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ANNOTATION

Call for a national plan for rare diseases

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Abstract: Australia requires a national plan, similar to plans developed internationally, to address the impacts of rare diseases on individuals, the community and health services. Rare diseases often present in childhood, many are chronic, some life threatening and others associated with significant disability. However, diagnosis is often delayed, because of lack of knowledge and experience of health professionals and uncertainty about where to refer. Specialised health services are frequently lacking and specific therapies are often not available, partly because of lack of research funding directed towards rare diseases. A national plan would facilitate a coordinated response to service development, carer support, and health professional and community education, and would promote research and advocacy for affected children and their families.

Key words: disease burden; orphan drugs; rare diseases.

There is an urgent need for a coordinated national response to rare diseases in Australia. Rare diseases impose a significant burden on affected individuals, their carers, health providers, health resources and the community.[1] Although, by definition, rare diseases occur infrequently (most have a prevalence of less than 1 in 2000), it is estimated that there are approximately 7000 rare diseases and that they affect 6–10% of our population. This equates to 1.2 million Australians – similar to the number that suffer from diabetes.[1]

Rare diseases often have their onset in childhood and most continue throughout life and may be disabling or life threatening and difficult to diagnose and treat. Although early diagnosis and appropriate treatment of rare diseases may save health resources, diagnosis is often delayed because of lack of health professional knowledge and lack of specialised clinics. Such multi-disciplinary clinics, which promote access to specialists and decrease health costs, are themselves rare.[1] For many rare diseases, no specific treatment is available and this relates, in part, to the lack of research funding directed at treatments for rare diseases. When diseases are rare, there is little financial incentive for the pharmaceutical industry to develop new therapies and individual patients often lack effective advocates. Rare diseases that are potentially preventable (e.g. fetal alcohol syndrome, genetic disorders that can be diagnosed prenatally and severe seatbelt injuries) deserve particular attention from public health agencies.

Key Points

1. Collectively, about 7000 rare diseases impose a significant burden on affected individuals, their families, health professionals and the community.
2. The United States and the European Union have responded to this burden by developing integrated national plans that support development and access to health and community services, education and research, and advocate for equitable access to treatment for people affected by rare diseases.
3. Australia needs a coordinated National Plan to respond to the significant burden of rare diseases.

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Despite the number of people in our population affected by a rare disease, there is no coordinated effort to provide current information to health professionals and carers who, as a result, often feel isolated and under-supported.[1–3] Similarly, there is no national effort to support the development of specialised health and community services for children affected by rare diseases and their families or to promote research into rare diseases. Australia needs a national plan for rare diseases.

The idea of a National Plan for Rare Diseases is not new. The French, who developed a National Plan in 2004,[4] identified 10 strategic priorities to address the multiple impacts and complex nature of rare diseases.

The aim of the plan is to:

1. increase knowledge of the epidemiology of rare diseases;
2. recognise the specificity of rare diseases;
3. develop information for patients, health professionals and the general public concerning their diseases;
4. train health professionals to better identify rare diseases;
5. organise screening and access to diagnostic tests;
6. improve access to treatment and the quality of patient care;
7. continue efforts in favour of orphan drugs;
8. respond to the specific accompanying needs of people suffering from rare diseases;

9 promote research on rare diseases; and
10 develop national and international partnerships.

The French Parliament has also adopted the law on equality of rights and opportunities, participation and citizenship of the handicapped *(loi sur l'égalité des droits et des chances, la participation et la citoyenneté des personnes handicapée)*, which specifically highlights rare diseases.[4] In the United States, the Rare Diseases Act was passed in 2002 and the Office of Rare Diseases was established within the National Institutes of Health, with the rationale that: '*Because of the small number of affected patients in any one location, rare diseases research requires collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed national research resources and patient populations. Knowledge about rare diseases may offer leads for scientific advancement in other rare diseases and in more common diseases*'.[5] The European Union's (EU) European Task Force on Rare Diseases is charged with supporting research into rare diseases that will provide evidence to support policy development, provision of specialised services and community support. Having recently completed a public consultation on rare diseases, the EU Task Force is currently calling on all EU nations to adopt a co-ordinated national plan similar to the French National Plan.[6]

Over the last 10 years, the EU and the United States have also responded to rare diseases through initiatives that include the development of internet portals that provide information for patients and health professionals (http://www.orpha.net and http://rarediseases.info.nih.gov/). To raise awareness of rare diseases, the EU recently held a Rare Diseases Day,[7] which culminated in a Public Hearing on Rare Diseases at the European Parliament in Brussels. In the United States, the Office of Rare Diseases recently launched its Undiagnosed Diseases Program,[8] which brings together expertise from all over the United States to provide diagnostic and management services that can be accessed by patients and doctors. Australia is well placed to benefit from this extensive overseas experience by adapting some of these strategies to the local situation.

In our local region, a national organisation for rare diseases has been established in New Zealand (New Zealand Organisation for Rare Disorders (NZORD); http://www.nzord.org.nz). This charitable trust was set up in 2000, originally to support families of patients with rare diseases. The formation of a similar Australian Organisation for Rare Diseases would provide the focus needed to respond to the burden of rare diseases and to advocate on behalf of people affected by rare diseases.

Australia has a high international profile in the area of rare diseases in children. The Australian Paediatric Surveillance Unit (APSU; http://www.apsu.org.au) is a unique national research resource established in 1993 to facilitate the study of rare childhood diseases of significant public health importance and which impact on health resources. Over the last 15 years, more than 300 researchers, mostly paediatricians, have used the APSU to study over 40 rare conditions (including rare infections, vaccine-preventable diseases, mental health disorders, congenital and genetic conditions and injuries). Cases are identified and reported each month by paediatricians throughout Australia. The resulting data have informed clinical practice, public health policy and advocacy, and have contributed to improving the health and well-being of Australian children.[9] The APSU is funded by the National Health and Medical Research Council and the Department of Health and Ageing. It is a Unit of the Division of Paediatrics of the Royal Australasian College of Physicians (RACP) and a member of the International Network of Paediatric Surveillance Units and has close links with paediatric surveillance units overseas.

Another Australian initiative is the Pulmonary Interstitial and Vascular Organisational Taskforce (http://www.lungnet.com.au/content/view/5/5/), a Rare Lung Diseases Taskforce established by The Australian Lung Foundation to address these rare respiratory disorders. The Foundation Supporting Medical Innovation for Life Enhancement (http://www.smilefoundation.com.au) and the Steve Waugh Foundation (http://www.stevewaughfoundation.com.au) are the only Australian charities focused exclusively on rare childhood diseases and provide information and financial support to affected families. It funds Australian medical research into rare conditions, providing financial grants in an area where there is a severe funding gap, and offers information and support to affected families.

The need for a systematic national approach to rare diseases is increasingly being recognised, as indicated by a flurry of recent commentary in the medical literature, which highlights the important medical and social issues associated with rare diseases.[1,3,10–12] Australia must respond to the challenges commonly faced by children, families and health professionals, by ensuring provision and equitable access to appropriate diagnostic, treatment, community and educational services; by supporting scientific, clinical and social research into rare diseases; and by equipping heath professionals and carers to advocate for affected children.

As a first step, we have established a task force to develop a national plan for rare diseases in Australia. The task force includes clinicians, researchers, epidemiologists, public health personnel, consumers and parent advocates, as well as representatives from the NZORD, RACP, Association of Genetic Support of Australasia, the APSU and the European Organisation for Rare Diseases. We have met to draft an outline of the plan and our next step will be to engage with government, and peak health professionals and research organisations to seek input into and endorsement of the plan.

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