Medical Genetics:Principles and Application

医学遗传学——原理与应用

（第2版）

主　编　杨保胜 丰慧根 王天云

中国人口出版社

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编委会

第二版说明

本书第1版出版以来，深受广大读者的欢迎与喜爱，但由于医学遗传学在此期间有了长足的进展，涉及的新技术和新成果不断涌现，尤其是在人类基因组学和分子遗传学技术方面的进展更为显著。为了能及时地加入新的资料、反映新的动态，虽然本书出版至今只有2年时间，我们还是决定对其进行修订。

本书第2版保持了第1版的结构体系和写作风格，但鉴于目前学科发展的现状，补充了大量的新内容，有些地方完全是重新撰写的，在必要的地方进行了增加、删节、修改和订正。由于有关网络信息日新月异，瞬息万变，本书对所有参考网站进行了订正。考虑到双语教学的需要，在正文中增加了三级英文标题。在医学遗传学导论一章中不仅对医学遗传学资料与互联网进行了改写，而且增加了遗传病研究策略一节；在染色体与染色体病一章中增加了染色体病发病的分子基础等内容；对基因与基因组学一章进行了改写，补充了基因突变的分子细胞生物学效应的内容；在单基因遗传与单基因病一章中，对动态突变疾病、遗传印记和遗传异质性等内容进行了扩展，并增添了一些新的内容。删节较多的是遗传伦理学、行为遗传学和毒理遗传学中的部分繁琐内容。同时对第1版的图、表、文字、标点做了修改，增加了一些新的图表，删去不必要的图表，对表达欠妥的文字做了订正。

新参加本次修订的有来自教学和科研第一线的3位博士，使参加本书编写的队伍达到5位博士和11位教授及副教授，对他们为本书的辛勤工作，表示感谢。

尽管我们在修订时仍然秉承第1版写作时的宗旨，力求使本书内容全面而新颖、概念准确，能系统地反映本学科的历史和最新研究进展，形成一部有自己特色的、比较完整的医学遗传学基础科学论著，但限于作者的知识水平和写作能力，虽自决定修订之日起，夜以继日，不敢懈怠，惟望以勤补拙，减少疏漏，但书中错误和不妥之处在所难免，欢迎读者和专家指正、赐教，作者不胜感激。

杨保胜

2005年7月

前　言

医学遗传学是医学领域中发展很快的带头学科之一。近年来，随着分子生物学技术的迅速发展，尤其是在人类基因组计划的推动下，已认识到的单基因遗传病达 14 000 种以上，遗传病对人类健康的危害日益重要。目前对人类疾病已有可能进行基因诊断和基因治疗，为从根本上预防遗传病的发生，开辟了光辉的前景，医学遗传学已成为培养高级医学专门人才的知识结构和能力的一个重要组成部分。

本书为河南省面向 21 世纪医学遗传学教学内容改革研究课题的成果。本书编写的指导思想为既照顾基本理论的系统性和完整性，又兼顾医学实践的需要；既介绍基本知识，又有一定的深度、适度和广度；既反映最新成就，如植入前遗传学诊断、人类基因组计划和单核苷酸多态性标记等，使学生了解本学科的新进展，又避免片面求新求多。

本书共分 15 章，主要包括导言、染色体与细胞分裂、染色体畸变与染色体病、基因与基因组学、单基因遗传与单基因病、多基因遗传与多基因病、线粒体遗传病、群体遗传学、生化与分子遗传学、药物遗传学、肿瘤遗传学、遗传病的诊断、遗传病的防治、医学遗传学专题（包括免疫遗传学、发育遗传学、行为遗传学、毒理遗传学和遗传伦理学）等。

本书编写体现三基（基本理论、基本知识和基本技能）、七新（新知识、新概念、新理论、新方法、新技术、新信息和新理念），力争达到七性（思想性、科学性、先进性、启发性、适应性、针对性和适用性）。充分体现对学生独立获取知识和信息能力的培养。如在各章后列出一些阅读材料、参考文献和专业网址，培养学生阅读参考文献的能力，有利于学生针对某些章节中的重要问题进一步进行探讨，使学生能从中吸取必要的信息和资料，总结出新的、更深入的认识；增加一些相关的英文文本框和中英对照的每章内容概要来扩充学生的知识面和奠定阅读英文专业文献的基础；每章后列出思考题，以启发学生领会相关的理论概念，并用来分析、认识某些问题，是培养学生“能力”的一个重要方面；书后附中英文索引，以利于查阅有关内容，提高其适用性。

