

Mc
Graw
Hill

Education

产前诊断

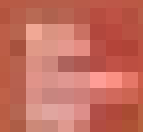
Prenatal Diagnosis



原著 Mark I. Evans Mark P. Johnson
Yuval Yaron Arie Drugan
主译 段 涛 胡娅莉 吕时铭

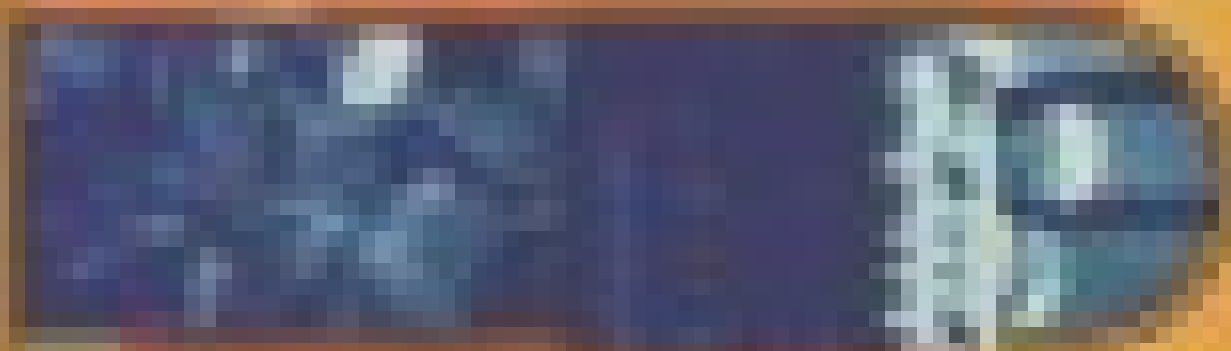


人民卫生出版社



产前诊断

Prenatal Diagnosis



主编 王卫平 副主编 王卫平 王卫平
编委 王卫平 王卫平 王卫平
王卫平 王卫平 王卫平

人民卫生出版社

产 前 诊 断

Prenatal Diagnosis

原著 Mark I. Evans Mark P. Johnson
Yuval Yaron Arie Drugan

主译 段 涛 胡娅莉 吕时铭



人民卫生出版社

Mark I. Evans, Mark P. Johnson, Yuval Yaron, Arie Drugan

Prenatal Diagnosis

ISBN: 0-8385-7682-6

Copyright © 2006 by The McGraw-Hill Companies

All Rights reserved. No part of this publication may be reproduced or transmitted in any form or by any means, electronic or mechanical, including without limitation photocopying, recording, taping, or any database, information or retrieval system, without the prior written permission of the publisher.

This authorized Chinese translation edition is jointly published by McGraw-Hill Education (Asia) and People's Medical Publishing House. This edition is authorized for sale in the People's Republic of China only, excluding Hong Kong, Macao SAR and Taiwan.

Copyright © 2010 by McGraw-Hill Education (Asia), a division of the Singapore Branch of The McGraw-Hill Companies, Inc. and People's Medical Publishing House.

版权所有。未经出版人事先书面许可，对本出版物的任何部分不得以任何方式或途径复制或传播，包括但不限于复印、录制、录音，或通过任何数据库、信息或可检索的系统。

本授权中文简体字翻译版由麦格劳-希尔（亚洲）教育出版公司和人民卫生出版社合作出版。此版本经授权仅限在中华人民共和国境内（不包括香港特别行政区、澳门特别行政区和台湾）销售。

版权© 2010 由麦格劳-希尔（亚洲）教育出版公司与人民卫生出版社所有。

本书封面贴有 McGraw-Hill 公司防伪标签，无标签者不得销售。

北京市版权局著作权合同登记号：01-2007-1126

图书在版编目 (CIP) 数据

产前诊断/段涛等主译. —北京：人民卫生出版社，

2010.4

ISBN 978-7-117-12535-2

I. ①产… II. ①段… III. ①妊娠诊断

IV. ①R714.15

中国版本图书馆 CIP 数据核字 (2009) 第 231793 号

门户网: www.pmph.com	出版物查询、网上书店
卫人网: www.ipmph.com	护士、医师、药师、中医师、卫生资格考试培训

版权所有，侵权必究！

产 前 诊 断

主 译: 段 涛 胡娅莉 吕时铭

出版发行: 人民卫生出版社 (中继线 010-59780011)

