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前　言

直到最近，医学遗传学和分子医学还被看作是三线医学中的学术专家们的独有领地。当问及对临床医学的分子遗传学方面的熟悉程度时，几年之前，大多数一线医务人员都会回答说，这与他们的日常医疗无关。

但现在这种回答听不到了。很少有实习医师或全科医师没有开过重组胰岛素、tPA或红细胞生成素；很少有儿科医师没有参加过对畸形或学习障碍儿童的分子医学诊断；很少有产科医师没有给遗传病高危夫妇做过羊膜穿刺或CVS；很少有普通外科医师没有遇到过关于遗传检验的作用问题，或者有明显乳癌或卵巢癌家庭病史的妇女的预防性外科手术问题。

与未来相比，目前分子遗传学在临床学中出现的程度还只是初步的，当人类基因组计划向着完全测量一个标准人的基因组的序列冲刺，并在2005年左右鉴定全部人类基因时，揭示人类疾病奥秘的步伐将不断加快。至今，大多数疾病-基因关系的发现还只建立在单个基因病之上（囊性纤维病，脆性X综合征，等等），或者孟德尔氏分类的更常见的疾病（BRCA1和BRCA2，遗传性非息肉病结肠癌综合征，等等）。但是，随着1998年启动的一个新的强化的基因组的计划目标，将对全部常见人类序列变异进行分类，涉及对所有疾病的较弱多基因作用的认识。许多产生的结果，个体化的预防医学策略，未来以基因为基础来确定发病的危度，将成为医学的常规工作。不依靠经验而依赖于对分子发病学的详细了解而新设计的药物将会出现。药物遗传学可以依据患者的遗传型预测一种具体药物配方的功效与毒性，并将成为给具体患者设计最佳治疗方案的标准程序。而基因疗法，建立在疾病-基因发现之上，将会成熟到成为内科医师对付疾病的装备的重要部分。

当我们看到列车进入铁轨时，这是一个收集分子医学信息并使之成为一本有权威性的教科书的理想时刻，《分子医学原理》的目标即在于此。弥合目前在基础科学与临床之间的鸿沟，这将有益于研究和临床。超过100章的篇幅覆盖广泛的论题，杰出章节作者及丰富的表格和插图，为目前和未来的分子遗传学和医学提供了一条捷径和急需的手册。

Francis S. Collins

（李洞译、白永权校）

序

对大多数内科医师来讲，分子医学和遗传学在传统的日常医疗工作中不起主要作用。但是，关于疾病的分子基础的新观念正在以不断加速的方式产生，使得我们对疾病的理解和治疗随之改变。各种技术进步激发了这种信息爆炸，而人类基因组计划的快速进展又为之推波助澜。众所周知，分子生物学目前正在引起医学教学和实践的示范性转变。分子医学的希望是激动人心的，但该学科的迅速渗透也对我们对疾病的理解和治疗提出了巨大挑战。《分子医学原理》这本书希望填补传统医学教科书与分子生物学新知识之间的鸿沟。

这本书定名为《分子医学原理》，反映了我们的努力，以使由遗传学和分子生物学提供的进展转移到医学的每一主要专业。通过与传统医学教科书的类比，本书依据主要器官系统进行编排。这种形式对大多数医学读者比较熟悉，从而符合许多作者和读者的专业范围。我们相信这本书对广大读者是有价值的，包括有理解力的学生、寻找各自领域知识更新的专家，或者他们尚未深入研究的论题，以及对分子医学的惊人变化很感兴趣的临床内科医师。

《分子医学原理》的每一专业章节均由各自领域的专家编写。本书编写了一系列导论章节，而每一专业章节包括额外的背景知识，以概括各自器官系统特殊的分子病理生理学论述。即使分子医学领域飞速发展，本书包括了最新的疾病的遗传学基础的综述，并强调了原理，从而使读者可以将所有最新突破整合到基础知识中去。我们的作者以表达细胞和遗传途径的许多清晰图表将许多复杂话题化繁为简，而且，我们特别着重强调了疾病的分子机制，以及由分子生物学工具的应用获得的新概念。本书最精彩之处在于将许多最新研究进展转变成临床有用的信息；我们的每一作者都提出了本专业的新近进展的未来意义。

