

# InterRett CONNECT

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December 2016

NEWSLETTER OF THE AUSTRALIAN RETT SYNDROME STUDY

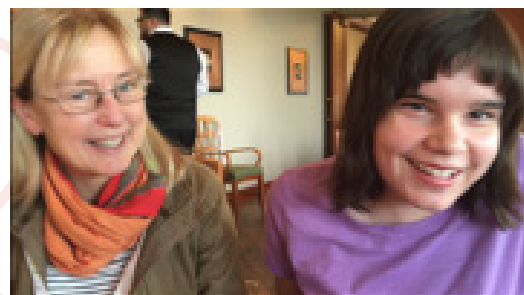


## Welcome

Welcome to our Christmas newsletter for 2016! Wishing you all Season's Greetings and a Happy New Year from Helen, Jenny, Amy, Barbara, Kingsley and the team at the InterRett. Have a safe, joyous, and relaxing festive season!

## Thank You to All!

A big thank you to all of the families of children and adults with Rett syndrome, for continuing to support our research by participating in the InterRett study. Thanks also to the members of our Parent Consumer Reference Group for your ongoing input and contribution to Rett syndrome research. We will continue meeting into the future so that our work is shaped by your ideas and experiences. If you are a parent who is interested in joining our teleconference meetings, please email us at: [rett@telethonkids.org.au](mailto:rett@telethonkids.org.au)



Jenny and Kylie

Helen and Jenny met some of you at the 32<sup>nd</sup> Family Education and Awareness Conference in Chicago in June. We work with families over long distances and it is wonderful when we meet face to face.

## Conference reports

Earlier this year, Helen attended the **8<sup>th</sup> World Rett Syndrome Congress in Kazan, Russia** where she gave a plenary presentation entitled "Rett syndrome journeys: What Russia and the World can learn from the experience of International and population-based registries of Rett syndrome". Helen also had the privilege of providing consultations to Russian families who had a child with Rett syndrome at a clinic held at the Children's Hospital in Kazan. This was a humbling experience and demonstrated how far advanced management of Rett syndrome in Australia is in comparison to other countries in the world.



Kazan



Panel Discussion at the IASSIDD (International Association for the Scientific Study of Intellectual and Developmental Disabilities) 15<sup>th</sup> World Congress in Melbourne in August

Later in the year, our team attended the **2016 IASSIDD (International Association for the Scientific Study of Intellectual and Developmental Disabilities) 15<sup>th</sup> World Congress in Melbourne** where we had the opportunity to present our research on quality of life and sleep problems. Helen's presentation focused on the need in Rett syndrome for an international register such as InterRett to complement the Australian population-based data and also the need for international registers in conditions like the CDKL5 Disorder and the MECP2 Duplication syndrome, which are even rarer than Rett syndrome. Helen also participated in a panel discussion regarding the genetic architecture of neurodevelopmental disabilities.

## Vienna

This year is the 50 year anniversary since Rett syndrome was first described in the German medical literature by Dr Andreas Rett who was a neurologist in Vienna. To celebrate, a group of Austrian clinicians and researchers organised an anniversary conference held in Vienna. The conference ran over 3 days and included many excellent presentations by clinicians, researchers and families, all dedicated to Rett syndrome. Many presentations had historical contexts and these were important because they illustrate why, relative to many other rare disorders, our understanding of Rett syndrome has made so much progress. The Rett syndrome research and clinical communities have been extremely well supported by

proactive family associations who have been truly visionary in their work and planning for a better future for Rett syndrome. Helen and Jenny attended and both presented - Helen on the value of databases and Jenny on gross motor skills and activity.