参加本书编写的主编和副主编共 7 位，都是教学和科研第一线的教授和副教授，具有丰富的教学和科研经验，主编或参编过多部教材或专著。

本书以新颖的结构体系，比较系统地介绍了现有的医学遗传学的知识，配含大量的图表（图 95 幅，表 38 个，英文文本框 30 个），以方便读者学习。本书不仅可作为医学本科学生的教材，也可作为医护人员、从事遗传医学、生殖医学、预防医学、环境医学、计划生育、人口学和生物学等有关部门工作者的参考书。

由于编著具有双语特点的《医学遗传学》是一项新的尝试，可供参考和借鉴的资料不多，加之我们水平有限，虽经集体审稿，本书从形式到内容上的欠妥之处在所难免，殷切希望读者提出宝贵意见，以便在修订时加以改进，不断提高质量，以期在培养合格的医学人才中发挥应有的作用。

杨保胜　丰慧根

2003 年 7 月

Preface for the Second Edition

The book has been very popular with the readers since the 1st edition was published. Medical genetics has been developed rapidly during the periods and new perspective and concepts have been emerging, especially the approach on human genomics and molecular genetics, made it necessary to add information and concept to reflect currently view. Above on, we decide to make this book revise although the first edition was printed only 2 years ago.

The configuration and style of the 1st edition have been retained in the 2nd edition, some new contents are added, and some sections are rewritten, revised and deleted. Because the net information changes with each passing day, the new edition has revised all reference network web information. In order to satisfy the need for bilingual learning, we superinduce the third class title of English. In the chapter of introduction to medical genetics not only the network information is revised, but also the research strategy of genetic disease is added. The molecular structure and fundamental are added in the chapter of chromosome aberration and chromosome disease. The chapter of gene and genomics is also revised and the effect of molecular cytobiology of gene mutation is attached to it. Extensions on gene mutation, genetic imprinting and heterogeneity have been carried out while some new contents have been supplied to the chapter of monogenic inheritance and monogenic disease. Complicated contents of behavior genetics, toxicogenetics and genetic ethics are deleted and simplified. Meanwhile, some figures, tables and punctuations used in the 1st edition are amended, superinduced and expurgated. Many commentaries and conceptions without properly expressed have also be corrected.

There are five MDs and eleven professors or associate professors in all who take part in this revision, including three new MDs coming from the front line of teaching and scientific research. We thank them for their contributions to this text.

We recieve the tenet of the 1st edition when proceed the new edition, which make it comprehensive, new and reflect the subject history and current progress of medical genetics, and produce one special and relative intact medical genetics book. Because of the editor's limited knowledge and writing ability, though we lose evening and weekends and work night and day, some errors and misgivings cannot be ensured to avoid in this book, we hope that readers and experts can put forward some good suggestions.

Professor Baosheng Yang

July, 2005

PREFACE

Medical genetics is one of leading subjects developing rapidly in medical field. Recently, the known number of diseases of monogenic inheritance has been beyond 14 000 with the rapid development of molecular biology technology, especially driven by human genome project (HGP). It is possible for gene diagnosis and therapy to be applied in some human diseases, making resplendent prospect for preventing inherited disease fundamentally. Medical genetics has become an important part for culturing senior medical worker knowledge and ability.

This book is fruit of medical genetics teaching content innovation topic facing 21 st century in Henan province. This book is aimed not only at considering system and integrity of basic theory, but also the need of medical practice; not only introducing elementary knowledge, but also possessing definite depth, moderation and extend; not only reflecting the last developments, for example genetics diagnosis before implantation, HGP, SNP labeling etc, but also avoiding pursuing too new and excessive knowledge.

This book is divided into 15 chapters, mainly including introduction, chromosome and cell division, chromosomal aberration and chromosome disease, gene and genomics, monogenic inheritance and monogenic disease, polygenic inheritance and polygenic disease, mitochondrial genetic disorder, population genetics, bichemical and molecular genetics, pharmacogenetics, oncogenetics, diagnosis of hereditory disease, prevention and therapy of hereditary disease, techniques of medical molecular genetics, medical genetics subjects (immunogenetics, development genetics, behavior genetics, toxicogenetics and genetic ethics) etc.