地 址: 北京市朝阳区潘家园南里 19 号

邮 编: 100021

E - mail: pmph@pmph.com

购书热线: 010-67605754 010-65264830

印 刷: 中国农业出版社印刷厂

经 销: 新华书店

开 本: 889×1194 1/16 印张: 44 字数: 1359 千字

版 次: 2010 年 4 月第 1 版 2010 年 4 月第 1 版第 1 次印刷

标准书号: ISBN 978-7-117-12535-2/R · 12536

定 价: 130.00 元

打击盗版举报电话: 010-59787491 E-mail: WQ@pmph.com

(凡属印装质量问题请与本社销售中心联系退换)

原 著

Ernest L. Abel, MD

Professor
Departments of Obstetrics/Gynecology
and Psychology
C.S. Mott Center for Human Growth
and Development
Wayne State University
Detroit, Michigan

Jill Ablett, MD

Westmead Hospital
Westmead New South Wales
Australia

N. Scott Adzick, MD

Professor and Chairman
Department of Surgery
Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

Svetlana Arbuzova, MD

Interregional Medico-Genetic Center
Central Hospital
Donetsk, Ukraine

D. Randall Armant, PhD

C.S. Mott Center for Human Growth
and Development
Professor, Department of Obstetrics/Gynecology and
Anatomy and Cell Biology
Wayne State University School of Medicine
Detroit, Michigan

Gerard Barki

Adviser
Karl Storz GmbH and Co.
Tuttlngen, Germany

David A. Beckman

Jefferson Medical College
Philadelphia, Pennsylvania

Mary P. Bedard, MD

Children's Hospital of Michigan
Detroit, Michigan

Beryl R. Benacerraf, MD

Diagnostic Ultrasound Associates
Boston, Massachusetts

Richard L. Berkowitz, MD

Professor of Obstetrics and Gynecology
Columbia University College of Physicians
and Surgeons
New York, New York

Ryan Blackwell

University Women's Care
Detroit, Michigan

Sean C. Blackwell, MD

Assistant Professor and Medical Director
of Informatics
Hutzel Hospital
Wayne State University
Detroit, Michigan

Pamela A. Boland, JD

Kitch Drutchas Wagner Valitutti and Sherbrook
Detroit, Michigan

Robert L. Brent, MD, PhD

Distinguished Professor of Pediatrics, Radiology,
and Pathology
Jefferson Medical College
Philadelphia, Pennsylvania
Head of Clinical Environmental and Teratology
Laboratory
Alfred I. DuPont Hospital for Children
Wilmington, Delaware

David W. Britt, PhD

Fetal Medicine Foundation of America
New York, New York

Bryan Bromley, MD

Associate Clinical Professor of Obstetrics, Gynecology,
and Reproductive Biology
Vincent Memorial Obstetrical Ultrasound Division
Massachusetts General Hospital
Boston, Massachusetts

Peter K. Bryant-Greenwood, MD, MBA

Clinical Proteomics Program
Department of Pathology and
Obstetrics/Gynecology
John A. Burns School of Medicine
Honolulu, Hawaii

The-Hung Bui, MD

Department of Molecular Medicine
Karolinska Hospital
Stockholm, Sweden

Bryon Calhoun, MD, FACOG, FACS, MBA

Professor of Obstetrics and Gynecology
Vice Chair Obstetrics and Gynecology
West Virginia University
Charleston, West Virginia

Darrell L. Cass, MD

Department of Pediatric Surgery
Baylor Medical School
Houston, Texas

Frank A. Chervenak, MD

Given Foundation Professor and Chairman
Obstetrician and Gynecologist In-Chief
Department of Obstetrics and Gynecology
Weill Medical College of Cornell University
New York, New York

