士，他们以耐心和勤奋完成了这个工作。他们是 Michael Rossa，Susan Baglino，Cheryl Ingram-Smith 和 Thomas H. Alton。此外，Betty Pessagno 负责编辑整个手稿，她对整部书的质量所作出的贡献值得特别感谢。

将上述各位的工作整合起来，并非凡人可以为之，我们有幸拥有一位天才 Dusty Friedman——我们的策划编辑。她不但使工作顺利进行，而且一直秉承愉悦有序的风范。

如同上述的致谢所清楚体现的那样，本书是集体智慧的结晶，上面的所有人都值得分享本书付梓的喜悦。我们不吝篇幅来叙述各位的贡献，仅以此表明我们对他们每个人都一样深怀感谢之情。

（佟向军　译）

Brief Contents

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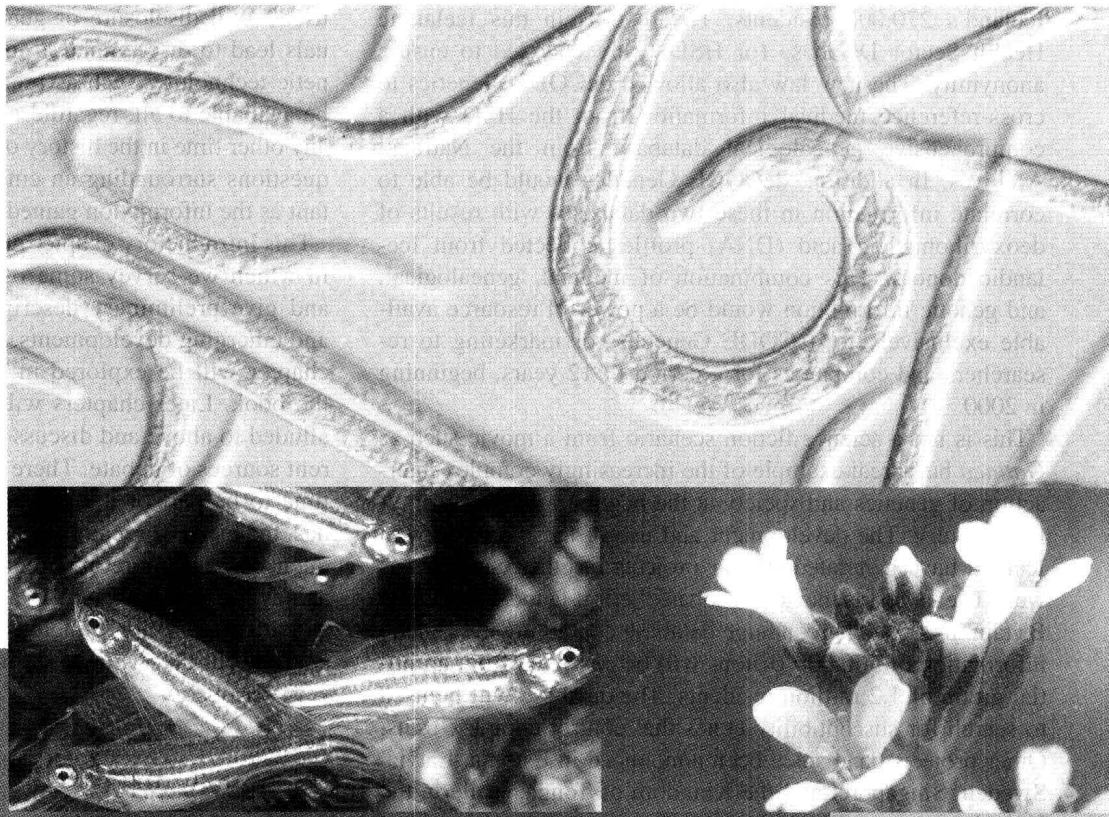

Newer model organisms in genetics include the roundworm C. elegans, the plant A. thaliana, and the zebrafish, D. rerio.

1

Introduction to Genetics

Chapter Concepts

- Transmission genetics is the general process by which traits controlled by factors (genes) are transmitted through gametes from generation to generation. Its fundamental principles were first put forward by Gregor Mendel in the mid-nineteenth century. Later work by others showed that genes are on chromosomes and that mutant strains can be used to map genes on chromosomes.
- The recognition that DNA encodes genetic information, the discovery of DNA's structure, and elucidation of the mechanism of gene expression form the foundation of molecular genetics.
- Recombinant DNA technology, which allows scientists to prepare large quantities of specific DNA sequences, has revolutionized genetics, laying the foundation for new fields—and for endeavors such as the Human Genome Project—that combine genetics with information technology.
- Biotechnology includes the use of genetically modified organisms and their products in a wide range of activities involving agriculture, medicine, and industry.
- Some of the model organisms used in genetics research since the early part of the twentieth century are now used in combination with recombinant DNA technology and genomics to study human diseases.
- Genetic technology is developing faster than the policies, laws, and conventions that govern its use.

