



A National Rare Disease Plan for Australia

Elizabeth Elliott

for the
**The National Rare Disease
Taskforce**



What is a rare disease?

- A rare disease (sometimes known as an orphan disease) has such a low prevalence in a population that a doctor in a busy general practice would not expect to see more than one case a year. wikipedia.org/wiki/Rare_disease
- US [National Institute of Health's](http://NationalInstituteofHealth.gov) (NIH) Office of Rare Diseases states that, "an [orphan](http://orphan.gov) or rare disease has a prevalence of fewer than 200,000 affected individuals in the United States"
- EU: 1/2000 population
- Australia (TGA): <2000 affected individuals (1/10,000)



Rare diseases are common

- There are ~ 8,000 rare diseases
- Estimated 6-10% of population affected

- 30 million in Europe
- 25 million in the US
- 2.0 million in Australia (>400,000 children)



Until recently rare diseases received little attention

Rare diseases are

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

Low prevalence \neq Low impact



Rare Diseases - high impact, common features

- onset in childhood
- difficult to diagnose
- no cure
- lifelong health problems, disability
- complex
- require multidisciplinary services
- impact on health and community resources
- significant social consequences

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



Rare diseases impact significantly on children and their families

- diagnostic delay (sometimes years)
- stress of 'not knowing'
- isolation (lack of parent support groups)
- lack of treatments
- lack of services (health, education, community)
- lack of information (local content)
- financial burden

Impacts on children and families

“Obtaining the correct diagnosis 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 children and families

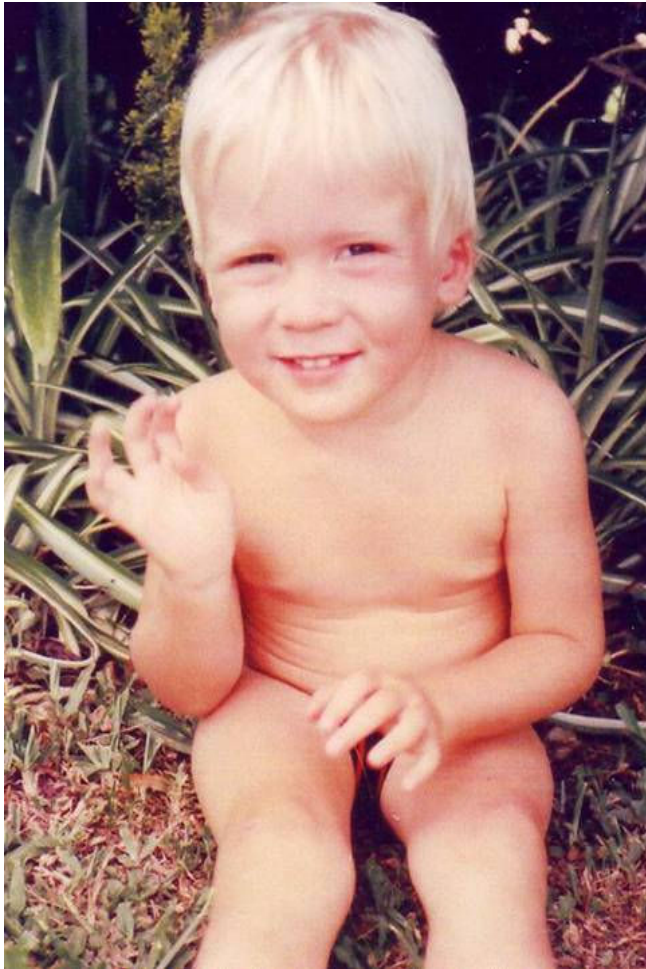


"When Mathilda was nearly 2 years old, she was still able to sit in her doll's bed"

"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"

(Dunkelberg S. BMJ 2006)

Impacts on children and families



I need to do something for him, but I don't know what to do next. I need a diagnosis so that I can help him manage" his problems

Elizabeth Russell.
Alcohol and Pregnancy: A mother's responsible disturbance. Zeus Publications, 2005

Impact on families



- Pompe's disease Dx infancy after death of sister 9y
- Respiratory, cardiovascular disease, Developmental delay.
- Parents heard about Myozyme (aglucosidase alpha) enzyme replacement therapy and relocated to Florida
- Age 4y, moved home, 2/52 Rx, normal Echo, OTx, PTx, STx



Rare diseases impact on clinicians

Primary care physicians, specialists, allied health professionals feel **ill-equipped**

- Lack of research evidence
- Lack of access to updated evidence-based information
- Lack of locally relevant information
- Lack of educational opportunities for HP
- Lack of specialised diagnostic and referral services
 - Diagnostic delays
 - Multiple, sometimes unnecessary investigations

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

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



Impacts on Health Services

- **Health expenditure high**
 - Frequent, specialised nature of services required
- **Scarce resources**
 - Multi-disciplinary staff, equipment, clinic time, expertise
- **Specialised clinics limited**
 - 2 multidisciplinary Rett Syndrome clinics in Australia
 - no specialised clinics for Dx children with FASD
- **Lack of continuity of care and transitional care**