Doina Ciorica, RDMS, MD

Maternal Fetal Medicine Associates
New York, New York

Joshua A. Copel, MD

Professor
Department of Obstetrics and
Gynecology and Reproductive Sciences
Section of Maternal-Fetal Medicine
Yale University School of Medicine
New Haven, Connecticut

Howard S. Cuckle, MD

University of Leeds
Institute of Epidemiology and Health Services
Research
Research School of Medicine
Leeds, England
United Kingdom

Jan Deprest, MD, PhD

Unit of Fetal Diagnosis and Therapy and Gynecological
Ultrasound
Department of Obstetrics and Gynecology
University Hospital Gasthuisberg
Leuven, Belgium

Russell L. Deter, MD

Professor, Obstetrics and Gynecology
Baylor College of Medicine
Houston, Texas

Jan E. Dickinson, MBBS, MD

Associate Professor, Maternal Fetal Medicine
School of Women's and Infants Health
University of Western Australia
Maternal Fetal Medicine Specialist
King Edward Memorial Hospital for Women
Perth, Western Australia

Rony Diukman, MD

Tel Aviv, Israel

Mitchell Dombrowski, MD

Chair, Obstetrics and Gynecology
St. John's Hospital
Detroit, Michigan

Alan E. Donnenfeld, MD

Women's Health Care Group of Pennsylvania Main
Line Perinatal Associates Division
Lankenau Hospital
Wynnewood, Pennsylvania

Arie Drugan, MD

Director, Genetics
Department of Obstetrics and Gynecology

Ram Bam Medical Center
Haifa, Israel

Sherman Elias, MD

Professor and Chairman
Department of Obstetrics and Gynecology
Northwestern University
Chicago, Illinois

Mark I. Evans, MD

President, Fetal Medicine
Foundation of America
Director, Comprehensive Genetics
Professor of Obstetrics and Gynecology
Mt. Sinai School of Medicine
New York, New York

Lynda Fawcett, PhD

Assistant Professor of Pediatrics
Department of Pediatrics
Alfred I. DuPont Hospital for Children
Wilmington, Delaware

Baruch Feldman, MD

Kfar Saba Medical Center
Kfar Saba, Israel

Charles W. Fisher, JD

Kitch Drutchas Wagner Valitutti and Sherbrook
Detroit, Michigan

Alan W. Flake, MD

Department of Pediatric Surgery
Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

John C. Fletcher, MD

(Deceased)

Robert S. Galen, MD

Professor and Interim Head
Department of Health Administration, Biostatistics,
and Epidemiology
The University of Georgia College of Medicine
Athens, Georgia

Natalie Gellman, PhD

Psychologist/Director
Humanistic Resources
Warren, Maine

James D. Goldberg, MD

San Francisco Perinatal Associates
San Francisco, California

Bernard Gonik, MD

Department of Obstetrics and Gynecology
Sinai Grace Hospital
Detroit, Michigan

Eduardo Gratacos, MD

Professor
Fetal Medicine and Therapy Programme
Department of Obstetrics
Hospital Clinic Barcelona
Universitat de Barcelona
Barcelona, Spain

原 著

Anne Greb, MS

Genetic Counselor
Assistant Professor
Director, Genetic Counseling Graduate Program
Wayne State University School of Medicine
Center for Molecular Medicine and Genetics
Detroit, Michigan

Anthony R. Gregg, MD

Department of Obstetrics and Gynecology
Medical College of South Carolina
Columbia, South Carolina

Edith D. Gurewitsch, MD

Assistant Professor of Gynecology and
Obstetrics
Division of Maternal Fetal Medicine
Johns Hopkins University School of Medicine
Baltimore, Maryland

Sinhue Hahn, MD

University of Basel
Department of Obstetrics and Gynecology
University Women's University
Basel, Switzerland

Ruth S. Hanft, PhD

Adjunct Associate
Center for Biomedical Ethics
University of Virginia
Charlottesville, Virginia

