

# MOLECULAR GENETIC PATHOLOGY

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*Edited by*

LIANG CHENG, MD

*Professor of Pathology and Urology  
Director of Molecular Pathology Laboratory  
Chief of Genitourinary Pathology Division  
Department of Pathology and Laboratory Medicine  
and Clarian Pathology Laboratory  
Indiana University School of Medicine, Indianapolis, IN*

DAVID Y. ZHANG, MD, PhD, MPH

*Associate Professor of Pathology  
Director, Molecular Pathology Laboratory  
Department of Pathology  
Mount Sinai School of Medicine  
and the Mount Sinai Medical Center  
New York, NY*

*Editors*

Liang Cheng  
Department of Pathology and Laboratory Medicine  
and Clarian Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN

David Y. Zhang  
Department of Pathology  
Mount Sinai School of Medicine  
and the Mount Sinai Medical Center  
New York, NY

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# Preface

We have had the opportunity to witness both the beginning and the subsequent growth of an exciting specialty that combines both pathology and medical genetics, a field commonly referred to as molecular genetic pathology. The birth of this specialty took place in 1988 when Kari Mullis developed a new DNA amplification technology called the polymerase chain reaction (PCR). Within a few years, this technology was no longer being used exclusively in research laboratories. The technique had found numerous new applications in clinical medicine as a tool for diagnosis and diseases monitoring. The use of PCR technology has greatly expanded the specialties of anatomic and clinical pathology and has increased the availability of genetic testing in the clinical setting. We expect that such advances as the completion of the Human Genome Project, the maturation of pharmacogenomics, the growth of proteomics, and the rapidly growing field of molecular genetic pathology will lead to a new era of personalized and customized patient care.

More recently, the American Board of Pathology (ABP), in conjunction with the American Board of Medical Genetics (ABMG), established a new subspecialty, molecular genetic pathology. Fellowship training for molecular genetic pathology is approved by the Accreditation Council for Graduation Medical Education (ACGME). Many pathologists and medical geneticists are applying for advanced training in this growing subspecialty. Training in molecular pathology is also becoming a required element in pathology residency curricula. To meet these demands, a team of more than 50 leading experts has compiled this quick reference book for medical students, general practitioners, medical technologists, pathologists, and medical geneticists. We also hope that residents or fellows who are training in pathology and medical genetics will find this book helpful in their preparation for board examinations.

*Molecular Genetic Pathology* contains two parts. Part I covers general molecular genetic pathology and technology, including principles of clinical molecular biology, principles

of clinical cytogenetics, diagnostic methodology and technology, tissue microarrays and biomarker validation, laser capture microdissection, clinical flow cytometry, conceptual evolution in cancer biology, clinical genomics in oncology, clinical proteomics, clinical pharmacogenomics, clonality analysis in surgical pathology, fluorescence *in situ* hybridization (FISH), conventional cytogenetics for hematology and oncology diagnosis, instrumentation, genetic inheritance and population genetics, and genetic counseling. Part II provides disease-based information, including prenatal diagnosis, familial cancer syndromes, molecular testing for solid tumors, molecular pathology of the central nervous system, molecular virology, molecular bacteriology, mycology and parasitology, molecular testing for coagulopathies, molecular hemoglobinopathies, molecular diagnostics of lymphoid malignancies, molecular diagnostics of myeloid leukemias, HLA system and transfusion medicine (molecular approach), molecular forensic pathology, gene therapy, ethical and legal issues in molecular testing, and quality assurance and laboratory inspection. Each chapter begins with a detailed Table of Contents for easy reference.

Assembling this diverse guidebook has truly been a team effort, cutting across many traditional specialty boundaries. We are most grateful for all the contributors who made this project possible. Our special thanks go to Mr. Ryan P. Christy from the Multimedia Education Division of the Department of Pathology at Indiana University, who has edited the illustrations for the book. We would like to thank the staff at Humana Press/Springer, including Ms. Mary Jo Casey, Mr. Paul Dolgert, Mr. Richard Hruska, and Mr. David Casey for their assistance in the development and editing of this text, and in particular Ms. Amy Thau, without whose outstanding work this book would have been an impossible achievement.

