Respiratory Medicine (2010) **104**, S1

available at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/rmed

EDITORIAL

Rare pulmonary diseases: A path to the future

In their professional lives, physicians involved in respiratory medicine may occasionally follow pulmonary disorders matching the definition of a rare disease. However, because of the rarity of these disorders the physician's experience in the field is usually limited to a few cases. Presented in this supplement of *Respiratory Medicine* are the proceedings of the Third International Congress on Rare Pulmonary Diseases and Orphan Drugs in Respiratory Medicine, held in Milan on 20–21 March 2009. This congress follows two similar meetings, both held in Milan, the first in February 2005[1] and the second in February 2007.

Like the 2005 and 2007 meetings the conference reported here focused on pathogenetic, clinical and therapeutic advances in the field of rare lung diseases. Topics presented over the 2-day conference included basic scientific data, genetic projects, molecular advances and other findings that have contributed to increase the therapeutic possibilities in patients suffering from these diseases. Valuable contacts have been generated between pulmonologists, internists and physicians involved in imaging, pathology and molecular biology, who have collegially discussed developments that have emerged in the last 2 years.

The conference reports that have been selected to be published in this issue begin with the contribution of Sitbon, who reviews current strategies in the management of pulmonary arterial hypertension. Humbert discusses an uncommon form of pulmonary arterial hypertension, pulmonary veno-occlusive disease (PVOD), and provides an outstanding overview of the clinical and therapeutic implications of this rare disease.

Three reports on the topic of idiopathic pulmonary fibrosis were selected by the editors of this issue. In the first, Cavazza reviews recent data that have strengthened the role of the pathologist in this disease, while Caminati gives an update on the clinical management of these patients. In the third report, Pesci provides data on his valuable experience in the controversial role of bronchoalveolar lavage in idiopathic pulmonary fibrosis.

The problem of clinical trials and registries in rare diseases is considered in the report by Luisetti, who presents and discusses the international experience with alpha1-antitrypsin deficiency achieved by the Alpha One International Registry, and national experience obtained with a large series of patients with pulmonary alveolar proteinosis. The role of bronchoalveolar lavage and transbronchial biopsy in the diagnosis of cystic lung diseases is reviewed by Torre in an interesting overview on this topic that suggests a number of considerations to the reader.

Various clinical aspects of interstitial lung disease in systemic sclerosis, and different therapeutic options for the severe restrictive lung disease that complicates this disease, are extensively reviewed by Mouthon in an outstanding report. Agostini discusses the controversial role of stem cell transplantation in the clinical management of rare lung diseases; regenerative medicine is expected to be a major challenge for the future in respiratory medicine and various sources of stem cells may be used in different disorders.

In his report Johnson describes the development of guidelines for lymphangioleiomyomatosis by the European Respiratory Society and discusses how these guidelines can help the standardization of diagnostic criteria in this disease, thus improving clinical care and facilitating research and clinical trials. Moss, taking advantage of his enormous experience in the field of lymphangioleiomyomatosis, addresses encouraging new developments in the treatment of this rare disease.

We hope that the reader taking a look at the reports included in this issue will receive an impression of the scope of the discussions that were generated by the data presented during the various sessions of the congress. Because of the rapidly evolving field, we plan a fourth international update within the next 2 years.

Conflict of interest statement

The authors have no conflict of interest to declare.

Reference

1. Harari S, Agostini C, editors. *The first international conference on rare pulmonary diseases and orphan drugs in respiratory medicine. Sarcoidosis Vasc Diffuse Lung Dis* 2005;**22**(Suppl. 1):S1–S107.

Sergio Harari*
Unità Operativa di Pneumologia e Terapia Semi-Intensiva Respiratoria e Servizio di Fisiopatologia Respiratoria ed Emodinamica Polmonare, Ospedale San Giuseppe Fatebenefratelli, Via San Vittore 12, 20123 Milan, Italy
*Corresponding author. Tel.: +39 (0) 2 8599 4580; fax: +39 (0) 2 8599 4400.
E-mail address: sharari@ilpolmone.it

Carlo Agostini
Padua University School of Medicine, Departments of Clinical and Experimental Medicine, Clinical Immunology and Hematology, 35128 Padua, Italy

Available online 28 April 2010

doi:10.1016/j.rmed.2010.03.011