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Maria M. Picken
Guillermo A. Herrera
Ahmet Dogan *Editors*

Amyloid and Related Disorders

Surgical Pathology and Clinical Correlations

Second Edition

Humana Press

CURRENT CLINICAL PATHOLOGY

ANTONIO GIORDANO, MD, PHD
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SERIES EDITOR

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This book is dedicated to our patients, past and present, with the hope that it will make a difference in the lives of future amyloidosis patients.

MMP, GAH, AD

Preface (Second Edition)

Amyloidosis, although known since the nineteenth century, retained for a long time the aura of a rare and obscure disease that was untreatable and mainly of purely academic interest. This state of affairs has, however, changed dramatically in recent years. With the new therapies that are now available, patients with systemic light chain amyloidosis (AL) may achieve a durable response and live for more than a decade from the time of their first diagnosis. Treatments for other types of systemic amyloidosis are also improving. Thus, in addition to liver transplantation, patients diagnosed with hereditary amyloidosis derived from a mutant transthyretin (ATTR) are currently being offered pharmacologic therapies that are in clinical trials. However, treatment outcomes are most successful when they are applied early in the disease process. Thus, now more than ever, early diagnosis is of the utmost importance. Although there are a number of excellent amyloidosis treatment centers around the world, early diagnosis of affected patients is reliant upon widespread and effective screening, and, despite advances in laboratory medicine, this still hinges upon the detection of deposits in tissues. Thus, the role of the pathologist in this process is critical. This book therefore has, as its primary focus, the diagnosis of amyloidosis in surgical pathology. Although written primarily for pathologists, it is hoped that this volume will also be helpful to those who would wish to gain insight into recent diagnostic and treatment options.

This second edition of "Amyloid and related disorders" has been expanded to include seven new chapters, while the prior content has been updated. The volume begins with a history of amyloid investigations and the latest nomenclature. Separate chapters are devoted to the mechanism of amyloidogenesis and an overview of AL, AA, ALECT2, hereditary, dialysis, and localized amyloidoses; a brief overview of cerebral amyloidoses is also included. In Part II, diseases that mimic amyloid and related disorders are discussed. Part III is entirely devoted to pathologic diagnosis, including the generic diagnosis of amyloid, and issues pertaining to amyloid typing that involve both antibody-based and proteomic methods. Part IV provides an overview of laboratory support for the diagnosis of amyloidosis, including serum, urine, bone marrow, and genetic studies. Part V provides an overview of amyloid pathologies in the genitourinary tract, cardiac, gastrointestinal/liver, and peripheral nervous systems; new chapters on lymph nodes and spleen, pulmonary, dermal, breast, and iatrogenic amyloidoses have been added.

Part VI discusses clinicopathologic issues and the role of solid organ transplantation, as well as recent advances in therapies for AL, hereditary, and AA amyloidosis. Brief chapters on relevant legal issues, and the patient's perspective, conclude Part VI.

Those who are interested in the amyloidoses are also encouraged to review the contents of "Amyloid: The Journal of Protein Folding Disorders" and contact the International Society of Amyloidosis (www.amyloidosis.nl). Resources available to patients include the Amyloidosis Foundation (http://www.amyloidosis.org) and the Amyloidosis Support Group (http://www.amyloidosissupport.org).

It also behooves us to acknowledge that abnormal protein folding, the very essence of amyloid fibril formation, affects many more aspects of our lives than those covered by the chapters in this book. While amyloid formation represents a fundamental process in many diseases and aging, it also plays an important role in vertebrate and invertebrate biology, as functional amyloid; amyloid fibrils also have applications in the fields of nanotechnology and bioengineering. Therefore, understanding the driving forces behind both the regulated and unregulated formation of amyloid structures may help us to enlist that knowledge in the fight against disease and the aging process and may, unexpectedly, also lead to improvements in many other areas of our lives.

Maywood, IL, USA — Maria Mrozowicz Picken
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