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基因科技传奇专辑（1）

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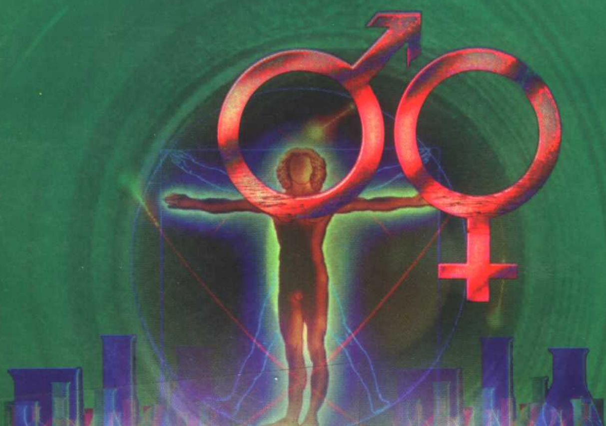
人类基因组计划

Brief Introduction to Human Genome

山河译注



● 人类基因组探秘
● 微生物基因组计划
● HGP将给人类带来什么

北京大学出版社

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A Brief Introduction to Human Genome

东辰通业教育网络技术有限公司
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图书在版编目(CIP)数据

英语活页文选·基因科技传奇专辑/《英语活页文选》编辑组编。
北京:北京大学出版社,2001.5.
ISBN 7-301-04956-0
Ⅰ.英… Ⅱ.英…Ⅲ.英语-语言读物
Ⅳ.H319.4

中国版本图书馆 CIP 数据核字(2001)第 79200 号

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书　　名:《英语活页文选》·基因科技传奇专辑(1)
　　　　　人类基因组计划
著作责任者:《英语活页文选》编辑组
标 准 书 号:ISBN 7-301-04956-0/H·0611
出　版　者:北京大学出版社
地　　　址:北京大学校内 100871
排　版　者:北京环鹏伟业文化发展有限公司
印　刷　者:北京牛山富各庄福利印刷厂
发　行　者:北京大学出版社
经　销　者:新华书店
850×1168　32 开本　5.875 印张　147 千字
2001 年 5 月第一版　2001 年 5 月第一次印刷
全套 6 册定价 45 元,每册定价 7.50 元

前　言

19世纪60年代奥地利学者孟德尔根据豌豆杂交实验首次提出遗传因子概念；1909年丹麦植物学家和遗传学家约翰逊第一次提出基因这个名词；1944年三位美国科学家分离出细菌的DNA(脱氧核糖核酸)，并发现DNA携带生命遗传物质的分子；1969年科学家成功地分离出第一个基因；1990年10月国际人类基因组计划正式启动；2000年成功地绘制了人类基因组工作框架图；2001年2月12日中、美、日、德、法、英等六国科学家联合公布了人类基因组图谱及初步分析结果。从以上大事记，人们不难看到被誉为"生命科学"的基因及基因组研究已经取得突飞猛进的发展。

为了使广大读者对基因和与之有关的学科能有一个概括的了解，并学习相关的英语知识，我们特意编写了这套丛书。它们是：《基因——生命之本》、《人类基因组计划》、《操作DNA，操作生命的未来》、《第四次医学革命——基因治疗》、《揭开癌症的秘密》、《器官移植》。《基因——生命之本》：介绍了基因与人类健康、人体发育和人类行为的关系。《人类基因组计划》：简单介绍人类基因组计划和相关的知识，并简述人类基因组计划将给人类带来什么。《操作DNA，操作生命的未来》：专门介绍转基因技术，该项技术在农业和医学研究中的应用，以及对人类健康、生态、环境和生物伦理学的影响。《第四次医学革命——基因治疗》：综合介绍科学家从疾病的根源异常基因来医治各种顽症的尝试和失败，并预言这场革命将在未来的三四十年内彻底改变医学界。《揭开癌症的秘密》：寻找致癌基因，叩开癌症之门。通过本书可以对医学界的抗癌之战有一个综合的了解。《器官移植》：概述器官移植的现状，重点介绍科学家对于基因移植方法的种种探索。

