



分子诊断与 肿瘤个体化治疗原则

中文翻译版

Principles of Molecular Diagnostics and Personalized Cancer Medicine

主 编 Dongfeng Tan (谈东风)
Henry T. Lynch (亨利 T. 林奇)
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北 京

图字：01-2015-1440 号

内 容 简 介

本书英文版由国际知名专家谈东风教授和 Henry T. Lynch 教授共同主编，系统、全面地阐述了肿瘤分子诊断和个体化治疗领域的常识、原则和理念。全书共 70 章，分为 7 个部分：癌症生物学以及癌症基因组学介绍；分子诊断技术；分子检测和癌症治疗；常见恶性肿瘤的分子诊断；癌症治疗的分子基础；癌症的个体化治疗与靶向治疗；分子诊断和癌症个体化治疗的未来。本书英文版作者均是来自于癌症研究、病理、临床试验、分子诊断、个体化治疗、生物信息学以及政策法规等各个领域的国际知名教授，而中文版译者也是来自于国内各大医院、科研院所以及企业单位在相关领域的工作人员，确保了整体的翻译质量。

本书可以作为肿瘤相关科室临床医生的推荐读物，可以供肿瘤分子诊断领域的科研和技术人员参考，同时也适合从事肿瘤相关药物及诊断试剂研发、生产以及销售等工作的企业人员阅读使用。

图书在版编目(CIP)数据

分子诊断与肿瘤个体化治疗原则 / (美) 谈 (Dongfeng Tan), (美) 林奇 (Henry T. Lynch) 主编; 张绪超等译. —北京: 科学出版社, 2017. 1

书名原文: Principles of Molecular Diagnostics and Personalized Cancer Medicine
ISBN 978-7-03-049862-5

I. 分… II. ①谈…②林…③张… III. 肿瘤-诊疗-研究 IV. R73

中国版本图书馆 CIP 数据核字 (2016) 第 214882 号

责任编辑: 丁慧颖 杨小玲 / 责任校对: 桂伟利 李 影 张凤琴

责任印制: 肖 兴 / 封面设计: 陈 敬

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Dongfeng Tan, Henry T. Lynch: Principles of Molecular Diagnostics and Personalized Cancer Medicine

ISBN: 978-1-4511-3197-0

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科学出版社 出版

北京东黄城根北街 16 号

邮政编码: 100717

<http://www.sciencep.com>

中国科学院印刷厂 印刷

科学出版社发行 各地新华书店经销

*

2017 年 1 月第 一 版 开本: 889×1194 1/16

2017 年 1 月第一次印刷 印张: 65 1/2 插页: 18

字数: 2 120 000

定价: 398.00 元

(如有印装质量问题, 我社负责调换)

非常感谢我的妻子 Helen，女儿 Connie 和 Christina，及我父母亲对我的支持、关心和鼓励。

Dongfeng Tan, MD, FCAP

非常感谢我的妻子 Jane 与我携手研究，以及对家庭无微不至的照顾。

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中译本序

肿瘤的精准诊疗是一部发展史，一部正在演化的发展史。

20 世纪 80 年代以来，以 SRC、KRAS、RBI 等首批癌基因或抑癌基因的发现为标志，揭开了从分子层面逐渐认识肿瘤发生、发展的序幕。

之后，基因变异的功能学研究促进了分子靶点的认识和转化应用，分子变异被用作药物研发的对象，于是出现了可药物靶向（druggable）或可作用（actionable）的分子靶点变异的概念，可见，分子医学应用于临床肿瘤学的过程中，遵循“研以致用”的原则，即我们耳熟能详的转化医学。但真正将癌症科学的新发现转化应用于患者，则是以 BCR-ABL、CD20、HER2、EGFR 等分子靶向治疗的成功为标志性事件，这些分子靶向治疗的经典在当代肿瘤学中依然熠熠生辉，照亮了前进的方向！

21 世纪初以来，由于人类基因组计划及基因组测序通量技术的发展使得人类对肿瘤相关基因变异的全面探索成为可能。“暴涨”的癌症医学知识和新兴技术的发展进一步催生了“个体化”及“精准”肿瘤学的诊疗模式。过去十余年，在肺癌中成功研发了 EGFR 突变、ALK 基因重排、ROS1 重排等重要分子亚型的诊疗方案；在黑色素瘤中也成功研发了 BRAF 突变型及免疫靶点的靶向治疗方案等。肿瘤突变新抗原、嵌合抗原受体 T 细胞（CART）等新技术新方法的出现，更将进一步推动临床免疫治疗的精准性。

当今，临床肿瘤学仍处于分子标志物驱动的快速发展时期。其中，尤其突出的是分子诊断技术、新药创制及令人眼花缭乱的临床方案研究。精准医学需要诊断先行，即分子靶点诊断的精准性决定了治疗的精准性。在我国，临床精准医疗中的分子诊断已广泛开展，但尚处于逐步规范的时期。2015 年我国 EGFR 突变的受检率在肺癌患者群中约为 45%，较之以往有很大进步，但仍存在很多不足。随着分子分型和靶向治疗方案的进步，更需要对 ALK、MET、ROS1 等更多的基因进行检测和规范应用。其他多个瘤种及相关分子靶点的临床检测同样需要标准化和规范应用。中国临床肿瘤学会（CSCO）也正在致力于制订、推广临床肿瘤分子标志物在我国的应用规范或共识，并开展相关的分子标志物或分子靶点驱动的多药临床研究。

