

Merry Christmas and a Happy New Year from Helen and all the team at the Australian Rett Syndrome Study. We hope that this time of year brings you much joy, rest and relaxation as you spend time with family and friends.

This newsletter features the voices of several parents and beautiful photos of children with Rett syndrome. Trish Donnelly has written about her family's journey to making their goal of a Rett syndrome specific respite facility a reality. Emma Reilly has written about a photography exhibition featuring photos she took of her daughter Eva. Annabelle Canaider has written about her daughter Ruby's plans for the summer holidays. We also bring you an update on the journal article written by Marisa Knott of her family's experience of her daughter's diagnosis of Rett syndrome. These stories highlight the power of families as advocates and voices for their children and others with Rett syndrome.

We are delighted to feature your family stories and photographs so please contact the team if you would like to contribute to the next newsletter in 2012.

Ruby's summer Written by Annabelle Canaider

This summer during the school holidays Ruby will go SURFING again with The Disabled Surfers Association of Australia (www.disabledsurfers.org). Ruby will visit her Grandma lots and also go on the Liberty swing at our local park (Laguna Park). The Liberty swings are so much fun. They're always locked but we have our own key and can go whenever we want. We'll also go to Nerang Library and borrow switch operated toys from the Special Needs Library. For more lovely photos of Ruby, see "Ruby has Rett Syndrome" on YouTube.



This photo of Ruby with the super cute smile was taken by her Dad at his place. That's a smile just for Daddy.



Ruby with our closest family friends, Grace and her little brother Oscar. They have a trampoline and a pool AND a space which Ruby loves. We spend a lot of time at their place, especially over summer school holidays.

'The Rett Syndrome Respite Initiative'

Written by Trish Donnelly, Queensland

In June 2010, our family was at breaking point. Lily was suffering from a period of regression and was inconsolable, day and night. It was the last day of the school holidays, a pupil free day. The thought of facing another day of screaming and crying after another night of sleep deprivation was just too much to bear. At 7.30am I began calling every phone number on my list of respite services whose waiting lists we had been placed on. I could not contain my emotions and with every phone call cried as I explained our desperate need for respite. Every call I made was answered by an answering machine, but one.

Julie Johnson from MAMRE answered the phone and turned out to be my guardian angel, for just a little while. She consoled me and assured me that she could help even if just for a short time. She provided us with some emergency funding for in-home respite. This would allow us to employ a person of our choice and in her words, "give us some breathing space" until we were able to make arrangements for more formal, ongoing supports. Julie gave me hope and assured me that we were not alone. She committed to supporting us for a period of 8 weeks.

The in-home respite was helpful in allowing us to leave our daughter at home and in the care of someone we trusted whilst we left home to spend some quality time together and with our other children. However, what we really needed was to sleep, to just rest so that we could cope with all that Rett Syndrome would have in store for us for another week. This would mean out of home respite.

This period came with the sad realisation that we could not do this alone. As strong, as resilient and as good a parents that we were, we

felt that we had somehow failed in our role. This took a little working through. We came to understand that along with letting go comes the opportunity for Lily to have meaningful relationships with others. The first thing we did was to set about creating a 'Circle of Friends'. These are people, young and old in Lily's life who care about her, are interested in her well-being and with whom Lily has a personal, mutually satisfying relationship with. The second was to have Lily attend an out of home respite service where we hoped that not only would we receive some respite, but where Lily's needs would also be met.

We made contact with Disability Services Queensland with a view to gaining access to a centre based respite service they offered for children in Brisbane. The process took nearly 6 months and by December, 2010 Lily was to have her first practice stay of 2 hours. All went well and Lily seemed happy when we arrived to pick her up. The next stay was a full day and then finally came the practice over-nighter.

As I packed Lily's bag, I was overwhelmed with feelings of guilt, fear and trepidation. We didn't need the respite right this minute and yet, here I was packing her up to leave her with people that I didn't know, in a place that I didn't feel was entirely appropriate. I reminded myself I must continue on with the process as we would need it 'one' day. Should we withdraw her from the program now, we would face not having any out of home respite options available to us if we did hit another difficult patch.

