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Rare lung diseases — should we pay more attention?

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Rare diseases are defined as the diseases that affect less than one person in 2,000 people. There are about 6000 such disorders, including diseases, anomalies and syndromes. About 80% of them have genetic origin. However, the number of patients with a single disease is small, particularly in one country, but if we take into account the entire world and the number of disorders and patients, there are millions cases worldwide.

Many of rare diseases are orphan diseases, which are defined as those of limited interest to scientists, doctors, healthcare and pharmaceutical companies. Some of orphan diseases are very rare but others are common as so-called neglected infectious diseases, which are endemic to areas ravaged by poverty in Africa, Asia and the Americas (tropical infections and infestations: lymphatic filariasis, leishmaniasis, African trypanosomiasis, schistosomiasis, trachoma, onchocerciasis, Chagas disease, etc.).

The epidemiological data on that field are spare and with high probability of not being registered. Frequently, the clinical and radiological picture of these diseases vary widely, and in spite of patients with spectacular symptoms, there are some cases with not pronounced symptoms or even occult. It often influences underdiagnoses and delayed correct diagnosis.

In addition, only for few diseases diagnostic and therapeutic recommendations have been established and many statements still require proving in prospective randomized studies.

Rare lung diseases may involve only the lung, or the lung can be one of multiple involved organs. Usually patients with rare disease require multidisciplinary care in highly specialized centers, however, with tight cooperation with family doctors.

It is underlined that special systems of education focused on rare diseases should be employed for medical students and young doctors thus improving awareness of these diseases. All respiratory physicians should have some clinical and scientific knowledge of the field and should be open-minded about diagnosis.

Usually rare diseases are present throughout the person's entire life and apart from medical problems, there are many social and psychosocial problems that should be resolved. The role of patients' organizations in the field of stimulation of health care systems for the improvement of health care, social welfare, scientific research, implementations of new drugs is outstanding.

Recently The European Union Committee of Experts on Rare Diseases (EUCERD) has established recommendations for the criteria of centers of expertise for rare diseases in member states, their mission and scope, and the criteria of their designation. Emphasis has been placed upon the development of European reference networks, as well as registries and databases, and the necessity of a multidisciplinary approach.

The Orphanet website (www.orpha.net) is the principle source of important and validated information about the rare disorders, for both

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Table 1. Rare pulmonary diseases

Autoimmune diseases
Anti-basement membrane syndrome
Pulmonary alveolar proteinosis
Amyloidosis
Common variable immunodeficiency disease
Vasculitides
Granulomatosis with polyangitis (Wegener's disease)
Microscopic polyangitis
Eosinophilic granulomatosis with polyangitis
(Churg-Strauss syndrome)
Behçet's disease
Takayasu's arteritis
Disorders of genetic origin
Lymphangioleiomyomatosis sporadic and associated
with tuberous sclerosis
Birt-Hogg-Dubé syndrome
Primary ciliary dyskinesia
Alpha-1-antytripsin deficiency
Elhers-Danlos syndrome
Neurofibromatosis
Gaucher disease
Other idiopathic disorders (limited to the lungs)
Idiopathic pulmonary fibrosis
Idiopathic eosinophilic pneumonias
Tracheobronchopathia osteochondroplastica
Tracheobronchomegaly (Mounier-Kuhn syndrome)
Idiopathic bronchiolitis
Other rare diseases
Thoracic endometriosis
Langerhans' cell histiocytosis

patients and doctors. The Polish version of this page is full of many recent and important information. Also EuroHistionet (www.eurohistio.net/index_eng) and Histiocytosis Society (www.histiocytesociety.org),

American LAM Foundation (www.thelamfoundation.org) and national LAM web pages provide useful information regarding histiocytic disorders and LAM.

Improved clinical and basic scientific knowledge, as well as research on rare pulmonary diseases should be one of the most important goals for all respiratory physicians at the present time and in the future.

Conflict of interest

The author declares no conflict of interest.

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