My doctor was simply unaware of this disorder…

Stephen A Margolis

Medicine is a science of uncertainty and an art of probability (William Osler).1

To the untrained eye, the multitude of symptoms and signs our patients reveal suggest confusion and uncertainty. Development of the clinical method has provided a systematic means to unravel this apparently disparate information, sort the core data from the ‘noise’ and proceed in a reductive fashion towards a single diagnosis. The principle attributed to the 14th century friar William of Ockham remains central to this process: ‘an explanation of the facts should be no more complicated than necessary’.2

The success of this process is without question, leading to a vast catalogue of clearly defined clinical entities whose natural history and pathways for management are well known. Interestingly, with the accelerating pace of basic and clinical research, especially over the last generation, many previously obscure maladies that were loosely associated with mental health or the consequences of specific lifestyles have been reclassified as diseases with a clearly defined pathophysiological/pathological basis. Perhaps the best known is the transformation of gastric ulcers from a somatoform, stress-related lifestyle disorder to infectious disease. Apart from a dramatic increase in our knowledge and understanding of common diseases, there has been a similar-sized but much less well-known explosion in the discovery of distinct, newly described diseases. Unlike well-known diseases such as diabetes and hypertension, many of these new conditions are individually rare (prevalence <1 per 2000 people).3 Yet, as detailed by Elliott and Zurynski,4 when rare diseases are considered as a group, 8% of the population have a rare disease.

This leaves the general practitioner (GP) in somewhat of a conundrum when confronted with a patient with persistent undifferentiated and somewhat confusing clinical findings. As discussed by Stone,5 there is an intrinsic dilemma in deciding when and how to pursue atypical collections of symptoms and signs, (ie those patients without a ‘diagnostic story’). Is this merely a manifestation of an unusual presentation of a common problem, masked anxiety or depression, or a contested illness (eg Lyme disease)? Perhaps this is a truly elusive disease requiring more intense investigation and targeted referral, while being mindful of the dictums in the Choosing Wisely program, to focus on judicious and selective use of tests and other resources.6

Maybe it is none of the above as this patient is actually presenting with the standard features of a known and well-defined rare disease, which requires less common tests to diagnose. If so, it would be unrealistic to expect the GP to be familiar with each and every rare disease, even though timely diagnosis is often the key, as rare diseases are more likely to result in chronic, progressive, degenerative and often life-threatening consequences. Once diagnosed, patients with rare diseases are often confronted with a healthcare system less attuned to their needs. The learning curve for patients and their clinicians is often steep and tortuous in the face of limited and incomplete information. Ypinazar7 illustrates some of the challenges from a patient/parent perspective, especially the need to look outside traditional spaces to find the answers. Dudding-Byth8 considers the challenges for the GP.

Interestingly, this is similar to the challenges of managing cancer of unknown primary. Despite these patients being situated in a specific disease category, as detailed by Vajdic and Goldstein,9 the logistics and detail of management are far less structured than those for patients with common cancers such as breast cancer.

General practice has traditionally been based on the longitudinal relationship between clinician and patient. Integrated diagnostic, therapeutic and supportive pathways have been developed for the common diseases, with GPs playing an integral role in the care of these patients. Rare diseases have not participated in these developments, leaving many of these patients adrift in the system. Hence, a strong connection between patients and their GP is even more important for this group. As rare diseases present special challenges, these patients may find a strong relationship with their GP particularly rewarding.

Author
Stephen A Margolis MBBS, MFM, MD, GEM, DRANZCOG, FRACGP, FACRRM, Senior Medical Officer with the Royal Flying Doctor Service, DRANZCOG, FRACGP, FACRRM, Senior Medical Editor, Australian Family Physician, Professor, School of Medicine, Griffith University, Queensland; Medical Officer with the Royal Flying Doctor Service.

References
3. EURORDIS. What is a rare disease? Available at www.eurordis.org/sites/default/files/publications/Fact_Sheet_RD.pdf [Accessed 14 August 2015].
4. Elliott EJ, Zurynski YA. Rare diseases are a ‘common’ problem for clinicians. Aust Fam Physician 2015;44:630–33.