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基因科技传奇专辑（6）

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基因——生命之本

Genes—the Basis of Life

孟庆禾 杨晓红 译注

- 基因和人类健康
- 基因和人体发育
- 基因和人类行为
- 解码生命的蓝图

北京大学出版社

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策 划 人:叶 乡
宗 和
责任编辑:范一亭
钱王驷(特邀)
李 顶(特邀)
封面设计:张 虹
电话:
(010)62752028(编辑)
(010)62752019(邮购)
(010)62754140(发行)
e-mail:
gl@pup.pku.edu.cn

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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实飞猛进的发展。

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。《人类基因线计划》: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

19世纪60年代奥地利学者孟德尔首次提出遗传因子概念。经历了整整一个世纪，科学家成功分离出第一个基因。新世纪伊始，人类基因组计划绘制出了完整的人类基因组图谱。影响人类健康、发育，甚至于人类行为的基因技术正在取得突飞猛进的发展。

Chapter 1 Introduction

1.1 Genes: Key to Human Health?

Understanding how genetic factors contribute to human health is gathering speed. In the 1950s, the structure of DNA had just been solved and the precise number of human chromosomes was still under debate①. We now know that there are 46 human chromosomes, which between them house 3,000 million base pairs of DNA and encode② about 60,000 to 80,000 proteins. These coding regions make up only about 2% of the genome and some chromosomes have a higher density of genes, but others not.

Sequencing our DNA is all about hunting genes. As well as providing an excellent framework③ for the complete sequencing of the human genome, the physical map has assisted directly in identifying about 100 disease-causing genes. However, one of the biggest difficulty ahead is to find genes involved in diseases that have a complex pattern of inheritance, such as those that contribute to diabetes, obesity④, cancer and mental illness. In all these cases, no one gene has the yes/no power to say whether a person has a disease or not. It is likely that more than one mutation is required before the disease is manifest. A number of genes may each make a subtle contribution to a person's susceptibility to a disease; genes may also affect how a person reacts to environmental factors. Unravelling⑤ these

第一章 简介

1.1 基因:人类健康的关键?

人们对遗传因素对人类健康影响的认识速度正在加快。在20世纪50年代,DNA的结构刚被确定,但人类染色体的确切数目仍存在争议。现在我们知道人类有46条染色体,它们由30亿个DNA(脱氧核糖核酸)碱基对组成,编码6万到8万种蛋白质。这些编码区只占人类基因组的2%,而且某些染色体含有的基因密度较大,其他染色体含有基因的密度小。

我们的DNA测序的目的就是寻找基因。基因组的物理图谱除了为人类基因组的全序列提供了一个非常好的框架,它还直接帮助确定了100多个致病基因。但是,今后存在的最大困难是发现能产生像糖尿病、肥胖症、癌症和精神病那样的复杂遗传方式的基因。在这些情况下,没有一个基因能通过开/关来决定一个人患病与否。在疾病被确定以前,可能有一个以上的基因发生了突变。可能存在许多基因,它们之中的任何一个都或多或少地使人容易患病;基因也可能影响一个人怎样对环境因素作出反应。勿庸置疑,揭示

① debate [di'beit] *n*. discussion 争论,辩论

② encode [in'kəud] *vt*. convert ordinary language into code 编码(把…译成电码,代码化)

③framework ['freimwəːk] *n*. a structure supporting or containing something 构架,框架,结构

④ obesity [əu'biːsəti] *n*. more than average fatness 肥胖,肥大

⑤ unravel [ʌn'rævəl] *v*. disclose 解开,弄清楚(秘密)

networks of events will undoubtedly be a challenge for some time to come.

[From: http://www.ncbi.nlm.nih.gov/disease/]

1.2 Genes and Development: How Did We Get Here?

It's a question both basic and profound①. A child might give a guileless answer: First comes love, then comes marriage, then comes the baby in the baby carriage.

How did we get here? Each of us begins as a zygote②, as the sum of egg and sperm, as a primal cell. Genetically complete and raring to go, this single cell contains the biological software necessary to create a human, written in three billion base pairs of DNA, twisting through 23 pairs of chromosomes.

Understanding exactly how this software moves an organism from zygote to adulthood is another matter. What switches genes on and off, directing them first to develop a primitive③ skin, a crude nervous system, and a chamberless, but beating, heart? And then to refine—to become eyes that can see...with lashes and brows! To become hands and fingers swirled④ with a print unique in all the world. To become a mind that not only controls basic functions like breath and heartbeat, but one that burns with the electricity of reason and imagination!

How did we get here? Embryologists, developmental biologists,

这些事件之间的关系在很长一段时间里将是一种挑战。

1.2 基因和发育:我们怎样来到这儿?

这是一个基础而又复杂的问题。小孩的答案可能很天真:首先恋爱,接着结婚,于是婴儿车中就会有婴儿出现了。

我们怎样来到这儿?我们每个人都由一个受精卵开始,由卵子和精子融合而来,由一个最初的细胞开始。这个单细胞在遗传上是完整的,很容易死亡,它包含了创造一个人所需要的所有的生物学软件。这个软件由30亿个DNA碱基对书写而成,缠绕形成23对染色体。

准确地理解这些软件怎样使生物体由受精卵转变为成年个体是另一件事情。是什么把基因打开和关闭,从而指导它们首先产生原始皮肤,粗糙的神经系统,以及一个没有房室但可以跳动的心脏?接下来进行精炼——成为具有睫毛和眉毛而又能用来观察的眼睛!成为世上无比灵巧的手和手指。成为不但能控制呼吸和心跳这样的基本功能而且富有理智和想象的心智!