我们衷心希望广大读者能够喜欢这套丛书并从中受益。

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内容简介

2001年2月11日，美、英、日、德、法和中国等六个国际人类基因组计划参与国的科学家联合宣布，准确、清晰、完整的人类基因组图谱已经绘制完成。揭示人类基因组面貌是一项出色的成就，也为人类医学新发展提供了崭新的、激动人心的前景。

Chapter 1 Introduction to Human Genome Project

At the end of the road in Little Cottonwood Canyon, near Salt Lake City, Alta is a place of near-mythic renown among skiers. In time it may well assume similar status among molecular geneticists. In December 1984, a conference there, co-sponsored by the U.S. Department of Energy (DOE), pondered a single question: Does modern DNA① research offer a way of detecting tiny genetic mutations②—and, in particular, of observing any increase in the mutation rate among the survivors of the Hiroshima and Nagasaki bombings and their descendants? In short the answer was, **Not yet.** But in an atmosphere of rare intellectual fertility, the seeds were sown for a project that would make such detection possible in the future—the Human Genome Project.

In the months that followed, much deliberation and debate ensued. But in 1986, the DOE took a bold and unilateral step by announcing its Human Genome Initiative, convinced that its mission would be well served by a comprehensive picture of the human genome. The immediate response was considerable skepticism—

第一章 人类基因组计划简介

阿尔塔坐落在通往小棉木峡谷的路的尽头，盐湖城附近。在滑雪者眼中它是一个近乎神秘的地方。在分子遗传学家眼里，它也同样有着类似的地位。1984 年 11 月，美国能源部(DOE)参与主办了一个研讨会，会议提出了这样的一个问题：现代分子生物学研究能不能提供一个检测微小的遗传突变的方法——尤其值得一提的是能否检测出在广岛和长崎原子弹爆炸之后的幸存者和他们的后代在突变的比例上是不是有增加？当时答案是：还没有。但是就在这种珍贵的学术氛围中，科学家开始播撒下了人类基因组计划的种子，而这个计划将使这种检测在将来成为现实。

在接下来的几个月里，人们进行了反复的思考和辩论。在 1986 年，DOE 大胆地单方面提出了它的人类基因组研究意向，并宣称其目标是描绘出完整的人类基因组图。当时有很大一部分人持怀疑的态度，怀疑科学界

①DNA (deoxyribo nucleic acid): The molecule that encodes genetic information. DNA is a double-stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner DNA (脱氧核糖核酸)：编码遗传信息的分子 DNA 是靠核苷碱基对间的弱键结合的双链分子 DNA 中还有的四种碱基为：腺嘌呤(A)，鸟嘌呤(G)，胞嘧啶(C)和胸腺嘧啶(T)。自然界中 A 只与 T，G 只与 C 配对；这样每条单链的碱基序列都可以由另一条推出来

②mutation [mju'teiʃən] *n*. any heritable change in DNA sequence 突变：DNA 序列上的任何的可遗传的变化

skepticism about the scientific community's technological wherewithal for sequencing the genome① at a reasonable cost and about the value of the result, even if it could be obtained economically.

Things have changed. Today, a decade later, a worldwide effort is under way to develop and apply the technologies needed to completely map and sequence the human genome, as well as the genomes of several model organisms. Technological progress has been rapid, and it is now generally agreed that this international project will produce the complete sequence of the human genome by the year 2005.

And what is more important, the value of the project also appears beyond doubt. Genome research is revolutionizing biology and biotechnology②, and providing a vital thrust to the increasingly broad scope of the biological sciences. The impact that will be felt in medicine and health care alone, once we identify all human genes, is inestimable. The project has already stimulated significant investment by large corporations and prompted the creation of new companies hoping to capitalize on its profound implications.