John H. Hannigan, PhD

C.S. Mott Center for Human Growth
and Development
Wayne State University
Department of Obstetrics and Gynecology
Detroit, Michigan

Michael R. Harrison, MD

Professor of Surgery, Pediatrics, and Obstetrics,
Gynecology and Reproductive Sciences
Director, Fetal Treatment Center
Division of Pediatric Surgery
Department of Surgery
Fetal Treatment Center
University of California, San Francisco
San Francisco, California

John C. Hobbins, MD

Professor of Obstetrics and Gynecology
University of Colorado Health Sciences
Center
Denver, Colorado

Eric P. Hoffman, MD

Children's National Medical Center
Washington, DC

Wolfgang Holzgreve, MD

Professor and Chair
University of Basel
Department of Obstetrics and Gynecology
University Women's Hospital
Basel, Switzerland

Roderick F. Hume, Jr., MD

Hutzel Women's Hospital
Wayne State University
Detroit, Michigan

Jon Hyett, MD

King's College
Denmark Hill
London, England
United Kingdom

Suzanne M. Jacques, MD

Department of Pathology
Hutzel Hospital
Detroit, Michigan

Donna D. Johnson, MD

Assistant Professor
Department of Obstetrics and Gynecology
Medical University of South Carolina
Charleston, South Carolina

Mark Paul Johnson, MD

Associate Professor of Obstetrics and
Gynecology and Pediatric Surgery
Children's Hospital of Philadelphia
University of Pennsylvania
Philadelphia, Pennsylvania

Heung Bae Kim, MD

Department of Pediatric Surgery
Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

Charles S. Kleinman, MD

Professor of Pediatric Cardiology
Columbia University
New York, New York

Daniela Koch, MD

Rambam Medical Center
Haifa, Israel

Bruce R. Korf, MD, PhD

Professor and Chairman, Department of
Genetics
University of Alabama at Birmingham
Birmingham, Alabama

Christine Kovac, MD

Department of Obstetrics and Gynecology
Madigan Army Medical Center
Tacoma, Washington

Deborah Krakow, MD

Cedars Sinai Hospital
Los Angeles, California

Ralph L. Kramer, MD

Stuart Ob Gyn
Stuart, Virginia

Eric L. Krivchenia, MS

Genetic Counselor
Cherry Hill, New Jersey

Sanja Kupesic, MD

Ultrasonic Institute
University of Zagreb
Zagreb, Croatia

Asim Kurjak, MD

Ultrasonic Institute
University of Zagreb
Zagreb, Croatia

Allen N. Lamb, PhD

Laboratory Director
Molecular Cytogenetics
Genzyme Genetics
Santa Fe, New Mexico

Ellen J. Landsberger, MD

Department of Obstetrics and Gynecology
Albert Einstein College of Medicine
Bronx, New York

Zvi Leibovitz

Genetics Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

Pamela Lewis-Matia, MD

Greater Washington Maternal Fetal Medicine and Genetics
Rockville, Maryland

Elisa Llurba, MD

Fetal-Maternal Unit
Obstetrics and Gynecology Department
Vall d'Hebron Hospital
Barcelona, Spain

James N. Macri, PhD

Laboratory Director
NTD Laboratories, Inc.
Huntington Station, New York

Laura S. Martin, MD, FAAP, FACMG

Director, Medical Genetics
Rockford Health System
Department of Medical Genetics
Rockford, Illinois

Donald R. Mattison, MD

Senior Advisor
National Institute of Health
Bethesda, Maryland

Laurence B. McCullough, PhD

Professor of Medicine and Medical Ethics
Center for Medical Ethics and Health Policy
Baylor College of Medicine
Houston, Texas

Heather Shane Michaels, MS, MHS, CGC

Genetic Counselor
Genzyme Genetics
New York, New York

Ana Monteagudo, MD

Associate Professor
Department of Obstetrics and Gynecology
New York University Medical Center
New York, New York

George B. Mychaliska, MD

Assistant Professor of Surgery and Obstetrics and Gynecology
Division of Pediatric Surgery
University of Michigan
Ann Arbor, Michigan

