



Gene Advocate

Issue 63

Rare Diseases

What is a rare disease?

Internationally, definitions vary among countries. Australia, for example, defines a rare disorder as a condition that affects about 2,000 people, while in the United States it is about 200,000 people.

In Europe, a disease or disorder is defined as rare when it affects less than 1 in 2000 citizens (Orphan Drug Regulation 141/2000). Rare diseases may affect 30 million European Union citizens.

Although by definition each rare disease occurs infrequently, collectively, approximately 8000 rare diseases affect approximately 6-10% of the population. This is equivalent to 30 million people in Europe, 25 million people in the US and 1.2 million Australians.

It is increasingly acknowledged that low prevalence does not equate with low impact. Although rare diseases vary considerably in aetiology, there are many commonalities. Rare diseases often have their onset in childhood, continue throughout life, are difficult to diagnose, are disabling and have significant impacts and burdens on the patient, family,

community and health services.

Despite this, rare diseases receive little attention. For many rare conditions epidemiological and scientific data are lacking, making the development of evidence based practice and policy impossible.

Many rare diseases are serious and/or chronic. Many are life-threatening. Even so, people with rare diseases often have trouble accessing the medical or other services they need because those making the decisions are not familiar with their diseases.

Characteristics of rare diseases

- Rare diseases are often chronic, progressive, degenerative, and often life-threatening
- Rare diseases are disabling: the quality of life of patients is often compromised by the lack or loss of autonomy
- High level of pain and suffering for the patient and his/ her family
- No existing effective cure
- There are between 6000 and 8000 rare diseases
- 50% of rare diseases affect children

- ✓ 80% of rare diseases have identified genetic origins. Other rare diseases are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative.

Rare diseases are characterised by a broad diversity of disorders and symptoms that vary not only from disease to disease but also from patient to patient suffering from the same disease. Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis.

Rare disease patients face common problems:

- Difficulty in obtaining an accurate diagnosis
- Limited treatment options
- Little or no research being done on the disease
- Difficulty finding physicians or treatment centers with experience in treating a particular rare disease
- Treatments that are generally more expensive than those for common diseases
- Reimbursement issues related to private insurance, and Medicare
- Difficulty accessing medical, social, educational or financial services or assistance because those making the decisions are not familiar with the disease
- Feelings of isolation and of having been “forgotten” by our healthcare system.

How can things change?

- ✓ By implementing a comprehensive approach to rare diseases
- ✓ By developing appropriate public health policies
- ✓ By increasing international cooperation in scientific research
- ✓ By gaining and sharing scientific knowledge about all rare diseases, not only the most “frequent” ones
- ✓ By developing new diagnostic and therapeutic procedures
- ✓ By raising public awareness
- ✓ By facilitating the network of patient groups to share their experience and best practices
- ✓ By supporting the most isolated patients and their parents to create new patient communities or patient groups
- ✓ By providing comprehensive quality information to the rare disease community

Rare Disease Day February 28th 2011

This will be the fourth annual Rare Disease Day. The concept was launched in 2008 by EURORDIS, NORD’s counterpart in Europe. Last year, EURORDIS asked NORD to host Rare Disease Day in the U.S. In 2010, Rare Disease Day was observed in 46 nations around the world including Australia. The observance is always on the last day of February.

Information Source:

Australian Paediatric Surveillance Unit. <http://www.apsu.org.au/index.cfm?objectid=64B91B18-A313-7C8D-864397E5A22934D0>
Canadian PKU and Allied Disorders Inc. 4/6/2010. <http://canpku.org/pku-news/90-canada-not-fit-to-deal-with-rare-disorders>
Eurordis, Rare Diseases Europe. <http://www.eurordis.org/content/what-rare-disease>
NORD Rare Disease Day. <http://rarediseaseday.us>

Rare Disease Resources



The NORDD Rare Disease Support Community connects patients, families, friends and caregivers for support and inspiration.

This community is sponsored by the National Organization for Rare Disorders, an Inspire trusted partner.

Joining Inspire is completely free and usually takes less than a minute, visit: <http://www.inspire.com/groups/rare-disease/>

The Australasian Genetic Alliance (AGA)



The Australasian Genetic Alliance (AGA) facilitates networking

between groups, health professionals and patients and families to improve community and professionals knowledge of genetic conditions, their impact and available services.

The Alliance has taken a lead advocacy role in representing members interests in national genetics issues.

The AGA provides written and verbal submissions to government and others in the field of genetics and participates with like bodies nationally to further represent genetic support groups and their members.

AGA Member Groups

- Genetic Support Council WA (GSCWA)
- Genetic Support Network Victoria (GSNV)
- New Zealand Organisation for Rare Disorders (NZORD)

- ACT Self-Help Organisations United Together (SHOUT)
- The Association of Genetic Support of Australasia (AGSA)

Visit: www.australasiangeneticalliance.org.au

Genetic Support Groups



Our recently updated Directory of Genetic Support Groups & Community Support Organisations contains some information and details of rare genetic diseases and conditions.

The directory is available in PDF format on our website www.geneticsupportcouncil.org.au or you can ph 9485 8999 to order a hard copy.

Become a Rare Disease Day Partner or Ambassador

Rare Disease Day is about realizing the similarities among all individuals impacted by rare diseases. By working together we can raise awareness and create a global impact. Whether you are an individual or part of an organization, we want you to show your support by becoming a Partner or Ambassador.

Visit: <http://rarediseaseday.us>