But the DOE's early, catalytic decision deserves further comment. The organizers of the DOE's genome initiative recognized that the information the project would generate—both technological and

能否得到足够的必需的资金以可接受的成本对基因组进行测序，怀疑这个结果的价值，甚至于怀疑能不能经济地完成这项计划。

但是，时代变了。十几年后的今天，全世界都在为了开发和使用能完整地进行人类基因组图谱绘制和测序的技术而努力着，这些技术应用的对象还包括了几个模式生物。技术进步得很快，现在普遍的观点是这项国际计划将在2005年得到人类基因组完整的序列。

更重要的是，人们对这项计划的意义也确信不疑了。基因组研究正给生物学和生物技术带来革命，为在越来越广阔的领域中研究生命科学提供了一个至关重要的推动力。一旦我们识别出所有的人类基因，在医疗和健康护理方面将产生不可估量的变革。这一计划已经刺激了很多大公司进行数目可观的投资，并推动了新公司的产生，这些新公司希望能借助它深远的影响来得到发展。

DOE早先提出的催化剂般的决定还值得进一步探讨。DOE的基因组意向的组织者早就认识到这项计划所产生的技术上和遗传

① genome ['dʒiːnəum] *n*. the whole set of genes of a species 基因组：指某种生物的全套的基因 ② biotechnology ['baiəutek'nɔlədʒi] *n*. a set of biological techniques developed through basic research and now applied to research and product development. In particular, the use by industry of recombinant DNA, cell fusion, and new bioprocessing techniques 生物技术，一套在基础研究中开发出来的技术，现在用于研究和产品开发。尤其指重组DNA技术，细胞融合，和新的生物处理技术的工业运用

genetic—would contribute not only to a new understanding of human biology, but also to a host of practical applications in the biotechnology industry and in the arenas of agriculture and environmental protection. A 1987 report by a DOE advisory committee provided some examples. The committee foresaw that the project could ultimately lead to the efficient production of biomass for fuel, to improvements in the resistence of plants to environmental stress, and to the practical use of genetically engineered microbes to neutralize toxic wastes. The Department thus saw far more to the genome project than a promised tool for assessing mutation rates. For example, understanding the human genome will have an enormous impact on our ability to assess, individual by individual, the risk posed by environmental exposures to toxic agents. We know that genetic differences make some of us more susceptible, and others more resistant, to such agents. Far more work must be done before we understand the genetic basis of such variability, but this knowledge will directly address the DOE's long-term mission to understand the effects of low-level exposures to radiation and other energy-related agents—especially the effects of such exposure on cancer risk. And the genome project is a long stride toward such knowledge.

The Human Genome Project has other implications for the DOE

上的数据，将不仅仅有助于我们对人类生命有全新的理解，而且还是生物技术工业、农业和环保领域里实际应用的出发点。DOE顾问委员会1987年的报告提供了几个例子。这个委员会预见到，这项计划将最终使生物燃料的有效开发成为可能，能够提高植物对环境压力的抵抗能力，使得用基因改造过的微生物进行有毒废料的中和成为现实。这样，这个部门并不仅仅把基因组计划看成是测评突变率的有效工具，而是看到了它更为广阔的前景。例如，对人类基因组的理解将大大改变我们评估环境毒物对每个个体造成的危险的能力。我们知道，遗传上的区别使得我们中的某些人对一些物质比较敏感，而另一些人则相对更有抵抗力。在我们完全理解这种多样性的遗传基础之前，还有许多工作要做，但是这些知识，将直接有助于完成DOE的长期目标：理解低剂量的接受放射物和其他能源物质照射的后果——特别是这些照射的致癌危险。而人类基因组计划正是我们获取这样的知识需要迈出的一大步。

人类基因组计划对DOE来说还有别的

as well. In 1994, taking advantage of new capabilities developed by the genome project, the DOE formulated the Microbial Genome Initiative to sequence the genomes of bacteria of likely interest in the areas of energy production and use, environmental remediation and waste reduction, and industrial processing. As a result of this initiative, we already have complete sequences for two microbes that live under extreme conditions of temperature and pressure. Structural studies are under way to learn what is unique about the proteins[1] of these organisms—the aim being ultimately to engineer these microbes[2] and their enzymes for such practical purposes as waste control and environmental cleanup.