Shivani B. Nazareth, MS, CGC

Department of Obstetrics and Gynecology
St. Luke's—Roosevelt Hospital
New York, New York

Kypros Nicolaides, MD

Harris Birthright Research Centre for Fetal Medicine
King's College, London University
London, England
United Kingdom

Beth A. Nordstrom, PhD

Department of Family Medicine
James H. Quillen College of Medicine
Eastern Tennessee State University
Johnson City, Tennessee

Avi Orr-Urtreger

Prenatal Genetic Diagnosis Unit
Genetic Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

**Wayne H. Persutte, MD, RDMS,
FAIUM, FSDMS**

Director of Sonography
Platte River Perinatal Center
Department of Obstetrics and Gynecology
University of Colorado Health Science Center
Denver, Colorado

Lawrence D. Platt, MD

Professor of Obstetrics and Gynecology
David Geffen School of Medicine at UCLA
Department of Obstetrics and Gynecology
Los Angeles, California

Dolores H. Pretorius, MD

Professor
Department of Radiology
University of California, San Diego
La Jolla, California

Peter G. Pryde, MD

University of Wisconsin
Madison, Wisconsin

Faisal Qureshi, MD

Department of Pathology
Hutzel Hospital
Detroit, Michigan

Charles H. Rodeck, MD

Head, Department of Obstetrics and Gynecology
University College and Middlesex
School of Medicine
University College, London
London, England
United Kingdom

Kevin P. Rosenblatt, MD, PhD

Department of Pathology
Division of Translational Pathology
University of Texas Southwestern Medical Center
Dallas, Texas

Guy Rosner, MD

Prenatal Diagnosis Unit
Genetic Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

Jack Rychik, MD

Director, Pediatric Cardiology
Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

Aimen F. Shaaban

Genetic Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

Shay Ben Shahr

Prenatal Genetics Diagnosis Unit
Genetic Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

Seetha Shankaran, MD

Children's Hospital of Michigan
Detroit, Michigan

Chris Shimizu, MD

Hawaii Pathologists Laboratory
The Queens Medical Center
Honolulu, Hawaii

Jana K. Silva, MD

Assistant Professor
Department of Native Hawaiian Health
Native Hawaiian Center of Excellence
Honolulu, Hawaii

Robert J. Sokol, MD

Distinguished Professor
C.S. Mott Center for Human Growth and Development
Department of Obstetrics and Gynecology
Wayne State University School of Medicine
Detroit, Michigan

Lorien Tambini, MS, CGC

Genetic Counselor
Department of Obstetrics and Gynecology
Division of Genetics
St. Luke's—Roosevelt Hospital
New York, New York

Carol Tarnowsky

Assistant General Counsel
St. Joseph Mercy Oakland
Pontiac, Michigan

Marjorie C. Treadwell, MD

Professor, Obstetrics and Gynecology
Division of Maternal-Fetal Medicine
Director, Obstetric Ultrasound
Center for Fetal Diagnosis and Therapy
Hutzel Women's Hospital

Wayne State University
Detroit, Michigan

Ilan E. Timor-Tritsch, MD

Professor of Obstetrics and Gynecology
Department of Obstetrics and Gynecology
New York University Medical Center
New York, New York

Brian Trudinger, MD

Westmead Hospital
Westmead New South Wales
Australia

Dominique van Schoubroeck, MD

Unit of Fetal Diagnosis and Therapy and Gynecological
Ultrasound
Department of Obstetrics and Gynecology
University Hospital Gasthuisberg
Leuven, Belgium

Ronald J. Wapner, MD

Professor of Obstetrics and Gynecology
Columbia University College of
Physicians and Surgeons
New York, New York

Charles Weber, MD

Madigan Army Medical Center
Tacoma, Washington

Carl P. Weiner, MD

Professor and Chairman of Obstetrics and Gynecology
University of Kansas Medical Center
Kansas City, Kansas

Jane Wegner, MD

Waukesha, Wisconsin

R. Douglas Wilson, MD

Department of Pediatrics General and Thoracic Surgery
The Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

