

AussieRett CONNECT



www.aussierett.org.au

December 2012 - ISSUE FIFTEEN

NEWSLETTER OF THE AUSTRALIAN RETT SYNDROME STUDY

AussieRett Christmas Newsletter 2012

Merry Christmas and a wonderful New Year from Helen, Jenny, Katherine and all of the team at the Australian Rett syndrome study. We hope this time of year brings you and your families much joy and relaxation as you spend time unwinding and being with loved ones.

One focus of our research is on functional abilities in Rett syndrome for which we are currently collecting video footage. This is a very important study as the results will illustrate the natural history of Rett syndrome for families, clinicians and researchers. The video footage allows us to examine specific abilities in detail and develop assessments especially for Rett syndrome which will be relevant for future clinical trials. We would love to be able to follow the development of your daughters in person. Since this is not possible, we instead use the video footage and questionnaires that you send us.

We are grateful to the many families who have already taken video on their cameras and phones and sent clips to us either via the mail or electronically; your time and support are very much appreciated. If you are yet to take video or are doing the video study over the holidays and need support please do not hesitate to contact the AussieRett team on aussierett@icmr.uwa.edu.au or call 0419956946.

Family stories

Ward family 2012 holiday to the USA

The Ward family from NSW enjoyed a wonderful holiday in the USA earlier this year. There were many highlights: a luau in Hawaii, a cable car ride in San Francisco, the Lumberjack breakfast deal in 'Denny's' which fed four family members, and a horse and carriage ride in Central Park at night.

There was much warmth and friendliness towards Jess throughout their travels; she kissed an astronaut at the Kennedy Space Centre and a security guard on top of the Empire State Building! During the Lion King show at Disney's Animal Kingdom, the Disney characters included her in the Circle of Life finale, which was very moving.



left: Mark and his daughter Jess meet a security guard at the top of the Empire State Building.



right: The Ward family at Fisherman's Wharf, San Francisco.

Perth family picnic

A warm spring Perth day was the perfect backdrop for a purple-themed family picnic to celebrate Rett syndrome awareness month in October. Eight families joined members of the AussieRett team in a leafy park opposite the Telethon Institute to renew and develop friendships, share journeys and indulge in afternoon tea. It was a delightful afternoon for everyone and we are sure that other gatherings for Rett syndrome awareness month held around the country were just as successful.



Congratulations

Our sincerest congratulations go to Bill Callaghan who was awarded an Order of Australia medal in the Queen's Birthday Honours List in June for service to community health through founding and leading the Rett Syndrome Association of Australia. Bill is a very deserving recipient of this award. A link to the media article and contact details for the Rett Syndrome Association of Australia can be found on our website www.aussierett.org.au under 'Resources'.

Bill (left) is presented with his award by the Governor of Victoria, the Honourable Alex Chernov, at Government House Melbourne.

Congratulations

Congratulations to Trish and James Donnelly for their dedication and hard work in making their dream of "Lily Grace Place" a respite initiative for Queensland families a reality. In the 2011 AussieRett Connect Christmas newsletter, Trish wrote about the events impacting on her family that led to James and Trish working with the Queensland government on a respite model which would meet the specific needs of children with Rett syndrome and their families. Trish spoke at the Family Conference in May about the progress of the project. In October, a QLD government media release confirmed that \$1 million in recurrent funding would be granted to deliver respite services to families caring for a member with Rett syndrome and other fragile conditions from a purpose-built home that Trish and James will be building. This is an incredible accomplishment which will have enormous impact for Queensland families. Services are expected to commence early 2013 and will encompass in-home, host family and centre-based respite services at Lily Grace Place along with other early intervention services to support families.

Thank you

We have been extremely fortunate this year to receive donations for our Rett syndrome research from incredibly generous families, their friends and supporters, as well as from members of the Australian community. We humbly acknowledge this incredible and much appreciated support. All donations will be used directly for research that will benefit those with Rett syndrome and their families.

You can read about the successes of two of our Rett syndrome research heroes on our Everyday Hero website at www.everydayhero.com.au/event/rettsyndromeresearch

CDKL5 Disorder Database

We are happy to announce that the new International CDKL5 Disorder Database was established in September 2012. All families with a child who has a mutation on the CDKL5 gene are invited to participate in the new database. Please visit cdkl5.childhealthresearch.org.au to register.

Australian Rett Syndrome Family Conference

In May this year over 160 family members, teachers, health professionals and care workers from around Australia met with researchers and clinicians at Riverglenn, Brisbane for the Australian Rett Syndrome Family Conference. Over two full days the audience learnt about the latest developments in genetics, scoliosis, epilepsy, bone health, communication, gastrointestinal health, mobility and recreation. A highlight of the conference was seeing all the wonderful interactions and the time spent affirming old friendships and making new ones. Many thanks go to the international and national speakers who gave of their time and expertise so generously, all the families who attended and made it such a success, and the Rett Syndrome Association of Australia for their support.

