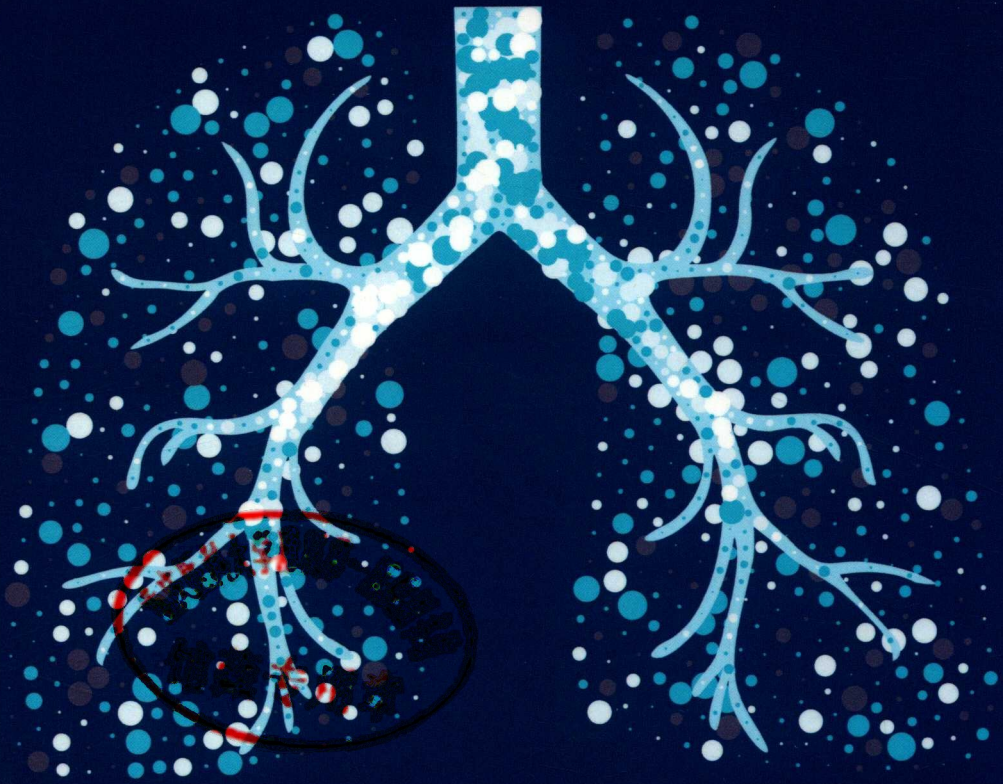

# RARE AND INTERESTING CASES IN PULMONARY MEDICINE

ALI ATAYA
ELOISE HARMAN

# Rare and Interesting Cases in Pulmonary Medicine

**Ali Ataya, MD**
Assistant Clinical Professor of Medicine, Division of Pulmonary Critical Care, and Sleep Medicine, University of Florida
Gainesville, FL, United States

**Eloise Harman, MD**
Professor Emeritus of Pulmonary and Critical Care Medicine
University of Florida, College of Medicine, and Staff Physician and MCU Director, Malcolm Randall VA Medical Center
Gainesville, FL, United States



**ACADEMIC PRESS**
**An imprint of Elsevier**
**elsevier.com**

Academic Press is an imprint of Elsevier
125 London Wall, London EC2Y 5AS, United Kingdom
525 B Street, Suite 1800, San Diego, CA 92101-4495, United States
50 Hampshire Street, 5th Floor, Cambridge, MA 02139, United States
The Boulevard, Langford Lane, Kidlington, Oxford OX5 1GB, United Kingdom



**Notices**
Knowledge and best practice in this field are constantly changing. As new research and experience broaden our understanding, changes in research methods, professional practices, or medical treatment may become necessary.

Practitioners and researchers must always rely on their own experience and knowledge in evaluating and using any information, methods, compounds, or experiments described herein. In using such information or methods they should be mindful of their own safety and the safety of others, including parties for whom they have a professional responsibility.