Yuval Yaron, MD

Director, Prenatal Genetic Diagnosis Unit
Genetic Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

Yoav Ben-Yoseph, PhD

Retired
Wayne State University School of
Medicine
Detroit, Michigan

Ivan Zador, PhD

Fetal Medicine Foundation of America
Los Angeles, California

Yaron Zalel, MD

Genetic Institute
Tel Aviv Sourasky Medical Center
Tel Aviv, Israel

译者序

产前诊断方面缺乏好的专著,一般可以在围产医学、胎儿医学、超声学或是遗传学专著中找到部分相关内容。但是这些内容往往不够全面,而且太分散。还好有这本《产前诊断》,Mark Evans 的这本专著是从事这一领域研究和临床工作的人不得不阅读的一本书,它几乎覆盖了产前诊断相关的所有内容。你可以把它当作教科书,也可以把它作为研究指南,还可以把它当作临床指导手册。这本书可以卖多少本我不能肯定,但是有一点我可以保证,就是这本书你不可能只读一次,你肯定还会不断地拿出来查阅、对照,甚至带着这本书上门诊;而且阅读这本书的人不可能只是你一个人,你的同事、学生甚至老师都可能会来向你借阅,你也会向他们推荐的。

在从事妇产科工作的同行中一直有一个误区,大多数人(包括多数的产科医生)都认为做产科不如做妇科。做妇科肿瘤既可以做手术,又可以做好的科学研究,产科不过是一个高级接生婆而已。我的看法恰恰相反,我经常和做妇科肿瘤的著名教授朋友们开玩笑说,“你们现在号称做妇科肿瘤的医生比妇科肿瘤病人还要多,拿了这么多科研经费,做了这么多研究,发表了这么多的 SCI 文章,开了这么多刀,也没有见妇科恶性肿瘤病人的五年生存率能提高多少”(玩笑而已,五年生存率还是有所提高的!)。我们产科的进展比妇科多得多,产前诊断是产科领域进展最快的,整个临床超声的进步要以胎儿超声最大,是胎儿超声引领了整个临床超声的发展。遗传性疾病的产前诊断是和基因组研究的进步同步发展的,胎儿宫内手术要比妇科内镜手术复杂得多,胎儿宫内基因治疗已经显现出不可估量的前景,临床遗传学里面发展最快的还是产前诊断。这些内容在《产前诊断》这本书里面都有很好的体现。

你可能没有听说过 Mark Evans 这个人,不知道他是何方神圣,听我说过以后你可能会忘记我是谁,但是绝对不会忘记他。Mark Evans 是在纽约工作的一位教授,也是我相识多年的一位朋友,他是一个产科医生 - 遗传学家,这是从事产前诊断的最佳人选(只是产科医生不行,只是遗传学家也不行),的确他做得也很好。他是美国胎儿医学基金会的主席,既从事临床又从事科学研究,是个重量级的专家(PS: 不仅仅是指学术,他的体重也是我体重的数倍!),他究竟有多好,在这里卖个关子,看了正文你就知道了!

在此书的翻译过程中,除了主译们倾注了大量心血进行审阅之外,傅静和熊诗诣两位博士也对全文进行了审校,特此表示诚挚的感谢!

段 涛

2010 年 3 月于上海

序 言

这真是一本“大书”，无论是从厚度还是从覆盖的内容，都令人印象深刻！考虑到 40 年前，这只能是很小的一本书，甚至是写不出什么内容。在那时候人类细胞遗传学才刚刚开始，直到 1967 年羊膜腔穿刺才开始被用于遗传性疾病的产前诊断，超声只是用于发现中线的偏移，还没有把胎儿作为病人进行讨论。然而 40 年以后，需要写 68 个章节来全面覆盖产前诊断及其相关内容。即使只是作为遗传学革命的一个部分，这已经是相当了不起的发展，医学领域的发展中只有很少一些内容能与之相比。