我们是怎样来到这里的?胚胎学家、发

①profound [prə'faund] *adj.* far-reaching and thoroughgoing in effect especially on the nature of something 深奥的

② zygote ['zaigəut] *n.* the cell resulting from the union of an ovum and a spermatozoon (including the organism that develops from that cell) 受精卵,接合子

③primitive ['primitiv] *adj.* belonging to a nearly stage of technical development; characterized by simplicity and (often) crudeness 原始的,简单的

④ swirl [swəl] *vi.* *turn in a twisting or* spinning motion 弯曲

fetal pathologists① and others have worked for years to sketch in the outlines of development. But it's still pretty much a mystery, and like all the best mysteries, it defies easy resolution. Clues are elusive②. Methods, mechanisms, and motives are hard to deduce. Solving it will take more than good detective work; scientific sleuths also need good hunches and good luck.

In detective novels, it's frequently the chance clue—the stray③ fingerprint, the contradictory statement, the forgotten letter—that cracks the case. So it is too in science, as ORNL's Waldy Generoso has come to experience. From a seemingly ordinary experiment, Generoso has stumbled④ upon extraordinary results, clues that might help crack the mystery of how we got here, evidence that might also help to explain how things can go wrong on the journey.

[**From: Kit Carlson, Science & Technology from Oak Ridge National Laboratory, *Detectives at The Dawn of Life*, 1993**]

1.3 The Gene Exposed

"We have caught the first glimpses⑤ of our instruction book, previously known only to God," said Dr Francis S. Collins, director of the National Human Genome Research Institute. The Human Genome Project (HGP) in its first major announcement on Monday, June 26, 2000, gave the draft plan of a typical human—the rough

育生物学家、胎儿病理学家和其他人已经工作了多年以期勾勒出发育的轮廓。但这仍旧是个谜，并且像所有的谜一样，它不能被轻易地解决。线索令人难以捉摸。方法、机理和目的难以推导。解决这个问题光靠良好的探测工作是不够的，科学的敏感还需要良好的预感和不错的运气。

在侦探小说中，经常是偶然的线索（偶然遇到的指纹、矛盾的陈述及被遗忘的信件）使案件侦破。在科学上亦是如此，就像国立橡树岭实验所的韦尔蒂·吉列罗索的体验一样。在一个看起来很平常的试验中，吉列罗索对异常的结果，也就是能帮助解决我们怎样来到这儿的线索，以及帮助解释在这个行程中事情是怎样误入歧途的。

①pathologist [pəˈθɑlɔdʒist] *n.* a doctor who specializes in medical diagnosis 病理学家

② elusive [iˈluːsiv] *adj.* be difficult to detect or grasp by the mind 难懂的，难捉摸的

③ stray [strei] *adj.* met by chance 偶然遇见的

④stumble [ˈstʌmbəl] *v.* make an error 使困惑

⑤glimpse[glimps] *n.* a brief or incomplete view 一瞥，一看

1.3 暴露的基因

“我们已经第一眼瞥见了我们的说明书，在这以前只有上帝知道它。”人类基因组国家研究所所长弗朗西斯·S.柯林斯博士说道。人类基因组计划（HGP）在2000年6月26日（星期一）第一次主要宣言中，给出了典型的人类

sketch of DNA, the molecule that determines① the hereditary and structural instruction set of a human.

The information provided by the current work—the gaps will be filled by 2003—has already led to significant efforts② to locate the genes that cause diseases and attempt genetic modifications to effect cures for them.

Such a catalogue of all human genes will help us understand better and possibly treat many inherited diseases and other human illnesses such as heart disease, diabetes and even cancer. It will be useful in the prevention, diagnosis③ and treatment of other diseases. Scientists believe it is fundamental to the progress of medical research throughout the world. It provides the basic set of inherited instructions for the development and functioning of a human being and this knowledge is likely to transform medical care in the 21st century.

While there are specific genes that increase susceptibility④ to certain diseases, a large number of genes are often involved, and this is further complicated by environmental factors, making the determination of the source of most genetic diseases very difficult.

What are the medical benefits of this new knowledge? Within a decade, you may be able to go to your doctor, give a small blood sample⑤ and be told what is wrong with you—almost. By checking your genetic code—your personal version of the human genome—the

草图计划——DNA这种决定人类遗传和结构机制的分子的草图。

当前的工作提供的信息(序列间隔将在2003年之前被填补)已经在定位致病基因方面取得重要成就,并试图进行基因修饰来治疗它们。

那样一种人类所有基因的目录将帮助我们更好地理解以及治疗许多遗传病和其他人类疾病,比如心脏病、糖尿病甚至癌症。它在防止、诊断和治疗其他疾病方面会很有效。科学家们相信这对全世界医学研究的进步将是很重要的。它为人类的发育和功能行使提供了基本的遗传指令,这些知识在21世纪可能使医疗保健发生变革。

虽然有某些基因能提高患某些疾病的可能性,但是通常有大量的基因参与其中,而且环境因素使这进一步复杂化,从而使确定许多遗传疾病来源十分困难。

这些新知识的医学用途是什么?在10年之内,你可以去看医生,给他一点血样,然后你就能被告知害了什么病——大多情况下如此。通过检查你的遗传密码(你自己的基因

①determine [di'tɜmin] *v*. reach, make, or come to a decision about something 决定,确定

②effort ['efət] *n*. a notable achievement 成就

③diagnosis [daiəg'nəusis] *n*. identifying the nature or cause of some phenomenon 诊断

④susceptibility [səsepti'biliti] *n*. the state of being susceptible; easily affected 易感性,感受性

⑤sample ['sæmpl] *n*. all or part of a natural object that is collected and preserved as an example of its class 标本,样品