World Rett Syndrome Congress

The seventh World Rett Syndrome Congress took place in New Orleans, USA in June 2012. The congress comprised three symposia each with a central theme: basic research, translational and pre-clinical research, and family education and awareness. Five members of the Australian Rett syndrome team attended the congress and participated in one or more of the symposia. At the translational and pre-clinical symposium Dr Jenny Downs gave a presentation on how to measure physical activity in Rett syndrome. At the family conference, Dr Helen Leonard spoke on the importance of family and patient contribution to our collective understanding on Rett syndrome and Dr Jenny Downs coordinated a workshop on scoliosis. Alison Anderson promoted our international study InterRett. The congress also provided opportunities for PhD students Stephanie Fehr and Anna Urbanowicz: Stephanie presented the framework for the recently launched CDKL5 registry while Anna presented two posters on communication abilities, networked with experts in her field and was able to trial the latest communication technologies!

Communication Research

This year PhD student Anna Urbanowicz is investigating the communication abilities of girls and women with Rett syndrome. One of her studies will involve the development of a detailed video assessment tool for communication. In September this year, Anna participated in the 'Three Minute Thesis Competition.' The competition is run at various Australian universities and requires higher degree research students to captivate and educate an audience about their research in only three minutes! If you're interested in viewing the presentation about the communication abilities of girls and women with Rett syndrome please go to this link: www.youtube.com/watch?v=4G7Az71jsT8

If you would like any additional information about this research please contact Anna Urbanowicz on aurbanowicz@icmr.uwa.edu.au or (08) 9489 7786.

Celebrating 10 years of InterRett

Did you know that there is an International repository of information on over 2500 cases of Rett syndrome built with input from 610 clinicians and 1294 individual families from over 40 different countries...and it is right here! InterRett – the IRSF Rett phenotype database celebrated its 10 year anniversary this year and a flyer was produced to celebrate its achievements and highlights.

You can find a link to this flyer on the homepage of our website www.aussierett.org.au. You can also explore graphs generated from the InterRett database, which may also be of interest to you. From the homepage of our website www.aussierett.org.au, go to "Our Research" and then "Search Our Databases".





MECP2 duplication research

MECP2 duplication syndrome is a rare neurodevelopmental disorder that, unlike Rett syndrome, mostly affects boys. Australian families with children with *MECP2* duplication syndrome are raising awareness and much needed funding for research into treatment and a cure for this disorder. Part of these funds will go to a research project headed by Dr Helen Leonard at the Telethon Institute for Child Health Research.

Recently, the story of two Australian families and their sons was shown on the ABC program, the 7.30 Report in NSW.

The Van Wright Foundation was launched in March 2012 to raise awareness of and funds for research into *MECP2* duplication syndrome by the parents of Van Wright who was diagnosed in November 2011. Visit their website at www.vanwrightfoundation.org to learn more.

In the Media

In issue 46 of "That's Life" magazine, Perth mother, Teresa Pracilio, described the time around her daughter Kayla's (4) diagnosis and her search for answers about her daughter's condition. Accompanied by a beautiful photo of Kayla with her family, this story raised awareness about Rett syndrome.

Jovie Hoyng (4) and her family were featured in their local paper Rouse Hill Times on September 25 to raise awareness and funds for our Rett syndrome research. Jovie's family took part in the Rebel Sport Run4Fun on November 4 and encouraged the community to join or sponsor them in this community event.

Keep in touch with AussieRett CONNECT

Every AussieRett family helps to improve our knowledge and research into Rett syndrome. Please let us know if your contact details change. We don't want to lose you! If you would like to make a contribution to the next AussieRett CONNECT, please contact:

AussieRett

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2012 Publications

The following papers and book chapters written by members of the AussieRett team have been published or accepted for publication in 2012. You can read snapshots of these papers on our website under 'Our Research', or copies of full papers can be obtained by emailing the team at aussierett@ichr.uwa.edu.au.

1. Using a large international sample to investigate epilepsy in Rett syndrome. *Developmental Medicine and Child Neurology*. Forthcoming 2012.
2. Pubertal trajectory in females with Rett syndrome: a population-based study. *Brain and Development*. Forthcoming 2012.
3. The experiences of mothers of young adults with an intellectual disability transitioning from secondary school to adult life. *Journal of Intellectual & Developmental Disability*. Forthcoming 2012.
4. Caring for a child with severe intellectual disability in China using the example of Rett syndrome. *Disability and Rehabilitation*. Forthcoming 2012.
5. Longitudinal and population-based approaches to study the lifelong trajectories of children with neurodevelopmental conditions. In *Health Outcomes in Children, and Young Adults with Neurological and Developmental Conditions: Theory, Concepts, Evidence and Practice*. MacKeith Press. Forthcoming 2012.
6. The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy. *European Journal of Human Genetics*, 2012.
7. The phenotype associated with a large deletion on *MECP2*. *European Journal of Human Genetics*, 2012.
8. The conductive environment enhances gross motor function of girls with Rett syndrome. A pilot study. *Developmental NeuroRehabilitation*, 2012.
9. Initial assessment of the StepWatch Activity Monitor™ to measure walking activity in Rett syndrome. *Disability and Rehabilitation*, 2012.
10. The diagnostic odyssey to Rett syndrome: The experience of an Australian family. *American Journal of Medical Genetics Part A*, 2012.
11. Barriers to diagnosis of a rare neurological disorder in China – lived experiences of families affected by Rett syndrome. *American Journal of Medical Genetics Part A*, 2012.
12. What does the nature of the *MECP2* mutation tell us about parental origin and recurrence risk in Rett syndrome? *Clinical Genetics*, 2012.