This book has been designed to embody three bases (basic theory, basic knowledge, basic skills), seven innovations (new knowledge, new concept, new theory, new method, new technology, new information, new idea), and seven characters (idea, science, advance, enlighten, suitability, pertinency, applicability), make students take knowledge and information ability by themselves. Some reading material, references and special webs site are also listed to help to culture students' ability, discuss important problem about certain chapters, catch necessary information and data, get new and in-depth knowledge. Some English box and Chinese-English summary will be aided to extend students' knowledge and establish the background, improve ability for reading English special literature. Thinking problems can arouse students to understand related theories and concepts and analyze or learn some problems, an important aspect to culture ability of students. Chinese-English index can profit students to lock up related content and enhance its applicability.

Seven professors or associate professors took part in writing this book, all work in front line of teaching and scientific research, have rich teaching experiences and have edited or joined in writing many teaching materials and monographs.

The composing of this book is up-to-date, and knowledge existing on medical genetics is systematic introduced. There are large number of figures, tables and boxes (95 figures, 38 tables and 30 boxes) for readers to study. This book can be not only taken as teaching material by medical undergraduates, but also as reference by doctors, nurses and workers of related departments who are engaged in inheritance medicine, reproductive medicine, preventive medicine, environmental medicine, family planning (birth control), demography and biology etc.

It is a new attempt to write medical genetics teaching material using bilingualism, the reference is few and far between. Because of editors' limited knowledge, although draft was examined by collectivity, shortcomings can't be avoidable. All the editors wish sincerely that readers could bring forward precious opinions in order to improve when revising it. Hope this book could play its effect in culturing qualified doctors and nurses again!

Baosheng Yang and Huigen Feng
July, 2003

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第一章　医学遗传学导言

Introduction to medical genetics

遗传与变异是生物界存在的极其复杂的生命现象。遗传(heredity)是生命有机体在生殖过程中所表现出来的亲代与子代间的相似现象。遗传是高度稳定的,但这种稳定性又是相对的。亲子之间仅仅是相似,而不会完全相同,这种同种个体(亲代与子代、子代与子代)之间的差异称为变异(variation)。遗传的稳定性使物种生生不息,世代相传。人体的发育、分化是细胞中的 DNA 分子所携带的遗传信息依照精确的时空程序与环境相互作用,逐步表达的结果。当遗传信息改变表达程序(突变)而出现错误时,就会导致人体某些器官结构和功能异常,发生疾病乃至死亡。遗传学(genetics)是研究生物遗传与变异的科学。医学遗传学是遗传学与医学相互渗透的一门边缘学科。

第一节　医学遗传学及其研究范围

Medical genetics and disciplines of study

一、医学遗传学的研究对象和任务

Object and task of study of medical genetics

医学遗传学(medical genetics)是运用遗传学的原理和方法研究人类遗传病的发生、传递规律、诊断、预防和治疗的一门学科。它的研究对象是人类,它以遗传学的基本理论和方法为基础,以分子生物学为先导,来研究人类疾病与遗传的关系,进而达到生的健康、生的优秀的目的。它分别从分子水平、细胞水平、个体水平和群体水平探索遗传病的发病机制、诊治措施和预防策略。

人类遗传学(human genetics)主要探讨人类正常性状(character)与病理性状的遗传现象及其物质基础,即以人类为研究对象的遗传学。而医学遗传学则主要研究人类(包括个体与群体)病理性状的物质基础和遗传规律,同时也研究与人类疾病密切相关的非人类生物的病理遗传现象。狭义来讲,医学遗传学是人类遗传学的一部分,而广义的医学遗传学除了研究人类以外,还研究与人类疾病相关的其他生物(如药用动植物、病原微生物)的遗传本质及与人类健康的关系等。

临床遗传学(clinical genetics)是运用遗传学的原理和方法,研究各种遗传病的诊断、产前诊断、预防(包括遗传咨询)和治疗的学科,是医学遗传学的核心内容之一。

二、医学遗传学的研究范围

Disciplines within medical genetics

医学遗传学是一门由遗传病(genetic disease)这一纽带把遗传学与医学结合起来的一门边缘学科,是遗传学理论在医学领域中的应用。随着医学科学和生命科学的发展,人类已逐步从分子、细胞、个体和群体水平等各个不同层次去研究医学遗传学的各种问题,使其得到了迅速的发展,其研究范围逐渐拓展,已形成了一门由多个分支学科构成的综合性学科。