Specialised Clinics for Rare Diseases: The Agrenska Centre, Sweden

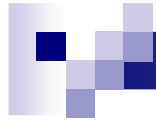
- Established 1989
- Integrated, multidisciplinary services
- Children with >140 rare diseases
- Family counselling, respite, referral services
- Educational resources, latest research
- Cost effective: Average cost per child: 33% of cost of non-integrated programs





Rare diseases are under-researched

- Knowledge gap is being addressed by clinicians e.g.
 - British Paediatric Orphan Lung Diseases Registry (also in Australia)
 - UK Obstetric Surveillance System (UKOSS)
 - Australian Maternity Obstetric Surveillance System (AMOSS)
 - InterRett Database



Australian Paediatric Surveillance Unit: facilitating rare disease research

- National surveillance for rare childhood conditions
- Systematic collection standardised data
 - epidemiology, diagnosis, risk factors, management, outcome
- 44 studies completed since 1993
 - Infectious, genetic, mental health disorders; rare injuries
- Translation into education, policy and practice
- Supported by NHMRC, DoHA, University of Sydney



Government Response to Rare Disease

Orphan Drugs Development Policies

- 1983 USA Orphan Drug Act
- 1991 Singapore
- 1993 Japan
- 1998 Australia
- 2000 European Union

Therapeutic Drugs Administration: Orphan Drugs Program (Australia)

Brings drugs to market more quickly by:

- Waiving fees associated with registration by TGA
- Priority review
- Approving with minimal review if already approved by US FDA

e.g. Lamivudine for HIV infection in children



Government Response to Rare Diseases

- **2002 USA:** Rare Diseases Act, NIH Office for Rare Diseases
- **2003-8 EU:** Rare diseases priority: Public Health Program
- **2005-8 France:** French National Rare Diseases Plan.
 - *To ensure 'equity in access to diagnosis, to treatment and to provision of care' for people with rare diseases.*
- **2008 EU:** EC Commission calls for European Action (EUROPLAN)
- **2008 USA:** NIH launched Undiagnosed Diseases Program
- **WHO:** to include rare diseases in ICD-11 (for release in 2014)
- **No national plan:** Australia, New Zealand or Canada



Resource allocation

- **French Plan:** € 65 million
- **EU Framework Program 6:** € 240 million
- **Italy:** €8.3 million p.a. basic and clinical research
- **USA NIH:** US \$9million Rett Syndrome research, 2008
- **Australia:** No priority funding or initiatives
(from NHMRC, ARC, DoHA)



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 and others
- **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



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
- Livewire (Starlight Foundation) provides information, support and on-line linkage for children with serious diseases in hospital



Draft National Rare Disease Plan: Australia

1. **Raise awareness** of the burden of rare diseases on families, health professionals and the community
2. **Increase knowledge** of the epidemiology and impacts of rare diseases
3. **Improve health care** for people with rare diseases through access to diagnostic tests, new treatments and specialised services
4. **Promote scientific and social research** through development of national and international multidisciplinary research partnerships
5. Develop and disseminate **information about rare diseases for parents, carers and the general public**, that is relevant in the Australian context
6. Provide **educational resources and networking opportunities for health professionals** to allow them to better identify and manage rare diseases
7. Support families affected by rare diseases by facilitating the development of integrated **peer support networks**
8. **Advocate** in partnership with families, for people affected by rare diseases

Rare Diseases in Australia: What is Needed?

Sydney Morning Herald: Common front to fight rare ailments

Julie Robotham Medical Editor, February 28, 2009

(Global rare diseases day: 29.2.09)



"Even if the future was quite grim, at least I knew what it was and we could start to make the best of it," said Zoe's mother.

Zoe, aged 4, who has a rare disorder called Rett syndrome plays with her sister Ella, 7. Photo: *Brendan Esposito*



Rare disease taskforce

- Elizabeth Elliott (APSU)
- Yvonne Zurynski (APSU)
- Carol Bower (TICHR)
- Adam Jaffe (**APOLDR**)
- Elizabeth Sullivan (NPSU)
- Elisabeth Murphy (NSW Health)
- Karen Gair (SMILE Foundation)
- Anne Cutler (AWCH)
- Jenny Proimos (RACP)
- Andrew Knight (General practice)
- Executive Director (NZORD)
- Margot Frazer (NSW Commission for Children)
- Jerome Parisse-Brassens (Eurodis)
- Elizabeth Chatham (WCHA)
- David Silence (AGSA)
- David Lester-Smith (APSU)
- Elizabeth Peadon (APSU)
- Lyndall Beville (SMILE)
- Fiona Stanley (TICHR)
- Melissa Wake
- Mark Farrell
- Neil Wigg
- John Christodoulou
- Dianne Petrie (AGSA)



References

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