**Liang Cheng, MD**  
**David Y. Zhang, MD, PhD, MPH**

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# Contributors

## DEBORAH E. BLUE, MD

*Assistant Professor of Pathology  
Associate Director, Molecular Pathology Laboratory  
Department of Pathology and Laboratory Medicine  
and Clarian Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN*

## LIANG CHENG, MD

*Professor of Pathology and Urology  
Director of Molecular Pathology Laboratory  
Chief, Genitourinary Pathology Division  
Department of Pathology and Laboratory Medicine  
and Clarian Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN*

## S. YOON CHOO, MD

*Associate Professor of Pathology and Medicine  
Director of HLA Laboratory, Associate Medical Director  
of Blood Bank  
Departments of Pathology and Medicine  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

## P. MICHAEL CONNEALLY, PhD

*Distinguished Professor Emeritus, Medical and  
Molecular Genetics and Neurology  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

## KENNETH CORNETTA, MD

*Joe C. Christian Professor and Chairman  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

## LISA J. CUSHMAN, PhD

*Certified Genetic Counselor  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

## MAGDALENA CZADER, MD, PhD

*Assistant Professor of Pathology  
Director of Clinical Flow Cytometry Laboratory  
Department of Pathology and Laboratory Medicine  
Indiana University School of Medicine  
Indianapolis, IN*

## PAOLA DAL CIN, PhD

*Associate Professor of Pathology  
Cytogenetics Laboratory  
Department of Pathology  
Brigham and Women's Hospital  
and Harvard Medical School  
Boston, MA*

## DARRELL D. DAVIDSON, MD, PhD

*Assistant Professor of Pathology  
Department of Pathology and Laboratory Medicine  
Indiana University School of Medicine  
Indianapolis, IN*

## STEPHEN R. DLOUHY, PhD

*Associate Research Professor  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

## LISA EDELMANN, PhD

*Assistant Professor  
Department of Genetics and Genomic Sciences  
Director, Molecular Cytogenetics  
Co-Director, Genetic Testing Laboratory  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

## MICHELLE P. ELIEFF, MD

*Staff Pathologist  
Diagnostic Pathology Services, Inc.  
Nampa, ID*

## Contributors

---

**VIRGINIA ESPINA, MS**

*Research Professor  
The Center for Applied Proteomics  
and Molecular Medicine  
George Mason University  
Manassas, VA*

**TAO FENG, MS, MP (ASCP)**

*Research Assistant  
Department of Pathology  
Mount Sinai School of Medicine  
and the Mount Sinai Medical Center  
New York, NY*

**TINA Y. FODRIE, BS, MT, MP (ASCP)**

*Supervisor, Department of Molecular Pathology  
Indiana University School of Medicine  
and VA Medical Center  
Indianapolis, IN*

**TATIANA FOROUD, PhD**

*Professor of Medical and Molecular Genetics  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

**DAVID H. GEHO, MD, PhD**

*Associate Director of Imaging  
Merck and Company, Inc.  
West Point, PA*

**LAWRENCE M. GELBERT, PhD**

*Research Advisor  
Eli Lilly and Company  
Indianapolis, IN*

**BRENT T. HARRIS, MD, PhD**

*Assistant Professor of Pathology  
Department of Pathology  
Dartmouth Medical School  
Lebanon, NH*

**EYAS M. HATTAB, MD**

*Associate Professor of Pathology  
Department of Pathology and Laboratory Medicine  
and Clarian Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN*

**HUGO M. HORLINGS, MD**

*Department of Pathology  
The Netherlands Cancer Institute/Antoni van  
Leeuwenhoek Hospital  
Amsterdam, The Netherlands*

**CAROL L. JOHNS, PhD**

*Supervisor and Technical Coordinator  
Clarian Molecular Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN*

**DAN M. JONES, MD, PhD**

*Professor of Pathology  
Department of Hematopathology  
Medical Director, Molecular Diagnostics Laboratory  
The University of Texas M. D. Anderson Cancer Center  
Houston, TX*

**TIMOTHY D. JONES, MD**

*Staff Pathologist  
Department of Pathology  
Floyd Memorial Hospital and Health Services  
New Albany, IN*

**NATALINE KARDON, MD**

*Associate Professor  
Director of Prenatal Cytogenetics Laboratory  
Department of Genetics and Genomic Sciences  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

**DANIEL L. KOLLER, PhD**

*Research Assistant Professor  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

**RUTH KORNREICH, PhD**

*Research Assistant Professor of Human Genetics  
Co-Director Genetic Testing Laboratory  
Department of Human Genetics  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

**MATTHEW KUCHAR, MD**

*Resident  
Department of Pathology and Laboratory Medicine and  
Clarian Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN*



**NEAL I. LINDEMAN, MD**

*Associate Pathologist, Clinical Chemistry  
Associate Pathologist, Molecular Diagnostics  
Brigham and Women's Hospital  
Assistant Professor of Pathology  
Harvard Medical School  
Boston, MA*