And other little-studied microbes hint at even more intriguing possibilities. For instance, Deinococcus radiodurans is a species that prospers even when exposed to huge doses of ionizing radiation. This microbe has an amazing ability to repair radiation-induced damage to its DNA. Its genome is currently being sequenced with DOE support, with the hope of understanding and ultimately taking practical advantage of its unusual capabilities. For example, it might be possible to insert foreign DNA[3] into this microbe that allows it to digest toxic organic components found in highly radioactive waste, thus simplifying the task of further cleanup. Another approach might be

应用。1994年，利用基因组计划已有的成果，DOE又定制了微生物基因组研究意向，对在能源生产和利用、环境净化、污物处理和工业生产中有重大价值的细菌进行序列的测定。随着计划的进行，我们已经得到了两种能在极端的温度和压力下生存的微生物的完整序列。为了找出这些物种的蛋白质所具有的特性，人们正在进行结构研究，最终的目的是对这些微生物和它们的酶进行改造，使之能在诸如废物控制和环境净化当中发挥作用。

另一些研究得很少的微生物甚至提供了更吸引人的应用前途。例如，*Deinococcus radiodurans* 是一种在极高剂量的电离辐射中也能很好生存的物种。这种微生物有着惊人的修复由辐射引发的DNA损伤能力。在DOE的支持下，人们正在测定它的基因组，希望能理解并最终利用它这种非凡的能力。举个例子，我们有可能把外源DNA插入到这个微生物中，使它能降解高放射性废物中有毒的有机物，这将大大简化后期处理工作。另一

①protein [ˈprəutiːn] *n*. a large molecule composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nucleotides in the gene coding for the protein. Proteins are required for the structure, function, and regulation of the body's cells, tissues, and organs, and each protein has unique functions. Examples are hormones, enzy-mes, and antibodies 蛋白质，由一条或多条具有特定顺序的氨基酸链组成的大分子；这一顺序由编码该蛋白的基因的碱基序列决定。机体的细胞，组织和器官需要蛋白质作为结构单位，行使功能并进行调节，每种蛋白质都有独特的功能。例如：激素，酶和抗体

②microbe [ˈmaikrəub] *n*. tiny living creature that can be seen only with the help of a microscope 微生物，指只能在显微镜下才可以观察到的生命体

③foreign DNA：外源DNA
DNA fragment from out-side an organism 来自生物体外的DNA片段

to introduce metal-binding proteins onto the microbe's surface that would scavenge highly radioactive isotopes out of solution.

Biotechnology, fueled in part by insights reaped from the genome project, will also play a significant role in improving the use of fossil-based resources. Increased energy demands, projected over the next 50 years, require strategies to circumvent the many problems associated with today's dominant energy systems. Biotechnology promises to help address these needs by upgrading the fuel value of our current energy resources and by providing new means for the bioconversion of raw materials to refined products—not to mention offering the possibility of entirely new biomass-based[1] energy sources.

We have thus seen only the dawn of a biological revolution. The practical and economic applications of biology are destined for dramatic growth. Health—related biotechnology is already a multi-billion-dollar success story—and is still far from reaching its potential. Other applications of biotechnology are likely to beget similar successes in the coming decades. Among these applications are several of great importance to the DOE. We can look to improvements in waste control and an exciting era of environmental bioremediation[2]; we will see new approaches to improving energy efficiency;

种应用是将金属结合蛋白引入到它的表面，使之能从溶液中消除高放射性的离子。

生物技术，得益于基因组计划带来的新的成果，将在改进化石资源的利用上扮演重要的角色。在未来五十年内，与日俱增的能源需求，使得今天的主要能源系统面临着一系列急需解决的问题。通过提高现代能源物的燃料值以及提供用生物转化对原料进行提炼的新方法，生物技术将帮助人们解决能源问题，即使我们还没考虑生物技术提供全新的生物能源的可能性。

这还只是生物革命的开始。生物学的实际而又经济的应用必将有巨大的发展。生物技术在保健领域已经书写了一个数十亿美元的成功故事——而且它的潜力还远未发挥出来。生物技术的其他应用也将在未来的数十年内取得相似的成就。其中有一些对于DOE来说有着重要的意义。我们期待着在废物控制和令人兴奋的生物净化领域的进步；我们将看到增进能源利用率的新发明；甚至可以

①biomass-based [ˈbaiəumæs] 基于生命物质的 of or come from argamisms 以来自于生物体的物质为基础的

②bioremediation [baiəuremidiˈeiʃən] *n*. clean up using living organisms 使用生物来净化环境