除了讨论疾病的病理生理学，遗传学实际上在每一个医学领域都发挥了作用。据估计，人类基因组包括 50 000 到 80 000 个基因，虽然许多疾病由关键基因的突变引起，越来越明确的是，人的遗传学背景可以引起对许多疾病的易感性，或者改变其对环境的反应性。在某些病例，如高血压和心血管病，遗传的作用是多基因的，而我们还处于认识与此有关的许多基因的早期。在另一些情况下，如血友病、囊性纤维化以及上百种其他疾病，有关的基因已经被充分定性。

可以预料，新的信息的猛烈冲击似乎是令人畏缩的，难以被消化利用，特别是许多新技术和新名词。有讽刺意味的是，新的观点事实上可以大大

简化过去难以猜测的领域。例如，一些不同的遗传缺陷可以引起外周神经疾病，但是，打乱了髓鞘的正常折叠似乎是其共同的最终途径。与此相似，一些遗传上不同形式的早老性痴呆似乎有着一个共同的最终特征，即神经纤维团的形成。鉴定缺陷基因的性质(如营养不良，CFTR、FGF 受体)可以弄清关键生理过程的途径。同样，转基因和基因"切除"模型可以暴露基因的生理学功能。

令人惊喜的是在分子医学这个新领域已经影响到医学的每一专业。例如，心脏病专家正在解决遗传性心肌病和离子通道缺陷的分子基础问题，这些患者易感心律不齐。神经学专家发现了惊人数量的能引起神经变性疾患的基因突变。毫不奇怪，血液从经典的血红蛋白病的遗传描述迅速发展到确定其他病的分子基础，包括红细胞膜缺陷、凝血病以及血栓形成病。白血病和淋巴瘤的遗传根源发展为理解肿瘤机制的重要领域。囊性纤维化转运蛋白的鉴定使得可以对其进行分子诊断，在一些研究中心，基因治疗方案已经起步。免疫系统的遗传性改变正瞄准对传染源的易感性，以及许多炎症和自身性免疫病。我们对肝炎和 HIV 等病毒感染的理解得益于重组 DNA 技术的巨大帮助。在内分泌学方面，关于激素作用、性别分化以及内分泌肿瘤的机制已经得益于遗传缺陷的性质的阐明。而且，激素和信息传导领域已经展现了一个非常错综复杂的相互作用网络，介导着细胞对外部信号的反应。在肾脏病学，多囊肾病的原因已经明确，缺血和炎症的分子病理学正在明朗化。在皮肤病方面，肿瘤的发病机制已经取得巨大进展，类天泡疮病的分子发病学，以及其他许多自身免疫性疾病的机制也进展神速。在精神病学方面，遗传的作用正在迅速显示其重要性；其进展的概貌概述于本书关于行为、早老性痴呆、感情性疾病，以及酒精中毒各章。许多"经典"遗传病的分子机制也已阐明。Prader-Willi、Angelman 及 Beckwith-Wiedemann 综合征的研究在其定性方面起了关键作用。脆性 X 综合征，以及 Huntington 等神经疾患也在核苷酸重复性疾病的冲击下得到证明。在大多数病例，我们的新知识已经改善了诊断试验的可能性和精确性，促进了我们对其病理生理学的理解，并正在开辟着新的治疗途径，包括基因疗法。

我们有幸得到国际有名的章节作者和编者参与本书编写。他们从自己已经满额的研究、教学和临床活动中挤出时间编写出这些无论对其专业领域内外均有教益的章节。虽然本书有 120 多章，但未能包含其全部。我们宁可着眼于最近取得实质性进展的疾病，以及存在领域鸿沟的疾病。希望读者再参考其他来源求得进一步的信息。一个特别有价值的来源是《孟德尔人类遗传》的在线版，其网址为 WWW3.ncbi. nlm. hih. gov. /OMIM。代谢病的分子基础已在内科教科书，以及 Scriver 所著的《遗传病的代谢和分子基础》中详尽阐述，我们预料，《分子医学原理》将涉及该领域，并加强临床分子医学。

除了各章节作者和编者的献身精神和博学之外，许多人对本书的成功作出了贡献。本书的设想来自与 Victoria Reeders 的讨论，为了按时完成这一计划，Mary Kay McMahon 付出了巨大努力，做了许多超出她的责任范围

