The common problem of rare disease in general practice

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ABSTRACT

- Rare diseases affect 6%–10% of the population, which equates to about 1.2 million people in Australia having a rare disease.
- The United States, the European Union and many other nations have coordinated policies and patient advocacy groups for rare diseases as a group.
- Australia has enacted orphan drug legislation, but there is no coordinated approach either from government or from patient groups.
- General practitioners see rare diseases commonly, but their role for this group has not been adequately described.
- People with rare diseases and their families have similar experiences despite their different diagnoses. GPs are well placed to help with these problems.
- The development of a generic general practice strategy for these patients may improve their overall care.

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In a combined total of 25 years of general practice experience, we have cared for patients with acromegaly, prolactinoma, Addison disease, glioblastoma, narcolepsy, corneal dystrophy, congenital hypothyroidism, listeriosis, juvenile-onset arthritis, Huntington disease, muscular dystrophy, syringomyelia, Marfan syndrome, Guillain–Barré syndrome, adamantinoma, Laurence–Moon syndrome, reflex sympathetic dystrophy, and the list goes on. Why are we seeing so many rare diseases?

Rare disease is common. Estimates suggest that between 6% and 10% of the community suffer from a rare disease at any one time.[1,2] This prevalence is similar to that of type 2 diabetes mellitus. There are significant funded strategies for rare diseases in Europe, the United States and other countries. What is happening in Australia, and what is the role of general practice in rare diseases?

How common is rare disease?

In the US, the National Organization for Rare Disorders (NORD), and the National Institutes of Health, Office of Rare Diseases (ORD), estimate that 25 million Americans suffer from a rare disease.[2,3] Based on a US population of 298 million, this gives a prevalence of 8.4%.

The European Organisation for Rare Diseases (Eurordis) estimates that the prevalence of rare diseases is 6%–8%,[1] which translates to about 30 million people in the European community. We have been unable to find original data on which these estimates were based. A current Eurordis study[4] has examined the prevalence of 230 rare conditions and reports a cumulative prevalence of 1.7%. Based on the lowest of European estimates (6%) and an Australian population of 20 million, it is probable that about 1.2 million Australians suffer from a rare disease.

Diverse diseases, common experiences

A 2005 Eurordis survey[5] of 5980 patients suffering from one of eight rare diseases identified delayed diagnosis as a major issue: 25% of respondents reported waiting between 5 and 30 years from onset of symptoms to a confirmed diagnosis. Forty per cent of respondents reported an initial wrong diagnosis. This resulted in inappropriate surgery (16% of respondents), medication (33%), or psychological care (10%). Forty-five per cent of respondents reported poor communication about the diagnosis.

Eurordis lists the problems faced by patients with rare diseases and their families as: lack of access to the correct diagnosis; lack of information; lack of scientific knowledge; social consequences; lack of appropriate quality health care; high cost of the few existing drugs and care; and inequities in treatment and care.[6]

International rare disease strategies

In 2002, the US Congress passed the Rare Diseases Act.[7] This was the culmination of 30 years of work by NORD, which had successfully lobbied for the 1983 Orphan Drug Act and for the establishment of the Office of Rare Diseases in 1993. The Act provided legislative authorisation for the Office of Rare Diseases and recurrent budgetary expenditure of US$25 million. The main focus of the US strategy has been to support coordination of disease-based support groups, dissemination of information about rare diseases, and research into commercially non-viable treatments.

Eurordis was established by a coalition of patient-support groups and the European Union in 1997. One of the major innovations funded by the European Union is Orphanet,[8] a web-based database of rare diseases, centres of excellence and patient-support groups. Furthermore, many European countries have developed specific public policies on rare diseases.[6] France has published a national strategy for rare diseases,[9] which includes specific training at all stages of medical education in the recognition and treatment of rare diseases. New Zealand (http://www.nzord.org.nz/) and Canada (http://www.cord.ca/) both have peak patient-representative groups for rare diseases. The original impetus for this effort came from patients seeking access to drugs that, without a focus on rare diseases, lacked regulatory approval or commercial viability (termed "orphan" drugs). The strategies appear to have had some success in increasing access to these drugs.[10] The wider impact of such national strategies on the problems faced by sufferers of rare diseases has yet to be assessed.