We arrived at the respite house and I was shocked to find that Lily would be staying overnight with a number of very agile, very boisterous and mostly uncontrollable teenage boys.



This was one of a few circumstances which we were to discover upon our arrival that was not ideal for Lily or in line with our wishes for her specific needs to be met. However, again, we pushed on for fear of losing our place in the service. We gathered our strength and kissed our little girl goodbye. Both James and I cried all the way home. It was a torturous evening after which we decided that if we were going to use a respite service again, it would be one that we created and one that catered to the very special needs of our Rett girls.

Within three months, we had purchased a parcel of land. James' vision was to develop part of the land into a complex of 30 town homes with a percentage of the profits from the sale of these used to build a house for the Rett community.

Our dream was to establish a place where our girls could not only have sleep-over's so that their families could have some respite from their caring roles, but to also provide families with a sense of community, a meeting place and somewhere to get together for the purposes of information sharing, learning and the opportunity for altruism. We wanted our community to gain control of how the services provided to us by the government were carried out. We wanted to have a say and a presence in the day to day operations of that service.

After researching the net, I came to the realisation that what we were dreaming of was already being done in the state, in the country

New AussieRett Website Launch

The Australian Rett Syndrome Study team are excited to announce the launch of our new website www.aussierett.org.au which has a great deal of interesting information.

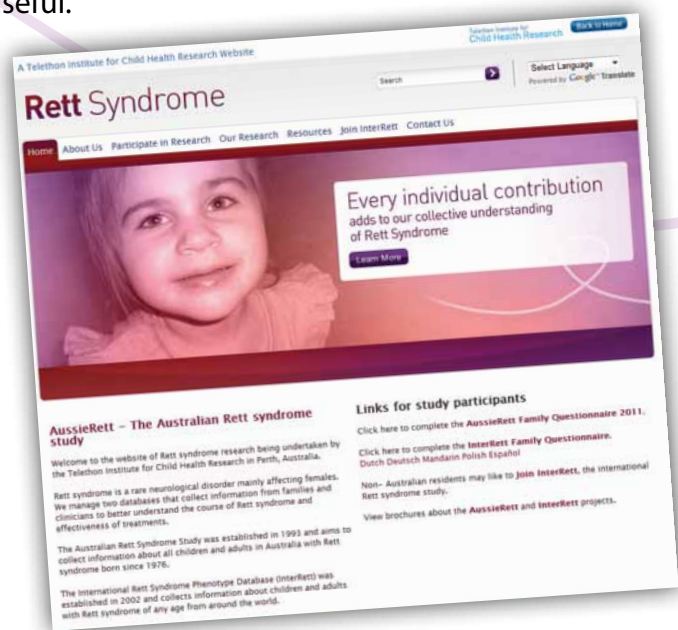
For families currently completing the 2011 Follow-Up Questionnaire online the link to the questionnaire can be found on the homepage under 'Links for Study Participants'. Please use the same username and password as is on your paper questionnaire. If you have any questions or problems with the online questionnaire please contact us.

As this is a new website we would very much appreciate feedback on the content or any other aspects of the website. Please email us at aussierett@ichr.uwa.edu.au if you have any comments or suggestions. We would also be happy to receive photos of your children or young adults with Rett syndrome to feature on the new website. We hope you find our new website informative and useful.

Published articles from our research

Below are the details of journal articles we have published since the last newsletter from the information you have generously given to us. If you would like to know more about any of these studies please contact Dr Helen Leonard on 0419 956 946. Snapshots and links to online abstracts will be available on the website soon.

