Introduction

Thomas O.F. Wagner[1,2], Marc Humbert [2,3,4,5], Marlies Wijsenbeek[2,6], Michael Kreuter [2,7] and Helge Hebestreit [2,8]

[1]Department of Pneumology, University Hospital Frankfurt, Frankfurt, Germany. [2]European Reference Network for Rare Diseases of the Respiratory System (ERN-LUNG), Frankfurt, Germany. [3]Assistance Publique – Hôpitaux de Paris (AP-HP), Dept of Respiratory and Intensive Care Medicine, Pulmonary Hypertension National Referral Center, Hôpital Bicêtre, Le Kremlin-Bicêtre, France. [4]Université Paris-Saclay, Faculty of Medicine, Le Kremlin-Bicêtre, France. [5]INSERM UMR_S 999 "Pulmonary Hypertension: Pathophysiology and Novel Therapies", Le Kremlin-Bicêtre, France. [6]Center of Excellence for Interstitial Lung Diseases and Sarcoidosis, Department of Respiratory Medicine, Erasmus Medical Center-University Medical Center Rotterdam, Rotterdam, The Netherlands. [7]Mainz Center for Pulmonary Medicine, Department of Pneumology, Mainz University Medical Center and Department of Pulmonary, Critical Care & Sleep Medicine, Marienhaus Clinic Mainz, Mainz, Germany. [8]Pediatric Pulmonology & Cystic Fibrosis, Children's Hospital, University Hospital Würzburg, Würzburg, Germany.

Corresponding author: T.O.F. Wagner (t.wagner@em.uni-frankfurt.de)

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Rare respiratory diseases pose a significant burden and can be challenging to diagnose and treat. This *Monograph* provides an up-to-date, comprehensive resource to the clinician, both for educational purposes and for clinical care. https://bit.ly/ERSM100intro

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Respiratory system disorders play a crucial role in the burden of disease and account for a huge portion of morbidity and mortality. People with rare diseases – according to the European definition, affecting not more than five in 10 000 – share symptoms and functional impairments with many more common diseases. Diagnosing these diseases can be challenging due to their rarity, and treating these conditions is complex because of often quite specific needs and treatment options.

To address this, the European Respiratory Society (ERS) has published Rare Diseases of the Respiratory System – the 100th issue of the *ERS Monograph*. The previous *Monograph* in this thematic area, entitled Orphan Lung Diseases and edited by Jean-Francois Cordier [1], was published in 2011 and needed an update.

To reflect the close collaboration of ERS with the European Reference Network for Rare Diseases of the respiratory system (ERN-LUNG; https://ern-lung.eu/), Thomas O.F. Wagner, was honoured to guest edit this new *Monograph* within a team of esteemed co-guest editors, since dealing with rare diseases nowadays requires networking and teamworking. This collaboration of networks has allowed for most of the recent progress made. A good example of the impact of networking on the improvement of care and research for people with rare lung diseases are the clinical trials networks. These collaborative infrastructures have fostered the development and market authorisation of many new drugs for rare diseases.

Another evolutional change of this book compared with its predecessor is that the reviews it contains now offer a more comprehensive overview of the whole spectrum of rare diseases of

the respiratory system, providing readers with essential information about these diseases, their diagnosis and their treatment.

The book is structured into thematic sections, with significant overlap between some of the sections. The first two chapters provide an overview of how to identify rare diseases of the respiratory system and their differential diagnosis [2, 3]. The sections that follow cover rare diseases of the lung interstitium [4–7], rare diseases of the airways or alveoli [8–13], and rare pulmonary vascular diseases [14–17]. The authors of each chapter are experts in their respective fields, and they provide valuable insight into the diagnosis and treatment of rare respiratory diseases. For instance, chapter 3 discusses ILDs and covers differential diagnosis, definitions, clinical phenotype, radiological classifications and histological characterisation [4]. Section 4 covers rare diseases of the airways or alveoli, such as bronchiolitis [8] and alveolar proteinosis [9], as well as primary ciliary dyskinesia [10], cystic fibrosis [11], bronchiectasis [12] and α_1-antitrypsin deficiency [13]. Section 5 provides a comprehensive overview of pulmonary vascular diseases, including pulmonary arterial hypertension [14], chronic thromboembolic PH [15], and PH complicating the course of other rare lung diseases [16]. The authors in this section provide clinical guidance, diagnostic and therapeutic approaches, and refer the reader to the right sources for detailed up-to-date information.

Overall, this book should constitute an excellent resource for healthcare professionals, researchers and students interested in rare diseases of the respiratory system. The authors provide comprehensive coverage of the whole spectrum of rare respiratory diseases, highlighting both the progress made in recent years and the areas where more work is needed. They also promote the idea of exchange, encouraging healthcare professionals to work together and share knowledge to improve diagnostic and therapeutic options for patients with rare respiratory diseases. Authors and editors belong to and/or support the vision and mission of ERN-LUNG. This network, funded by the European Commission in 2017, is the European information and collaboration hub offering expert support to patients and professionals and will be able to connect readers who want more detailed or specific information on any topic within the field of rare diseases of the respiratory system with the respective experts.

We are confident that Rare Diseases of the Respiratory System will prove a valuable resource for anyone interested in respiratory medicine. It provides a comprehensive overview of rare respiratory diseases, their diagnosis, and treatment, and promotes the exchange of knowledge among healthcare professionals.

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