写一本这一领域的书需要一定的勇气，幸好有 Mark Evans, MD, 在他 1992 年的那本书《生殖风险和产前诊断》的基础上进行了扩展，与他的 3 个学生一起完成了这个任务。这本书对这一领域来讲是一个很大的贡献，它告诉我们在这一领域的科学进展有多大。这些作者在各自的领域中都是非常了不起的权威，为读者呈现了整个产前诊断领域的现状。

John Fletcher 肯定会喜欢献给他的这本书，他的背景是神学，作为生物伦理学家（第一个），他在 NIH 的临床研究中心积累了无可比拟的经验，然后他又关注于生殖健康和母胎医学领域的生物伦理学问题，他在这一领域的发展和进步方面起了非常重要的作用。John Fletcher 坚信，不仅仅好的伦理学是从好的科学开始的，而且做出一个正确的伦理学决定需要对科学和医学实践有很好的理解。他肯定会仔细阅读此书，几乎每一页都会提出新的伦理问题，需要仔细的考虑。随着他的注意力逐渐转移到产前诊断和妊娠期的决定选择，他提供了两个非常有价值的贡献。首先，他是这一领域的主持正义者，他不断提出尖锐的问题，问一些有时过于大胆的围产学者他们究竟在做什么？是如何做的？让大家明白母亲 / 孕妇是最终的做决定的人。他在这方面扮演的角色非常重要，可以帮助临床医生和研究者避免这一领域的许多潜在问题，本来这些努力是出于良好愿望的，但是如果不能够审慎处理，就会带来很坏的名声。其次，他是这一领域的倡导者，他指出为了做出正确的决定和行动，需要进行研究来获得相应的知识。他喜欢与科学家、医生、鼓吹者、政客就伦理问题进行争论，但是在开始之前他总是要确认自己理解科学。

在阅读本书时，读者们应该像 John Fletcher 那样努力学习和理解科学，并以此与病人一起在伦理决策过程中作出最符合伦理的选择，为胎儿、婴儿、和母亲提供最好的服务。

Duane Alexander, MD

Director of National Institute of Child Health and Human Development

National Institutes of Health

Bethesda, Maryland

前 言

为《生殖风险和产前诊断》这本书写前言(参见下文)已经是 15 年以前的事情了,这些原则经历住了时间的考验。在 1992 年我就已经感叹在过去十几年当中生殖遗传学发生的巨大变化,这种情况不但继续,而且是以更快的速度在发生变化。进展的重点为,20 世纪 70~80 年代是临床技术,到了 90 年代是影像学进展,现在的最大变化是在实验室。这些新的工具为临床医生提供了新的武器,可以更早、更可靠、以更复杂的手段来诊断和治疗遗传疾病和先天异常。

这些新的技术对于传统的研究机构来讲是相当大的挑战,他们往往无法在基础设备上投资来应对我们这一领域的大量研究努力。因此,很多研究都不在这些传统的中心进行,这一趋势在以后的十几年里还会继续下去。

《生殖风险和产前诊断》的前言(1992)

我依然记得我在医学院第一天的情形,院长发表了比较标准的演讲,也就是在未来四年里我们学习的东西有一半会是错误的,但是他不知道哪一半是错误的。可能在医学领域里面没有一个领域像生殖遗传领域的不确定性和易变性这么大,遗传学领域里的许多基本原则在过去十几年里都被证实是错误的。对于许多临床医生来讲,除了医学院学习的那些内容,可能几乎没有再学习过遗传学。这些巨大的改变很混乱,这个领域看上去像外星人。本书的目的就是以一种易懂、可再述、临床容易应用的方式介绍胎儿异常诊断的新进展。本书的开篇介绍一些基本原则,之后是其在临床检查中的应用、实验室技术的应用,最后是对有问题胎儿的处理。尽管每个章节都是独立的,实际上它都为下一个章节打下基础,使得下一章节更容易理解。

