Introduction

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Imagine going to the doctor and being told that you have a rare lung disease—that you are one in a million. Any feeling of being special is rapidly replaced by one of frustration. Your general practitioner has never heard of the condition, your specialist has seen one case previously, and when you decide to seek out the medical literature you find that descriptions are largely based on reviews of five patients from the 1940s. When you have symptoms not described in the textbook no one can confirm whether they are “normal” or not. Worst of all, perhaps, is the isolation—you cannot commiserate or discuss your problems with other patients. As one patient said after being told that she had lymphangioleiomyomatosis: “I felt as if I had been put on an island all on my own”. Another frustration is the lack, or apparent lack, of research into the disease because of its rarity, thus removing the hope that often sustains patients with a severe disease that an effective treatment may appear.

Rare diseases are by definition uncommon, but as there are many of them the number of patients affected by a rare disease is appreciable. In this issue of Thorax we are therefore starting a series of articles on rare diseases of the lung, the first of which by Dr Simon Johnson deals with lymphangioleiomyomatosis. The articles have been written with two main objectives. The first is to provide a clear and comprehensive review of the clinical aspects of the disease covering natural history, diagnosis, and management. This is designed to help clinicians with limited experience of the disease to provide the best possible care for these patients. The second objective is to review current thinking about underlying mechanisms and the direction of research. This will, we hope, be of interest to clinicians with affected patients. It should also be of broader relevance and will encourage interest and research into these disorders.

Determining the aetiological factors and underlying mechanisms of a rare disease is inherently satisfying but it often has further ramifications, leading in several instances to an improved understanding of basic pathophysiological processes of relevance to other diseases. Research into sporadic and familial retinoblastoma by Knudson1 led to the “two hit” model of carcinogenesis which helped to explain relatively rare familial neoplasias. With time, however, his model has been seen as being of much wider relevance, helping to explain the role of somatic mutations in the aetiology of cancer more generally.2

Carrying out research into rare diseases is difficult and for many diseases had, until recently, progressed little beyond collecting demographic data such as age of onset and median survival. Advances in molecular biology, information technology, epidemiological methodology, including molecular epidemiology, are, with international collaboration, helping to push the research into areas where it is starting to unravel underlying mechanisms. Anecdotal reports of response to treatment are being replaced by more robust data, thereby improving the quality of the information and advice available to patients. Progress is variable and scientists looking for a challenge, whether clinical or laboratory based, will find several potential areas in the articles in this series.

Deciding what to include in this series has been somewhat arbitrary, but we have focused on diseases that most respiratory physicians will see relatively rarely and where help with management may be appreciated. We have concentrated on disorders that are usually persistent or progressive rather than rare infections and tumours. We have asked authors to review the literature so that the articles should also provide a useful reference source for both physicians and research workers.

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