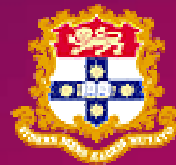


The Burden of Rare Diseases: How is Australia Responding?

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Literature search and review of web-based resources internationally to describe:

- The impacts of rare diseases on families, clinicians and health services
- Responses from governments, clinicians, and families
- To determine next steps



Are Rare Diseases Common ?

EU: 1/2000 population

US: Affecting < 200,000 affected individuals

Australia (TGA): < 2000 affected individuals

There are ~ 7,000 rare diseases

Estimated 6-10% of population affected

- **30 million in Europe**
- **25 million in the US**
- **1.5 million in Australia (250,000 children)**

In 26% of disabled children, the disability was attributed to a rare disease (*Guillem Arch Dis Child 2008*)

Until recently rare diseases received little attention

- **Forgotten**
- **Neglected**
- **Orphaned**

Low prevalence ≠ Low impact

Rare Diseases – common features

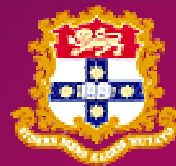
Many rare conditions:

- Have onset in childhood
- Difficult to daignose
- No cure
- Lead to lifelong health problems and disability
- Complex
- Require multidisciplinary services
- Impact on health and community resources
- Have enormous social consequences



Impacts on patients and families

- Diagnostic delays (sometimes years)
- Stress of not knowing
- Isolation
- Lack of services (health, education, community)
- Lack of information (local content)
- Financial burden



Impacts on patients and families

*“Obtaining the correct diagnosis has had significant implications... We were able to understand and accept Rett Syndrome... We are finally able to explain to others when they ask what is wrong and to foster more realistic expectations of our child’s future...and also to further extend our family”
(Mother of a girl affected with Rett Syndrome)*



Impacts on patients and families

"I had to find the right doctors and to be insistent but not obstinate. On the one hand, I had to save Mathilda from unnecessary diagnostic tests, often saying "no" and "why". On the other hand, I had to ensure that she received everything she needed"



"When Mathilda was nearly 2 years old, she was still able to sit in her doll's bed"

(Dunkelberg S. BMJ 2006)

Impacts on Clinicians

Primary care physicians, specialists, allied health professionals affected

Lack of, or lack of access to evidence based information and education educational opportunities

- Diagnostic delays
- Multiple, sometimes unnecessary investigations
- Lack of specialised diagnostic and referral services
- Lack of parent support groups

“Zebras on the commons” (*Phillips BMJ 2004*)

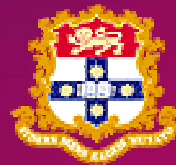
“Common problem of rare diseases in general practice” (*Knight, MJA 2006*)

Impacts on Clinicians

Rare diseases are under-researched

This Gap being addressed by clinicians themselves e.g.:

- British Paediatric Orphan Lung Diseases Registry
- UK Obstetric Surveillance System (UKOSS)
- InterRett Database



Impacts on Health Services

- Health expenditure – frequent and specialised nature of services required
- Scarce resources – staff, equipment, clinic time, expertise
- Access to specialised clinics difficult

For Example: Only 2 multidisciplinary Rett Syndrome clinics in Australia and no specialised clinics for children affected by FAS

- Lack of continuity of care

Specialised Clinics for Rare Diseases The Agrenska Centre, Sweden

Since 1989 has provided integrated multidisciplinary services for children affected by over 140 rare diseases

Family counselling and respite services, referral services, educational resources, information on latest research



Cost effectiveness:
Average cost per child is 33% of costs per child accessing non-integrated programs

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Government Response to Rare Diseases

Orphan Drugs Development Policies

- 1983 US Orphan Drug Act
- 1991 Singapore
- 1993 Japan
- 1998 Australia
- 2000 EU

TGA Orphan Drugs Program brings orphan drugs to market more quickly by:

- Waiving fees associated with registration by TGA
- Priority review
- Approving if already approved by US FDA (current evaluation report)

E.g. Lamivudine for HIV infection in children

Government Response to Rare Diseases

National Planning and Health Policy

- 2002 US Rare Diseases Act, NIH Office for Rare Diseases established
- Rare diseases priority EU Public Health Programme 2003-08
- 2005 France: Rare Diseases Plan 2005-08
- Now adopted by at least 9 EU countries

- No national plan in Australia or New Zealand

French National Rare Diseases Plan 2005-2008

Ten strategies to ensure 'equity in the access to diagnosis, to treatment and to provision of care' for people with rare diseases:

- Increase knowledge of the epidemiology of rare diseases
- Recognise the specificity of rare diseases
- Develop information for patients, health professionals and the general public concerning rare diseases
- Train health professionals to better identify rare diseases
- Organise screening and access to diagnostic tests
- Improve access to treatment and the quality of patient care
- Continue efforts in favour of orphan drugs
- Respond to the specific needs of people suffering from rare diseases
- Promote research on rare diseases
- Develop national and European partnerships



Government Response to Rare Diseases

Priority Funding:

Italy €8.3 million p.a. for basic and clinical research and clinical trials

NIH Office of Rare Diseases launched the Undiagnosed Diseases Program

No priority funding initiatives in Australia by the NHMRC nor the ARC

National Support Groups for Rare Diseases

USA NORD www.rarediseases.org

Canada CORD www.raredisorders.ca

New Zealand NZORD www.nzord.org.au

UK Contact a Family www.cafamily.org.uk

European Union – Eurordis www.eurodis.org

Orphanet www.orpha.net

Belgium, Bulgaria, Denmark, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Luxemburg, Netherlands, Portugal, Romania, Spain, Sweden

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National Support Groups for Rare Diseases

Australia

The Association of Genetic Support of Australasia (**AGSA**)

www.agsa-geneticsupport.org.au/links

Association for the Wellbeing of Children in Healthcare (**AWCH**) www.awch.org.au

The SMILE Foundation www.smilefoundation.org.au

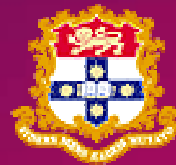
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Australian Paediatric Surveillance Unit APSU

- Surveillance for rare childhood conditions
- Infectious diseases, genetic diseases, mental health disorders, rare injuries (studies on 44 conditions completed)
- Systematically collected national enhanced data on epidemiology, diagnosis, risk factors, clinical management and short-term outcome
- Translation into policy and practice
- Supported by DoHA, RACP, University of Sydney, competitive grants (NHMRC, other grants)



Rare Diseases in Australia: What is Needed?

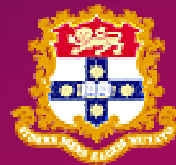
- Co-ordinated national approach to address the complex collective burden of rare diseases
- Priorities and funding for research to address knowledge gaps
- Development of new services based on existing models
- Improved access to information and improved education opportunities for clinicians, families, teachers
(Australian context needed to be useful)
- Co-ordinated collective patient/parent support group to provide information and advocacy

Knight MJA 2006, 2007

Zurynski et. Al. MJA 2007 (letter)

Zurynski et al. ADC, 2008

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Future?

- Determine impacts on children's hospitals – WCHA, AIHW
- Links with SMILE Foundation – development of a web-based support system for families, clinicians
- Creswick Fellowship – study support services overseas
- Advocacy

