

# Preface

Alpha-1 antitrypsin deficiency is an inherited condition that leads to lung disease in adults and liver disease in children and adults. The condition has a prevalence that varies from country to country, ranging from one in 2750 to one in 4500 live births, and currently is the only known genetic mutation firmly associated with chronic obstructive lung disease in adults. While the mechanisms underlying the clinical manifestations of alpha-1 antitrypsin deficiency have been largely clarified, specific treatment currently is only available for the lung disease. A growing interest of academic investigators and industry in finding new therapeutic solutions for lung and liver disease likely will lead to better clinical outcomes in the foreseeable future. This will necessitate a better effort to detect the condition, which is broadly underdiagnosed at present. The purpose of this book is to summarize what is known about the biology of alpha-1 antitrypsin and its deficiency, the clinical manifestations of alpha-1 antitrypsin deficiency, and the currently available therapeutic options.

The book begins with a chapter on the biologic role of serine protease inhibitors (SERPINS) including alpha-1 antitrypsin in general, followed by a chapter focusing on alpha-1 antitrypsin in particular. The next two chapters address the process of alpha-1 antitrypsin protein misfolding and polymerization, and their pathogenetic consequences in the liver and the lung, the principal sites of alpha-1 antitrypsin deficiency-related disease.

The condition is then characterized from a clinical perspective in the following three chapters that review the methods of and challenges to the detection of alpha-1 antitrypsin deficiency, and the manifestations and management of lung disease in adults, and liver disease in children and adults. With these three chapters we intend to provide clinicians with useful information on the diagnosis and treatment of alpha-1 antitrypsin deficiency.

The last two chapters cover topics that reach beyond alpha-1 antitrypsin deficiency per se. One chapter reviews what is currently known about the potential for using alpha-1 antitrypsin therapeutically in diseases not associated with alpha-1 antitrypsin deficiency. The final chapter addresses the important role of voluntary health organizations in the rare disease space, with a focus on alpha-1 antitrypsin deficiency. Voluntary health organizations raise awareness of the condition, support

research, promote new drug development, and bring the patient perspective to the table.

This book was conceived to provide the readership of health professionals and scientists with a comprehensive overview of alpha-1 antitrypsin deficiency. International authorities, who are widely recognized for their contributions to the basic and clinical science of alpha-1 antitrypsin deficiency, wrote the chapters. Certain difference of opinion may exist between authors. We have taken the position to allow for such differences and potential controversies and give the readers the opportunity to form their own conclusions.

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