**LANCE A. LIOTTA, MD, PhD**

*Director, The Center for Applied Proteomics  
and Molecular Medicine  
Professor of Life Sciences  
George Mason University  
Manassas, VA*

**ANTONIO LOPEZ-BELTRAN, MD, PhD**

*Professor of Pathology  
Department of Pathology and Surgery  
Cordoba University Medical School  
Cordoba, Spain*

**CATALINA LOPEZ-CORREA, MD, PhD**

*Principal Research Scientist  
Eli Lilly and Company  
Indianapolis, IN*

**HOLGER MOCH, MD**

*Professor and Chairman  
Institute of Surgical Pathology  
Department of Pathology  
University Hospital Zürich  
Zürich, Switzerland*

**RODOLFO MONTIRONI, MD, FRCPath**

*Professor of Pathology  
Institute of Pathological Anatomy and Histopathology  
Polytechnic University of the Marche Region (Ancona)  
School of Medicine  
United Hospitals  
Ancona, Italy*

**VESNA NAJFELD, PhD**

*Professor of Pathology and Medicine  
Director, Tumor Cytogenetics, and Oncology, Molecular  
and Cellular Tumor Markers  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

**THOMAS L. ORTEL, MD, PhD**

*Associate Professor of Medicine and Pathology  
Hemostasis and Thrombosis Center  
Duke University Medical Center  
Durham, NC*

**JODI A. PARKS, MD**

*Visiting Lecturer, Clinical Chemistry  
Department of Pathology and Laboratory Medicine  
and Clarian Pathology Laboratory  
Indiana University School of Medicine  
Indianapolis, IN*

**BRUCE E. PETERSEN, MD**

*Molecular Genetic Pathology Fellow  
Department of Pathology  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

**EMANUEL F. PETRICOIN, PhD**

*Professor of Life Sciences  
Director, The Center for Applied Proteomics  
and Molecular Medicine  
Chair, Department of Molecular and Microbiology  
George Mason University  
Manassas, VA*

**KIMBERLY A. QUAID, PhD**

*Professor of Medical and Molecular Genetics  
Department of Medical and Molecular Genetics  
Indiana University School of Medicine  
Indianapolis, IN*

**VESHANA RAMIAH, MD**

*Hematology/Oncology Fellow  
Duke University Medical Center  
Durham, NC*

**STUART SCHWARTZ, PhD, FACMG**

*Professor of Human Genetics  
Department of Human Genetics  
University of Chicago  
Chicago, IL*

**STUART SCOTT, PhD**

*Clinical Molecular Genetics Fellow  
Department of Human Genetics  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

**MONA SHARAAN, MD**

*Molecular Genetic Pathology Fellow  
Department of Pathology  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

## Contributors

---

### MARTINA STORZ, BS

*Director of TMA Core Facility  
Institute of Surgical Pathology  
Department of Pathology  
University Hospital Zürich  
Zürich, Switzerland*

### MARC VAN DE VIJVER, MD, PhD

*Professor of Pathology  
Department of Pathology  
The Netherlands Cancer Institute/Antoni van  
Leeuwenhoek Hospital  
Amsterdam, The Netherlands*

### FRANCISCO VEGA, MD, PhD

*Assistant Professor of Pathology  
Department of Hematopathology  
The University of Texas M. D. Anderson Cancer Center  
Houston, TX*

### JOSEPHINE WU, DDS, CLSp(MB), CLDir

*Assistant Professor  
Department of Pathology  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

### JULIA D. WULFKUHLE, PhD

*Research Professor  
The Center for Applied Proteomics and Molecular  
Medicine  
George Mason University  
Manassas, VA*

### RULIANG XU, MD, PhD

*Associate Professor of Pathology  
Department of Pathology  
New York University School of Medicine  
New York, NY*

### FEI YE, PhD

*Assistant Professor  
Department of Pathology  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

### C. CAMERON YIN, MD, PhD

*Assistant Professor of Pathology  
Department of Hematopathology  
The University of Texas M. D. Anderson Cancer Center  
Houston, TX*

### DAVID Y. ZHANG, MD, PhD, MPH

*Associate Professor of Pathology  
Director, Molecular Pathology Laboratory  
Department of Pathology  
Mount Sinai School of Medicine and the Mount Sinai  
Medical Center  
New York, NY*

### SHAOBO ZHANG, MD

*Associate Research Professor  
Department of Pathology and Laboratory Medicine  
Indiana University School of Medicine  
Indianapolis, IN*