Australia has had an orphan drug program since 1998.[11] The Australian Government is about to implement the National Chronic Disease Strategy. This rightly focuses on the health priority areas of the common chronic diseases. In September 2005, we contacted chronic disease groups of all state and territory health departments and the Australian Government Department of Health and Ageing; we have not been able to identify any coherent policy aimed at improving the care of the many Australians suffering from rare chronic disease.

The primary care gap

We have found no published information about the primary care role in rare diseases. The response to rare disease by organisations such as NORD and Eurordis has focused on making information more accessible and on coordinating research efforts into rare conditions. This approach seeks to connect isolated patients with specialised knowledge and specialist clinicians. However, general

practitioners also see rare conditions frequently.[12,13] What is the role of the GP in patients with rare disease?

Many patients with rare diseases will present their symptoms first to a GP. They will also attend a GP in between visits to the specialist, they will require diagnosis and treatment of common ailments, and will benefit from the preventive health services offered by general practices. They will require the accessible, relationship-based advocacy and support role that is at the heart of good general practice. The same GP will often perform this role for their patients' carers. A thoughtful, proactive, ongoing response in the context of a continuing relationship with a GP may reduce many of the negative experiences of patients with rare diseases listed above.

In the same way as we are starting to develop a comprehensive approach to the management of chronic disease in primary care,[14,15] the common experiences of those with rare disease may be improved by a systematic generic approach. The final structure of this approach should be developed in consultation with the profession, but some possible features of such an approach are given in the Box.

A systematic, generic primary-care approach to rare disease may reduce problems such as lack of coordinated care, lack of information, delayed diagnosis, and other difficulties encountered by people with rare diseases and their carers. It may enable the GP, inevitably confronted by a person with a rare condition, to approach their care systematically. Examples of generic approaches to clinical problems in primary care have been described for negotiating uncertainty[16] and managing chronic disease.[14] However, we have not found any evidence that a comprehensive approach to chronic rare disease is being developed in primary care in Australia or overseas.

Strategies for rare disease in Australia

We suggest four strategies to enhance the care of patients with rare disease in Australia.

First, the epidemiology of rare diseases needs to be detailed for Australia.

Second, it is time for better coordination of patient-support groups and centres of excellence at a national level. The establishment of an umbrella organisation, like the Office of Rare Diseases, is long overdue. Linkage with overseas databases and experts would improve access to information.

Third, the patient experience of rare diseases needs to be explored, looking for commonalities and intervention opportunities.

Finally, general practice as a specialty has the opportunity to develop a generic approach to the common problem of rare diseases.

A generic general practice approach to patients with rare disease

For a patient with a rare disease the GP will:

Diagnose. Ask more frequently "Could it be a rare disease?" Recognise deviations from common patterns of disease. Be judicious in testing for low-prevalence disorders. Wisely use specialist services for precise diagnoses.

Attend to the whole patient. Provide high-quality care for other health issues including unrelated common conditions and preventive activities (eg, immunisation, screening and health promotion).

Know the disease. Become knowledgeable about the rare diseases encountered, including natural history, evidence-based treatment options, systematic long-term care, associated problems, and genetics. Seek out appropriate specialist services, international centres of excellence, and local organisations which offer relevant services.

Empower the patient. Encourage patients and their carers to ask questions, and assist them with self-care and decision making.

Support the family. Contribute to the physical, emotional, psychological, spiritual, and social needs of the patient's support network.

Advocate. Support the patient's journey through social service and medical bureaucracies, and interpret written and verbal information. ◆

Competing interests

None identified.

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