Primary pulmonary hypertension: a GP comment

General practitioners (GPs) are subject to bombardments of medical information from many sources – local pharmaceutical formularies, local and national guidelines, national service frameworks, medical newspapers, peer-reviewed national journals and special interest publications.

Consequently, many of us struggle to keep abreast of developments, even in our own fields of interest. The GP confronted with a chronic rare disease may have difficulty in accessing relevant, clear information about it.

Primary pulmonary hypertension (PPH), with an incidence of less than two cases per million, is one such disease which most GPs and many physicians will not encounter in their working lives. Any knowledge possessed may date back to a poorly remembered paragraph in a medical student textbook.

To the patient and his or her family, the disease presentation may be tragically dramatic with the sudden death of one or more siblings in the family.

Ross Price

My first experience of the condition occurred when a young mother came to see me stating that her brother and sister had died young from PPH. Her sister had died suddenly in childbirth at the age of 21 from previously unsuspected pul-

monary hypertension. Her brother had collapsed at the age of 23 during a military exercise. He was investigated and also found to have pulmonary hypertension. Unfortunately, within a few days of the diagnosis being made, he died. Her questions were: was she at risk and were her son and daughter likely to have the disease? What was the prognosis and what, if any, therapy was available? Unsurprisingly, I did not know but referred her and her family to our local cardiologist and clinical geneticist who were able to define the risks and to monitor the family by serial echocardiography.

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The mother and daughter have remained free from the disease, but the condition became apparent in the son, with the onset of recurrent chest infections and progressive dyspnoea.

My role in counselling the family and patient was hindered by my lack of knowledge of the condition and the absence of a concise comprehensive source of information.

The two articles by Professor Yacoub's team[1,2] will do much to combat such a deficiency of knowledge. There have been significant developments in recent years in classifying PPH and in elucidating the disease mechanisms. Limited progress has been made in developing effective therapies, even though lung transplantation may still be the only long-term option.

Dr Mikhail and colleagues, in their first article last month,[1] provided a clear definition of PPH and demonstrated the current level of understanding of the pathology and pathobiology. The role of genetics in the aetiology of the familial form of PPH and the contribution of trigger factors was explained in a manner invaluable to the non-specialist dealing with the family. Discussion of the various underlying mechanisms, including the possible role of an imbalance of vasoactive inhibitors leading to PPH, ultimately may lead to the development of effective therapies. The second article on pages 330-6 deals with the various therapeutic options now available for the management of this condition;[2] the most important message being that if PPH is suspected, early referral to a specialist unit is mandatory.

As a family doctor, the knowledge imparted by the articles would have greatly helped my counselling of my index family and the reviews provide a sure base for further progress.

UK Diabetes (formally the British Diabetic Association) has provided a model where both professionals and patients can together make major contributions to the management of chronic disease. Recently, patients, families and professionals have been drawn together in meetings hosted by the British Cardiac Society. The Pulmonary Hypertension Association now provides a focus for patients, families and doctors to collaborate in research, in management, fundraising, mutual support and dissemination of knowledge about PPH. These review articles provide a foundation for such a process and will enable generalists to understand, explain and contribute to the management of this rare, life-threatening disease.

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References

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