



EMBARGOED: Saturday 28<sup>th</sup> February 2009

## Australian Health Professionals Call for National Response to Rare Diseases

On the eve of the second Rare Diseases Global Awareness Day on 28<sup>th</sup> February 2009, Australian health professionals are calling for a co-ordinated national response to rare diseases. Rare diseases are actually common with about 8000 rare diseases affecting an estimated 1.5 million Australians, including about 300,000 Australian children.

The Australian Paediatric Surveillance Unit (APSU), based at *The Children's Hospital at Westmead*, is a unique Australian resource that facilitates rare disease research. "In 2006, paediatricians reporting to the unit identified 570 children newly diagnosed with 16 different rare diseases," said Director Professor Elizabeth Elliott. "However, due to lack of research, there are thousands of rare diseases for which the number of affected children and the appropriate management is unknown."

Each rare disease is different, but they have some strikingly common similarities:

- Most begin in childhood (usually diagnosed in children aged < 2years) and continue throughout life
- Obtaining a definitive diagnosis is often difficult and delayed
- Many rare diseases have no cure or require new high-tech and often expensive treatments
- Neurological and intellectual disabilities occur in about half of all cases regardless of disease type and lead to reduced independence and opportunities
- Families experience isolation, psychological and financial stress
- Health professionals have inadequate access to information, education and resources
- It is estimated that about 35% of deaths in children aged < 1year are due to rare diseases.

Hundreds of children with rare diseases are admitted to *The Children's Hospital at Westmead* each year. Examples of rare diseases include genetic conditions such as Fragile X, muscular dystrophy, Kawasaki disease, Rett Syndrome, and rare infections such as congenital varicella, congenital rubella, neonatal herpes virus, perinatally acquired HIV.

The European Union, Canada, the UK and New Zealand have recognised the complex problem of rare diseases and are responding by establishing national plans for rare diseases that include developing national support organisations for people with rare diseases, making rare diseases a public health and research priority, and developing integrated clinics that pool expertise to enable quicker diagnosis and better access to treatments and interventions.

On the 9<sup>th</sup> February 2009, The Australian Paediatric Surveillance Unit convened a meeting in Sydney to discuss the need for a co-ordinated national approach to providing health care and support to Australians affected by rare diseases.

“The meeting was attended by eminent researchers, health professionals and family support groups, including representatives from the European Rare Diseases Organisation and the New Zealand Rare Diseases Organisation,” said Dr Yvonne Zurynski, Deputy Director APSU and Senior Lecturer, Discipline of Paediatrics and Child Health, University of Sydney

“We discussed the need for an integrated patient support organisation for Australians affected by rare diseases, improved access to information and education for health professionals, and new models of healthcare,” said Dr Zurynski.

A Creswick Foundation Fellowship will allow Dr Zurynski to study health and community services for rare diseases operating overseas and to implement this knowledge in Australia.

Nellie and Huw Evans understand only too well the challenges and frustrations faced by families affected by rare diseases. Their 4-year-old daughter Zoe was diagnosed with Rett Syndrome at the age of two, after first displaying symptoms at 12 months of age.

“Our second daughter Zoe was 12 months old when we sensed she wasn’t developing normally,” explained Ms Evans. “It took another year and a half before we were told our girl had Rett Syndrome. By the time the diagnosis was eventually given, it was almost a relief to be given a concrete reason why she kept losing skills and failing to meet normal milestones.”

Rett Syndrome is a rare disease predominantly caused by a sporadic mutation in the *MECP2* gene on the X chromosome. The syndrome becomes apparent from around six months of age when development stagnates and acquired skills, such as coordination, speech, communication skills and cognitive function deteriorate. Other problems can include, breathing, cardiac function, chewing, swallowing, and digestion. It is estimated that there are currently around 300 girls affected nationally by this condition.

Ms Evans said living with a loved one diagnosed with a rare disease “requires a reinvention of everything you are as a family”.

“There have been triumphs when we can access services and utter despair when we are told our daughter’s disability is too rare to warrant funding or help. Greater understanding of rare syndromes like Rett, and those who have them leads to greater inclusion. Our hope is that Zoe is afforded the same quality of life that we all take as a given.”

“We live with the reality of this disease, but we never ever stop hoping for a cure. Greater awareness equals more funding, which equals more research. At an everyday level, greater awareness means a better quality of life for Zoe.”

Rare Diseases Global Awareness Day will be held on the 28<sup>th</sup> of February 2009 and Australian organisations such as the Smile Foundation ([www.smilefoundation.com.au](http://www.smilefoundation.com.au)), The Association of Genetic Support of Australasia ([www.agsa-geneticsupport.org.au](http://www.agsa-geneticsupport.org.au)) will be holding an event to highlight the importance of rare diseases and the Australian Paediatric Surveillance Unit ([www.apsu.org.au](http://www.apsu.org.au)) have joined the global effort to raise awareness of rare diseases. For more information visit [www.rarediseaseday.org](http://www.rarediseaseday.org)

### ***The Children’s Hospital at Westmead***

*The Children’s Hospital at Westmead* is the largest paediatric centre in NSW, providing the most advanced care and treatment options for children from NSW, Australia and across the Pacific Rim. Established in 1880, *The Children’s Hospital at Westmead* is a stand-alone public Hospital and registered charity with 3,000 staff working in 150 departments. Over 60,000 sick children and their families are cared for each year in a family-focused, healing environment. Funded by the NSW Government, *The Children’s Hospital at Westmead* provides significant enhancements to services aided by the generosity of individual and corporate donors. For more information visit [www.chw.edu.au](http://www.chw.edu.au)

**Interview Opportunities:** Prof Elizabeth Elliott (Director) and Dr Yvonne Zurynski (Deputy Director) from The Australian Paediatric Surveillance Unit are available on request for interview opportunities

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