1. Fehr S, Bebbington A, Nassar N, Downs J, Ronen G. M, De Klerk N, Leonard, H. Trends in the diagnosis of Rett syndrome in Australia. *Pediatric Research*. 2011; 70(3): 313-9.
2. Lim F, Downs J, Li J, Bao X-H, Leonard H. Barriers to diagnosis of a rare neurological disorder in China—Lived experiences of Rett syndrome families. *American Journal of Medical Genetics Part A*. 2011; online.
3. Knott M, Leonard H, Downs J. The diagnostic Odyssey to Rett syndrome: The experience of an Australian family. *American Journal of Medical Genetics Part A*. 2011; online.
4. Downs J, Leonard H, Hill K. Initial assessment of the StepWatch Activity Monitor™ to measure walking activity in Rett syndrome. *Disability and Rehabilitation*. 2011; online.
5. Bebbington A, Downs J, Percy A, Pineda M, Ben Zeev B, Bahi-Buisson N, Leonard H. The phenotype associated with a large deletion on MECP2. *European Journal of Human Genetics*. In press.



and the rest of the world. This concept is called a 'family governed co-operative' model in which families work together alongside government and a service provider to create a co-operative working relationship for the purposes of providing families with a service. The board consists of family members and of people whom the service is being provided to, to ensure that the philosophies of 'family governance' are upheld.

We met with representatives from the Department of Communities (Queensland) to present our idea. Our timing couldn't have been better. The department was moving in a new direction and was in the process of change and fortunately for us, our concept and dream was in-line with a number of their new initiatives. We left with a commitment that the department would support our idea.

Soon after, we were advised that

we had been granted on-going ministerial approved funding to make our dream a reality. The department would fully fund a community house which would also provide a five bed respite service. The initiative is called 'The Rett Syndrome Respite Initiative'.

We are currently awaiting development approval from the council for commencement of construction and hope to have the house completed by October, 2012.

Celebrating Genetic Diversity Exhibition

Written by Emma Reilly



Emma Reilly Photography

In October this year Eva's portrait (Called 'Eva Triptych') was included in an exhibition called "Celebrating Genetic Diversity" and was exhibited at the Powerhouse Museum in Brisbane, Queensland. The exhibition featured works by American photographer Rick Guidotti who founded Positive Exposure in 1988 as a non profit organisation, that challenges stigma associated with difference by pioneering a new vision of the beauty and richness of genetic diversity.

The exhibition also brought together the works of local photographers, each one featuring someone who was living with a genetic condition. The event

organiser, developmental paediatrician Dr Honey Heussler, said the concept was to celebrate diversity and highlight the impact of genetic syndromes and the need for more research into this complex area.

EDITORS NOTE: Attendees of The Society for the Study of Behavioural Phenotypes International Research Symposium and Education day (where Dr Helen Leonard provided an update to delegates on Rett syndrome entitled "The value of a population database in the understanding of a rare genetic disorder: insights from AussieRett") were fortunate to view the exhibition as part of their recent symposium.

Diagnosis of Rett syndrome – An Australian mother's experience

In our previous newsletter we wrote about a paper that had been accepted for publication on the diagnosis experience of fourteen families in China. The journal editor asked for an Australian story and Marisa Knott has kindly written the story of her family's journey to a diagnosis of Rett syndrome for their daughter Isabella. This will be published in the American Journal of Medical Genetics.

Thank you Marisa for sharing your personal story and for informing clinicians and health care professionals world wide about what this experience is like for families. Hopefully it will have positive impact on the diagnosis experience of families in the future. Snapshots of both articles will available on the AussieRett website in 2012.

Are you working on a questionnaire?

Thank you very much to all families and carers who have completed the follow-up questionnaire either in paper form, online or over the phone.

If you are yet to start working on your questionnaire or have partly completed it, we would be most grateful if you are able to find some time over the holidays to work on it.

If you have misplaced your questionnaire, need help with completing it online (including your username and password) or would prefer to give answers over the phone, please contact Katherine at aussierett@ichr.uwa.edu.au or Helen on 0419 956 946 and we will be happy to assist you.

Thank you very much.

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

PO Box 855

WEST PERTH, WA 6872

Email: aussierett@ichr.uwa.edu.au

Mobile: 0419 956 946

Phone: 08 9489 7774/7790