目 录

第一部分 遗传学和生殖风险

第 1 章	经典遗传学原理·····	3
第 2 章	非整倍体的流行病学·····	18
第 3 章	遗传学新概念·····	33
第 4 章	染色体异常·····	41
第 5 章	产前诊断中的孟德尔遗传学·····	50
第 6 章	综合征:研究胎儿畸形的方法 ·····	55
第 7 章	基因与发育·····	61
第 8 章	产前遗传咨询·····	68
第 9 章	畸形学原理·····	76
第 10 章	药物 ·····	107
第 11 章	饮酒对孕期的影响 ·····	117
第 12 章	环境和职业对生殖和发育影响的评估 ·····	123
第 13 章	妊娠期细菌感染 ·····	139
第 14 章	病毒因素和生殖风险 ·····	144
第 15 章	妊娠期电离辐射暴露 ·····	156
第 16 章	妊娠期化疗 ·····	163

第二部分 超声诊断和筛查

第 17 章	胎儿全面超声检查:一种诊断和处理胎儿畸形的方法·····	173
第 18 章	胎儿脑图像 ·····	184
第 19 章	心血管畸形 ·····	210
第 20 章	胎儿腹部与腹壁正常和异常所见 ·····	220
第 21 章	泌尿生殖道畸形 ·····	236
第 22 章	胎儿骨骼发育不良 ·····	242
第 23 章	生化筛查 ·····	255
第 24 章	妊娠早期胎儿颈项透明层的超声筛查 ·····	266
第 25 章	妊娠中期非整倍体染色体异常的超声指标 ·····	285
第 26 章	先天性畸形的彩色多普勒 ·····	304

第 27 章	妊娠中期彩色多普勒和胎儿畸形	314
第 28 章	多胎妊娠的超声检查	319
第 29 章	三维超声在产前诊断中的应用	327
第 30 章	胎儿神经系统的三维超声	334
第 31 章	计算机和超声	351
第 32 章	胎龄确定和生长发育评估在产前诊断中的作用	354
第 33 章	胎盘的超声诊断	372

第三部分 操 作

第 34 章	羊膜腔穿刺术	381
第 35 章	早期羊膜腔穿刺术:风险评估.....	388
第 36 章	绒毛取样	398
第 37 章	脐带穿刺术	407
第 38 章	组织活检	412
第 39 章	胎儿皮肤采样及重症遗传性皮肤病的产前诊断	418
第 40 章	手术性胎儿镜	421

第四部分 实验室诊断

第 41 章	细胞遗传学和分子细胞遗传学	437
第 42 章	生化遗传学	446
第 43 章	产前分子诊断学	453
第 44 章	分子筛查	461
第 45 章	使用来源于母血的胎儿细胞进行产前诊断	464
第 46 章	临床蛋白质组学	471

第五部分 问 题 处 理

第 47 章	产前诊断和流产的心理学反应	481
第 48 章	胎儿异常的咨询	486
第 49 章	终止妊娠	494
第 50 章	胎儿尸检	505
第 51 章	多胎妊娠的减胎术	516
第 52 章	选择性终止妊娠	525
第 53 章	胎儿异常的产前处理	533
第 54 章	先天畸形新生儿的评估与处理	548

第六部分 胎 儿 治 疗

第 55 章	胎儿分流手术	557
第 56 章	胎儿手术治疗先天性膈疝的演变过程	568
第 57 章	胎儿开放性手术:先天性囊性腺瘤样畸形.....	577
第 58 章	孟德尔遗传病的胎儿药物治疗	583
第 59 章	叶酸与神经管缺陷的预防	590
第 60 章	胎儿心血管系统评估	600
第 61 章	产前心脏治疗	616
第 62 章	胎儿基因治疗——体细胞	627
第 63 章	胎儿基因治疗——干细胞	636

第七部分 伦理、法律和社会问题

第 64 章	产前诊断中的社会心理问题	645
第 65 章	伦理和产前诊断:多文化交汇的影响.....	651
第 66 章	胎儿治疗学的伦理规范	664
第 67 章	基因诊断与遗传咨询的法律问题	669
第 68 章	政治与遗传生育风险	682

第一部分

遗传学和生殖风险