To the fullest extent of the law, neither the Publisher nor the authors, contributors, or editors, assume any liability for any injury and/or damage to persons or property as a matter of products liability, negligence or otherwise, or from any use or operation of any methods, products, instructions, or ideas contained in the material herein.

**Library of Congress Cataloging-in-Publication Data**
A catalog record for this book is available from the Library of Congress

**British Library Cataloguing-in-Publication Data**
A catalogue record for this book is available from the British Library

ISBN: 978-0-12-809590-4



*Publisher:* Mica Haley
*Acquisition Editor:* Stacy Masucci
*Editorial Project Manager:* Samuel Young
*Production Project Manager:* Edward Taylor
*Designer:* Mark Rogers

Typeset by TNQ Books and Journals

# Rare and Interesting Cases in Pulmonary Medicine

# Dedication

To my parents
To my wife
And to all my mentors and patients over the years

– Ali Ataya

To my family
My mentors Dr. Jay Block and Dr. William Bell
And to the patients who have taught me so much

– Eloise Harman

# ORS Contributors

**Humberto E. Trejo Bittar, MD**
Assistant Professor, Department of Pathology, University of Pittsburgh Medical Center, Pittsburgh, Pennsylvania

**Mitra Mehrad, MD**
Assistant Professor, Department of Pathology, Microbiology and Immunology, Vanderbilt University Medical Center, Nashville, Tennessee

**Tan-Lucien Mohammed, MD**
Associate Professor, Department of Radiology, University of Florida, Gainesville, Florida

# Preface

Patients with rare lung disorders are often misdiagnosed or diagnosed late in the course of their disease. This is frequently due to physicians not considering the diagnosis in the first place. However, interest in rare and orphan lung diseases has been increasing among healthcare providers, pharmaceutical companies, and patients.

The goal of this book is to provide an introduction to various rare lung disorders, with the hope that this may assist in the recognition, diagnosis, and treatment of these diseases as they are encountered in clinical practice. This book may also benefit physicians studying for their pulmonary board exams. Each case begins with a case study of a patient with one of these rare diseases followed by a rapid review of the disease or syndrome. This case-centered approach is expected to help the reader to recall the information if they encounter patients with one of these rare diseases.

Ultimately, we hope this book will contribute to improving diagnosis and care of patients with rare lung disorders.

**Ali Ataya, MD**
**Eloise Harman, MD**

# Contents

## Case 22



## Case 23



## Case 24



## Case 25



## Case 26



## Case 27



## Case 28



## Case 29

## Case 30



## Case 31



## Case 32



## Case 33



## Case 34



## Case 35



## Case 36



## Case 37

## Case 38



## Case 39



## Case 40



## Case 41



## Case 42



## Case 43



## Case 44



## Case 45

## Case 46



## Case 47



## Case 48



## Case 49



## Case 50



## Case 51



## Case 52



## Case 53

## Case 54



## Case 55



## Case 56



## Case 57



## Case 58



## Case 59



## Case 60

# Case 1

A 60-year-old Caucasian female presents with progressive shortness of breath with exertion and a nonproductive cough for the last year and a half. She is a lifelong nonsmoker, has no significant past medical problems, and is not on any medications.

Examination of the heart and lungs is normal and there is no digital clubbing. A chest computed tomography scan revealed multiple small peripheral nodular opacities in the right upper and lower lobes as well as hilar and mediastinal adenopathy (Fig. 1.1). Endobronchial ultrasound with transbronchial needle aspiration of the mediastinal lymph nodes was performed. Histology is shown in Fig. 1.2. Further workup showed no other organ involvement of the disease.