的工作。Kristina Stanfield, Patty Kalan, Joanne McAndrews,以及 William Lowe 协助充实了最新资料。我也很感谢许多同事,他们抽出时间核对和评价了部分内容。特别感谢 Human Thomas Lanigan 出版社的热情支持和导向,使本书得以完成。我还很感谢许多顾问和学生,他们引起了我对分子医学的兴趣。其中,我特别感谢 Joel Habener 和 William Crowley,我与他们共享了来自新观点和新发现的兴奋。对我个人而言,我的妻子 Michele,以及我的孩子 Ryan, Christina 和 Jimmy,不断鼓励我,使我以一个不倦的学生的方式努力工作。

J. Larry Jameson

(李洞译、白永权校)

译者的话

九九年世界图书出版公司获得美国 Humana Press 公司的授权，委托我们遴选有关学科的专家翻译 J. Larry Jameson 编著的《分子医学原理》一书。我们从我校基础医学院、第一临床医学院和第二临床医学院中聘请了 20 名专家作为该书的主译，他们在相关学科中均有较高的学术造诣和扎实的专业英语水平。其中不少专家曾出版过译著。主译确定后，由他们根据所承担的翻译任务聘请翻译人员，共有 98 人参加了本书的翻译工作。为了保证翻译质量，我们采取了严格的翻译和译稿审定程序。即，翻译人员向主译负责，主译向总主译负责，总主译向出版公司负责的原则。主译对参译人员的译稿进行全文修改和审定，总主译组织基础学科专家与临床学科专家交叉审稿，最后对主译的稿件进行抽查审定。各位主译分别负责翻译的部分如下：

楚雍烈教授负责第 4 章至第 10 章的翻译；邱曙东教授负责第 1、2、3、57、58、60、61 章的翻译；吕卓人教授负责第 11 章至第 18 章的翻译；李洞教授负责第 19 章至第 27 章的翻译；范桂香教授负责第 28 章至第 35 章的翻译；李淳成、刘珊珊教授负责第 36 章至第 40 章的翻译；张树林教授负责第 41 章至第 45 章的翻译；朱本章教授负责第 46、47、50 至 55 章、63 章的翻译；宋天保教授负责第 48、49、56 章的翻译；曹缵孙教授负责第 59、62 章的翻译；袁育康教授负责第 64 章至 71 章的翻译；肖生祥教授、彭振辉教授及刘彤教授负责第 72 章至 91 章的翻译；闫剑群教授负责第 92 章至 95 章的翻译；武成斌、尹泉潮教授负责第 96 章至第 107 章的翻译；韩太真教授负责 108 章至 111 章的翻译；岳亚飞教授、郝炳华副教授负责第 112 章至 122 章的翻译。

书中的参考文献、人名没有翻译。对国内尚没有译名的新医学术语和新药名，我们参阅有关资料及工具书，按照其在文中的意思自拟了译名。对原文和图表中少数分子生物学新词、药名等，因对译文没有把握而直接引用了原文。

站在新世纪的大门前，医学科学发展前景令人振奋。人类对生命现象的认识，对疾病的认识逐渐向微观水平、分子水平深入。人类基因组计划的实施、耳聋遗传密码在我国的破译、“基因病”概念的确立等无不给人类疾病的诊断和治疗展示了新的希望。**《分子医学原理》是美国 Doody 出版编辑基金会对 1998 年出版的 2500 多种著作中评选出的最优秀健康科学书籍。**该书各章的作者均为相关学科的知名专家，书中所综述“基因病”种类的全面，对分子遗传病理生理学论述的详尽均是当前医学书籍所少见，它弥合了

目前基础学科和临床学科之间的鸿沟，使相关分子病理生理学及遗传学的理论和技术渗透到了各种“基因病”的诊断和治疗中。我们衷心希望《分子医学原理》在国内的出版，能帮助繁忙的临床医学工作者迅速了解有关疾病的分子医学原理和治疗策略，以指导自己的临床实践及基础研究，为发展我国分子医学事业起到积极作用。

在《分子医学原理》翻译出版过程中，我们与世界图书出版西安公司进行了精诚而愉快的合作。特别是张栓才总经理和任卫军编辑对我们的翻译给予了大力的支持和指导。他们严谨的编辑作风和高速的工作效率给我们留下了深刻的印象。他们为本书迅速出版问世做出了重要贡献。

最后需要说明的是，尽管我们全体翻译人员尽了最大的努力，但由于内容新、时间紧，书中肯定还存在有错误和不妥之处。衷心希望相关学科的同行、专家和读者及时指正，以期改进。

总主译　邱曙东　袁育康

二〇〇〇年八月于

西安医科大学

目 录