

Rare diseases are not so rare Australian Paediatric Surveillance Unit (APSU)

Guest editorial

Professor Elizabeth Elliott AM, MD, MPhil, MBBS, FRACP, FRCPCH, FRCP

Brothers Thomas and Bradley Farrell were born profoundly deaf and have never walked because of their progressive muscle weakness. Also, both boys are gradually losing their sight. Thomas is now 12 and Bradley is 13, and they have both been treated at The Children's Hospital at Westmead throughout their lives.

Their problems began early in life. Every time they caught a cold, it would progress to pneumonia and their breathing muscles would be totally paralysed. They spent many weeks of every year in ICU (Intensive Care Unit).

"The lack of specific health policies for rare diseases, and the scarcity of expertise, translates into delays in diagnosis and difficulty for families in accessing the care that they need. There is also a lack of research to guide health professions and where specific treatment and care does exist, the cost can be exorbitant," explains Professor Elizabeth Elliott AM, Director of Australian Paediatric Surveillance Unit (APSU).

This was the case with the Farrell boys. Initially no-one could give the family an accurate diagnosis. Eventually they were diagnosed with the very rare genetic disorder, Arts Syndrome.

Recently, researchers made a significant breakthrough, identifying the gene responsible. This has led to oral therapy for the boys, so for the last two years they have spent very few days in hospital.



Thomas and Bradley Farrell

Professor John Christodoulou AM, Director of the Western Sydney Genetics Program, part of Kids Research Institute at The Children's Hospital at Westmead, cares for the boys and leads the research team.

His work will improve understanding of these rare genetic disorders to enable development of better diagnostic tests and more targeted therapies.

"Paradoxically, rare diseases are common," said Prof. Elliott. "There are over 8,000 known rare diseases

which collectively affect 7-10% of our population. That equates to somewhere between 1.2 million to 2 million Australians. Many have their onset in childhood."

Patients with rare diseases and their families feel isolated and vulnerable. Life expectancy is often reduced. Severe disabilities can reduce educational, professional and social opportunities. There are unique and difficult medical, financial and social challenges these families face on a daily basis.

The impacts of rare diseases go beyond affected individuals. They have wide-reaching impacts on families and carers, health professionals, health resources and the wider community.

APSU is advocating for the development of a National Plan for Rare Diseases to improve health services, treatment and research. APSU convened a National Rare Diseases Working Party made up of medical experts, support groups and rare disease advocates. The working party drafted an agenda based on eight guiding principles that could be used as a basis for a national plan for rare diseases. The plan is now out for consultation at

www.apsu.org.au

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Further news about carer legislation in NSW

Laraine Toms, President, Carers NSW

Last time I wrote about the Carers Recognition Bill tabled in NSW Parliament by Liberal MP Andrew Constance. Thank you to the carers who have written to tell us about contacting their local member to seek support for the Bill. Thanks also to all of you who talked about the legislation to friends, family and colleagues. Your work has really made a difference. Never before have carers been so much the focus for politicians!

Since last writing the NSW Government has also tabled a Carers (Recognition) Bill. The Opposition has also introduced its Bill into the Upper House through Senator Robyn Parker. But what does it mean now that there are two Bills to be debated? Where does Carers NSW stand in all of this? As you know Carers NSW does not support any political party – we are non-political. We support good policy. We are delighted politicians of all parties are supporting carers. We do not support one Bill over the other. Essentially they are the same. Carers NSW was consulted in the development of both and we are confident that both Bills provide a good legislative base for supporting carers. We anticipate that the Government's Carer (Recognition) Bill will be the Bill passed into law because of the process that is followed in parliament.

In addition, the Federal Government has introduced a Carers Recognition Bill. We consider that Bill to be a start – recognition for carers at the national level but with no practical details. It is our hope that it will evolve into further legislation that has real 'teeth' as the NSW legislation has.

Carers are the clear winners in this confusing process. All carers will benefit. And that is our job here at Carers NSW – to work with and for all carers, no matter who they care for and what type of disability, illness or disorder their family member has. We work to ensure all carers are recognised and valued, their needs met and their voices heard. That is the advantage of being a peak body

for all carers. We have the strength, the numbers and the expertise to be credible to governments and the community. Your support for us has made this legislation possible. Through us your voices have been heard.

What will the legislation do for carers? You can read the entire Bill at [www.parliament.nsw.gov.au/prod/parlament/nswbills.nsf/131a07fa4b8a041cca256e610012de17/d908704e7ff4a0b1ca25770b001bccf3/\\$FILE/carers.pdf](http://www.parliament.nsw.gov.au/prod/parlament/nswbills.nsf/131a07fa4b8a041cca256e610012de17/d908704e7ff4a0b1ca25770b001bccf3/$FILE/carers.pdf)

The objects of the Bill are:

- (a) to enact a NSW Carers Charter to recognise the role and contribution to our community of persons who care for other persons who have a disability, a medical condition (such as a terminal or chronic illness), a mental illness or are frail and aged, and
- (b) to require certain public sector agencies to take action to reflect the principles of the NSW Carers Charter when providing services that affect carers
- (c) to establish a Carers Advisory Council (which will have, I am assured by Deputy Premier Carmel Tebbutt, amongst others, current carers as members).

One of the most important elements of the Bill is that carers will, if they agree, be assessed as to their own health and/or education needs. This will mean that their health will be better looked after. Young carers won't slip through the gaps, they will be identified and helped.

I have heard from some carers who are concerned that they will have to have an assessment before they can access services. That will not happen. It is the carer's choice whether or not to be assessed; the same services will still be available whatever their choice. Carers' privacy will be maintained – you will not have to give any information if you do not wish to.

We are in interesting times indeed. Let's hope that in the next newsletter I can write about the passing into law of the Carers (Recognition) Bill!

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"Developing a coordinated national plan for rare diseases will be the first step to improve the current fragmentation of services, promote research and family support, and enable health professionals to provide best practice," said Prof. Elliott.

A Professor in the Discipline of Paediatrics and Child Health at the University of Sydney and a consultant paediatrician at The Children's Hospital at Westmead, Professor Elliott founded the APSU in 1993.

In June 2008, Professor Elliott was awarded the Member of the Order of Australia for academic services to paediatrics and child health, research, education and the establishment of the Australian Paediatric Surveillance Unit.

The Australian Paediatric Surveillance Unit (APSU) facilitates surveillance and research into childhood diseases. Research findings from APSU have provided new knowledge that has significantly impacted on clinical practice and public health policy. APSU is affiliated with the Royal Australasian College of Physicians, the University of Sydney and the Kids Research Institute, and currently funded by the NHMRC, Australian Department of Health and Ageing, and the Australian Research Alliance for Children and Youth. Please go to www.apsu.org.au for more information.

Carers NSW is able to provide carers of people with rare diseases with support, information, referral and access to counselling and support groups. Please call **1800 242 636** (freecall, except from mobile phones).