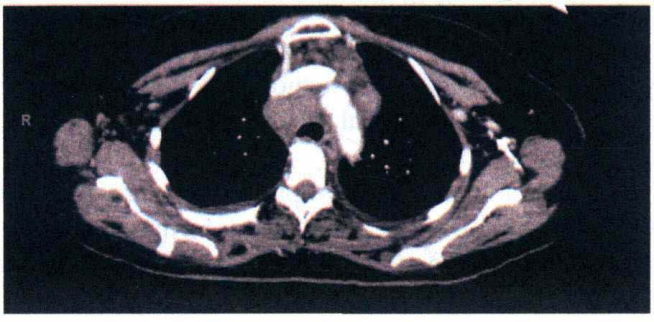


**FIGURE 1.1** Chest computed tomography scan with contrast showing enlarged mediastinal 4R node.

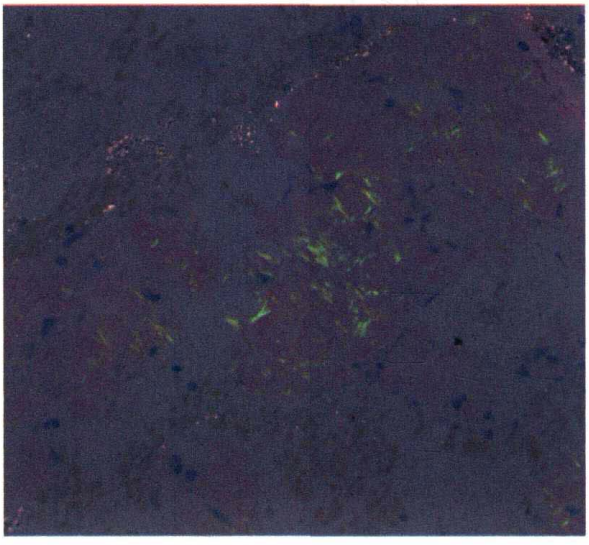

**FIGURE 1.2** Histology showing clumps of amorphous material with Congo red stain under polarized light.

**What is the diagnosis?**

## PULMONARY AMYLOIDOSIS

Amyloidosis is a systemic disease characterized by extracellular deposition of amyloid, which constitute insoluble β-pleated protein sheets, in different organs. Amyloidosis can be primary/idiopathic (AL type), or secondary/reactive (AA type). The secondary form may occur in the setting of an underlying malignancy, chronic inflammatory, or infectious disease, appear in the setting of chronic renal disease, or be heritable. Isolated pulmonary amyloidosis usually occurs in the setting of the idiopathic form of the disease. Isolated pulmonary amyloidosis is characterized by the occurrence of amyloidosis in the lungs without any systemic involvement.

Patients have nonspecific symptoms due to the diversity of its pulmonary manifestations and tissue biopsy is necessary to make the diagnosis. Isolated pulmonary amyloidosis comes in multiple forms:

1. Tracheobronchial amyloidosis: Most common form. Patients may present with cough, dyspnea, wheezing, or hemoptysis. Patients may have thickened trachea with stenosis. If proximal lesions are present, they may result in fixed upper airway obstruction.
2. Nodular form: Patients may be asymptomatic or present with a cough. A single nodule or multiple small nodular lesions may appear peripherally in the lower lobes. Amyloid nodules may be calcified and cavitate in 10% of cases.
3. Amyloid adenopathy: Amyloid is deposited in the hilar and mediastinal lymph nodes, usually bilaterally. This form of the disease rarely occurs alone or without systemic involvement.
4. Diffuse interstitial form: This is the rarest form of the disease. Amyloid gets deposited in the pulmonary interstitium between the alveoli and blood vessels, impairing gas transfer. Imaging will show a reticular or reticulonodular pattern that may present asymmetrically. Patients succumb to respiratory failure.

Tissue biopsy is the gold standard for diagnosis. Histology will show pink amorphous material that under polarized light will stain apple-green birefringence with Congo red stain.

There is no effective treatment for the disease. Patients with tracheobronchial involvement may undergo bronchoscopic treatment with Nd–YAG laser or clipping for obstructing lesions. For other forms external beam radiation and systemic immunosuppression have been used to halt progression.

This patient underwent further workup that showed no systemic involvement, including a bone marrow biopsy. She was diagnosed with nodular amyloid with hilar and mediastinal lymph node involvement and referred for systemic chemotherapy treatment.