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Rare pulmonary diseases and orphan drugs: where do we stand and where are we going to?

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Updates on rare pulmonary diseases from the 6th International Meeting on Pulmonary Rare Diseases and Orphan Drugs http://ow.ly/PYUCC

Rare diseases are a major problem for the patients who suffer from them, for their families and for selected specialists involved in their management. In addition, they cause economic, social and public health problems that should be acknowledged and discussed by the medical and scientific community. Because of the rarity of these illnesses, the experience of most physicians is usually limited to a few cases. Nevertheless, the essential steps in the diagnosis and management of a patient who is suspected to have, for example, an interstitial lung disease or pulmonary hypertension, should be common knowledge, as these diseases are uncommon, but not, in fact, exceedingly rare [1–3]. Furthermore, rare presentations of frequent lung diseases (*e.g.* rare thoracic cancers, severe asthma, and emphysema with α_1-antitrypsin deficiency) and the most typical rare pulmonary diseases can share similar aspects, both in research and in their clinical management [4].

Rare diseases also constitute an interesting challenge for the development of new trials and for the evaluation of surrogate end-points, as demonstrated in recent years with forced vital capacity in idiopathic pulmonary fibrosis trials [5–7] and with the criticism of the 6-min walk test in pulmonary hypertension studies [8].

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The possible gap between the efficacy of drugs tested in clinical trials and their effectiveness in real life represents another aspect of particular interest in the evaluation of orphan drugs, as indeed it is in the evaluation of drugs used in more common conditions. The strong links between basic research and clinical management can be witnessed in the way many centres around the world already manage rare pulmonary diseases. It represents an advanced model of translational approach to modern medicine.

In order to discuss these topics, the International Meeting on Pulmonary Rare Diseases and Orphan Drugs has been held in Milan (Italy) every other year since 2005. It is the only European meeting dedicated to all the various types of rare pulmonary diseases, both parenchymal and vascular. The 6th Congress was endorsed by the European Respiratory Society (ERS), and was held on February 27 and 28, 2015 (see [9] for more information and slide presentations), for the Rare Disease Day (February 28, 2015) and also in light of the fact that the Universal Exposition 2015 is taking place in Milan this year. The next Congress will be held on February 24 and 25, 2017. The success of this Congress is based on its many profiles (international, translational and clinical), which bring together scientists from different fields of expertise (from pulmonary hypertension to lymphangioleiomyomatosis, idiopathic pulmonary fibrosis, *etc.*), providing them with a valuable opportunity to encompass a broad range of insights within the area of respiratory medicine.

The scientific programmes of the different editions of the meeting were designed to build bridges between physicians specialised in the management of different respiratory diseases, which had the common feature of being rare or corresponding to rare presentations of common lung diseases. The translational approach has been improved in recent years, bridging basic research and clinical investigation, and the meeting has become more and more international. The way to provide new insights into science and clinical care is the sharing of knowledge and the exchange of information with regard to different areas of clinical and basic research in respiratory medicine, among experts with varied specialisms in pulmonary medicine.

An international scientific committee, comprising eminent physicians from Europe and the USA, suggested the topics for the programme. In 2015, the committee included F. Blasi, H.R. Collard, V. Cottin, M. Humbert, M. Matucci-Cerinic, G. Simonneau, C. Vancheri, A.U. Wells and S. Harari (Congress Chair).

In the past 20 years, there have been significant advances in the knowledge and management of rare pulmonary diseases. This has been made possible by several factors, including: 1) the better definition of some of the diseases (*e.g.* idiopathic pulmonary fibrosis and pulmonary arterial hypertension); 2) the collaborative effort of research carried out by the respiratory medicine community; 3) the process of information of the public, physicians and patients; 4) the coordinated action against rare diseases implemented by health authorities in several western countries and at a European level; 5) the significant role of patients' associations, which have raised awareness and created funds, allowing independent clinical trials; and, last but not least, 6) the investments made by major pharmaceutical companies, fostered by orphan drug legislation.

The increasing scientific interest in rare pulmonary diseases within the entire European medical community can be witnessed in the continuous rise in the number of scientific publications related to these topics and in the growing attention paid to this field in the publications of the ERS [1, 10]. In particular, in the past few years, the *European Respiratory Review* (*ERR*) has dedicated a lot of space to rare pulmonary diseases and orphan drugs.

In this issue of the *ERR*, selected papers from the Milan meeting are published and give an overview of important areas of pulmonary research. Cappelli *et al.* [11] provide a comprehensive review of the new advances in the pathogenesis, diagnosis and treatment of interstitial lung disease associated with systemic sclerosis. Adir *et al.* [12] discuss the epidemiology, pathogenic mechanisms and treatment approaches of the different causes of pulmonary hypertension in the setting of chronic myeloproliferative disorders: chronic thromboembolic pulmonary hypertension, pre-capillary pulmonary hypertension mimicking pulmonary arterial hypertension and drug-induced pulmonary hypertension. Caminati *et al.* [13] add important information to our knowledge regarding pitfalls in methodologies and data interpretation in epidemiological studies on idiopathic pulmonary fibrosis and analyse the main issues that should be carefully considered when comparing different studies. Margaritopoulos *et al.* [14] present an overview of what's new in smoking-related lung diseases. Harari and Caminati [15] discuss the role of real-life studies on idiopathic pulmonary fibrosis and the gap between randomised controlled clinical trials and everyday practice. We hope you will enjoy reading these articles and we look forward welcoming you in Milan at the next International Meeting on Pulmonary Rare Diseases and Orphan Drugs in February 2017.

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