值此分子肿瘤学蓬勃发展之际，来自美国 MD 安德森肿瘤中心的华人病理学家、肿瘤学家谈东风教授联合 Lynch 综合征的开拓者、克莱顿大学医学院的 Henry T. Lynch 教授出版了《分子诊断与肿瘤个体化治疗原则》（*Principles of Molecular Diagnostics and Personalized Cancer Medicine*）一书。这本书涵盖了癌症分子生物学基础、各类分子靶点变异的检测技术和通量分析技术、临床常见肿瘤的分子诊断与个体化治疗及未来的技术展望等。阅读这本书有助于对肿瘤的分子基础和当代个体化精准诊疗的理解，特别是对分子标志物检测技术在临床的合理应用提供了指导性认识。该书由 CSCO 肿瘤标志物专家委员会张绪超、刘毅等组织数十位我国肿瘤学界的专家共同翻译成中文，有助于扩大在我国的读者群，有助于当代肿瘤分子医学知识的推广和应用，也能促进肿瘤分子标志物的转化性研究。

祝贺该书中文版的出版！诗云：他山之石，可以攻玉。衷心希望我国的肿瘤学临床和科研人员能够多汲取这类书籍的知识，这有助于自身的临床实践与转化性研究水平的提高，更有助于推动我国临床肿瘤学事业的发展！

吴一龙
2016 年 9 月

译者前言

2015年1月，美国总统奥巴马在国情咨文中提出了“精准医学计划”并于当月正式启动，一个全新的精准医学时代由此而开启。美国国立卫生研究院在其官网的介绍中提到，所谓精准医学指的是疾病预防与处置的新方法，该方法需要考虑患者在基因、环境以及生活习惯方面的个体差异。毫无疑问，肿瘤的诊治是“精准医学计划”的重中之重，这是因为肿瘤是异质性最为明显的人类疾病，患者之间存在各不相同的生物学特征，最应该在治疗上做区别对待。近年来，随着越来越多临床证据的出现，通过生物标志物的分子诊断来指导患者个体化的精准治疗，已逐渐成为肿瘤临床治疗领域的共识，一批与早期筛查、预后判断、疗效预测以及疗效评价等密切相关的生物标志物已经被引入国内外多种肿瘤的临床治疗实践之中。肿瘤生物标志物已经成为未来肿瘤治疗的主要方向和实现个体化精准治疗的最有力武器。

中国临床肿瘤学会（CSCO）的肿瘤生物标志物专家委员会于2011年成立，由吴一龙教授和秦叔逵教授共同担任主任委员，旨在切实推动肿瘤临床生物标志物在我国的发展和运用、促进临床应用的规范性和普及，从而提高我国肿瘤诊治的医学水平，造福中国数以百万计的癌症患者。近年来，该委员会完成了大量卓有成效的工作，制定了包括EGFR、ALK等基因在内的检测指南，推出了二代测序技术应用于临床肿瘤精准诊治的共识等，有力地推动了肿瘤分子诊断检测的开展。然而，我们也清醒地认识到，针对我国肿瘤患者的生物标志物的发现与验证、新药物靶点的鉴定、疗效预测标志物的确立、检测方法学的建立和临床应用的标准化、标志物临床实践的规范化，均尚有许多建设性的工作需要携手来完成。特别是生物标志物的临床应用规范、新技术方法的标准化、检测人员的能力培养等现状仍然严重制约着肿瘤标志物在临床应用的普及化和标准化。这些问题亟须肿瘤学界全体同仁的共同努力并携手解决。

本书原著由谈东风教授和Henry T. Lynch教授主编，Lippincott Williams & Wilkins公司出版。两位教授均是肿瘤分子诊断和个体化治疗领域的国际知名专家，而本书各个章节的作者均是来自于癌症研究、病理、临床试验、分子诊断、个体化治疗、生物信息学以及政策法规等各个领域的国际知名教授，他们分别从癌症生物学和遗传学的综合知识、分子诊断技术、分子检测和癌症治疗、各个癌种的分子诊断、各个癌种的个体化医疗和靶向治疗以及分子诊断和个体化医疗的未来展望等7个不同的领域对肿瘤分子诊断和个体化治疗的原则做了详尽的介绍和说明，对于普及肿瘤分子诊断和个体化治疗的理念和规范具有重要的参考价值。

在本书的翻译过程之中，得到了业内同行的积极响应和大力支持，大部分译者均是来自于各大医院、科研院所以及企业单位相关领域的工作人员，他们在各自的领域均已有较好的建树和积累。感谢这些同道的共同参与，正是大家辛苦的努力，才使得本书得以顺利地出版。我们还要特别感谢医脉通公司在本书的译者组织、统筹协调以及组织翻译等方面所做的出色工作，他们的辛苦付出，是本书得以出版的重要保证。但是，由于本书所涉及的内容繁多，而且均是分子诊断和肿瘤个体化医疗领域的最新进展，虽经反复校审，仍无法完全保证没有疏漏和不当地方，恳请各位读者朋友能予以批评和指正。