In December 1998, following months of heated debate, the Icelandic Parliament passed a law granting deCODE Genetics, a biotechnology company with headquarters in Iceland, a license to create and operate a database containing detailed information drawn from medical records of all of Iceland's 270,000 residents. The records in this Icelandic Health Sector Database (or HSD) were encoded to ensure anonymity. The new law also allowed deCODE Genetics to cross-reference medical information from the HSD with a comprehensive genealogical database from the National Archives. In addition, deCODE Genetics would be able to correlate information in these two databases with results of deoxyribonucleic acid (DNA) profiles collected from Icelandic donors. This combination of medical, genealogical, and genetic information would be a powerful resource available exclusively to deCODE Genetics for marketing to researchers and companies for a period of 12 years, beginning in 2000.

This is not a science fiction scenario from a movie such as *Gattaca* but a real example of the increasingly complex interaction of genetics and society at the beginning of the twenty-first century. The development and use of these databases in Iceland have generated similar projects in other countries as well. The largest is the "UK Biobank" effort launched in Great Britain in 2003. There, a huge database containing the genetic information of 500,000 Britons will be compiled from an initial group of 1.2 million residents. The database will be used to search for susceptibility genes that control complex traits. Other projects have since been announced in Estonia, Latvia, Sweden, Singapore, and the Kingdom of Tonga, while in the United States, smaller-scale programs, involving tens of thousands of individuals, are underway at the Marshfield Clinic in Marshfield, Wisconsin; Northwestern University in Chicago, Illinois; and Howard University in Washington, D.C.

deCODE Genetics selected Iceland for this unprecedented project because the people of Iceland have a level of genetic uniformity seldom seen or accessible to scientific investigation. This high degree of genetic relatedness derives from the founding of Iceland about 1000 years ago by a small population drawn mainly from Scandinavian and Celtic sources. Subsequent periodic population reductions by disease and natural disasters further reduced genetic diversity there, and until the last few decades, few immigrants arrived to bring new genes into the population. Moreover, because Iceland's health-care system is state-supported, medical records for all residents go back as far as the early 1900s. Genealogical information is available in the National Archives and church records for almost every resident and for more than 500,000 of the estimated 750,000 individuals who have ever lived in Iceland. For all these reasons, the Icelandic data are a tremendous asset for geneticists in search of genes that control complex disorders. The project already has a number of successes to its credit. Scientists at deCODE Genetics have isolated genes associated with 12 common diseases including asthma, heart disease, stroke, and osteoporosis.

On the flip side of these successes are questions of privacy, consent, and commercialization—issues at the heart of many controversies arising from the applications of genetic technology. Scientists and nonscientists alike are debating the fate and control of genetic information and the role of law, the individual, and society in decisions about how and when genetic technology is used. For example, how will knowledge of the complete nucleotide sequence of the human genome be used? Will disclosure of genetic information about individuals lead to discrimination in jobs or insurance? Should genetic technology such as prenatal diagnosis or gene therapy be available to all, regardless of ability to pay? More than at any other time in the history of science, addressing the ethical questions surrounding an emerging technology is as important as the information gained from that technology.

This introductory chapter provides an overview of genetics in which we survey some of the high points of its history and give preliminary descriptions of its central principles and emerging developments. All the topics discussed in this chapter will be explored in far greater detail elsewhere in the book. Later chapters will also revisit the controversies alluded to above and discuss many other issues that are current sources of debate. There has never been a more exciting time to be part of the science of inherited traits, but never has the need for caution and awareness of social consequences been more apparent. This text will enable you to achieve a thorough understanding of modern-day genetics and its underlying principles. Along the way, enjoy your studies, but take your responsibilities as a novice geneticist very seriously.

1.1 Genetics Has a Rich and Interesting History

We don't know when people first recognized the existence of heredity, but archeological evidence (e.g., primitive art, preserved bones and skulls, and dried seeds) documents the successful domestication of animals and cultivation of plants thousands of years ago by artificial selection of genetic variants within populations. Between 8000 and 1000 B.C. horses, camels, oxen, and various breeds of dogs (derived from the wolf family) had been domesticated, and selective breeding soon followed. Cultivation of many plants, including maize, wheat, rice, and the date palm, began around 5000 B.C. Remains of maize dating to this period have been recovered in caves in the Tehuacan Valley of Mexico. Such evidence documents our ancestors' successful attempts to manipulate the genetic composition of species.

While few, if any, significant ideas were put forward to explain heredity during prehistoric times, during the Golden Age of Greek culture, philosophers wrote about this subject as it relates to humans. This is evident in the writings of the Hippocratic School of Medicine (500–400 B.C.), and of the philosopher and naturalist Aristotle (384–322 B.C.). The Hippocratic treatise *On the Seed* argued that active "humors" in various parts of the body served as the bearers of hereditary traits. Drawn from various parts of the male body to the semen and passed on to offspring, these humors could be healthy or diseased, the diseased condition accounting for the appearance of