## RECENT RESEARCH

### Rett syndrome Fifty years on

Along with Dr Stuart Cobb from the University of Glasgow, Helen and Jenny were privileged to be invited to write a review for Nature Reviews Neurology to commemorate the 50 year anniversary of Andreas Rett's first description of Rett syndrome in 1966. We were able to describe the clinical and biological research, that has over the last half century contributed to the immense volume of knowledge about Rett syndrome we now have today. In particular we had the opportunity to map out the milestones which have occurred in the course of this journey. We could not forget to acknowledge the roles of family organisations such as Rettsyndrome.org, originally known as IRSA, and its founder Mrs Kathy Hunter. This is a very prestigious journal and we hope the paper will reinforce the very important research progress that has been made and importantly help to raise awareness.

### Bone Health

We recently developed clinical guidelines for managing bone health in order to help doctors and families reduce the risk of fractures in those with Rett syndrome. Increased physical activity, even for those unable to walk independently (where body weight supported treadmill or assisted walking is recommended) is very important. If calcium intake is sub-optimal, increased dietary calcium or administration of calcium supplements is recommended and similarly, if Vitamin D levels are low, Vitamin D supplements are recommended. We hope that families will share these publicly available guidelines with their daughter's general practitioner and specialists. <http://journals.plos.org/plosone/article?id=10.1371/journal.pone.0146824>

### Sleep Problems

Sleep problems can have many impacts on children and families. Families, mainly from our international database, InterRett, contributed to our investigation of sleep problems and their specific determinants. Sleep problems in Rett syndrome occurred much more frequently than in the general population. Night waking was the most common sleep problem with nearly half of the girls and women currently waking frequently at night. Getting to sleep and staying asleep was most challenging for younger children and those with a p.Arg294\* mutation. Severe seizure activity and being unable to walk were associated with a greater likelihood of daytime napping.

### Quality of life

Last year, we started a program of research about quality of life. Firstly, a number of Australian families participated in an interview study about their daughter's quality of life. From the very rich set of interview data, we found that the important areas of quality of life related to wellbeing, daily function and activities, and to community immersion. Two of the areas were new to the literature. The security of routines in day to day living and in helping the child to accept and derive pleasure from new experiences, and activities in the natural environment were very supportive of quality of life.

Quality of life is about the important things in life and understanding this is crucial for us to be able to measure and then plan how to support it. As the first step, the interview data enabled us to construct a new measure and we are currently "road-testing" it in a short online questionnaire. This will tell us if it does what we want and need it to do! This measure will allow much clearer identification of support needs and we look forward to putting it to use in a new family questionnaire for 2017.

# How do girls and women with Rett syndrome communicate and what factors influence successful communication?

This year, team member Anna Urbanowicz completed her PhD research on the topic of communication in Rett syndrome. Her research described the communication abilities of girls and women with Rett syndrome and investigated factors that were positively and negatively associated with communication outcomes.

## What was found?

- During interviews parents reported their daughters were able to express discomfort and pleasure, and make requests and choices using a variety of modalities including body movements and eye gaze. They also reported that mobility and the skills and knowledge of the communication partner influenced communication.
- Data provided by Australian and international families showed that the majority (89%) acquired speech-language abilities in the form of babble or words. Of those, most (85%) then experienced a regression in those abilities. Those with a p.Arg133Cys mutation were the most likely to use one of more words, prior to and after speech-language regression.
- Many females with Rett syndrome communicate using eye gaze or gestures. Using questionnaire data (n=151) from Australian families we found that school-aged children had the highest scores for eye gaze. Those with better gross motor abilities had higher scores for the use of both eye gaze and gestures. The use of eye gaze did not vary across mutation type, but those with a C-terminal deletion had the highest scores for use of gestures.
- Video footage provided in our larger Australian video study found most 82.8% of the girls and women made a choice, most using eye gaze. Of those who made a choice, half did so within 8 seconds.

## What does this mean for girls and women with Rett syndrome and their families?

- Girls and women with Rett syndrome share communication strengths including the use of eye gaze and the ability to make choices.
- Communication interventions should target communicative strengths, such as the use of eye gaze, and factors shown to impact communication, including the skills of communication partners.

Anna is now working with a group of international researchers and speech-language pathologists to develop clinical guidelines for the management of communication in individuals with Rett syndrome. Just like our previous guidelines for scoliosis, gastrointestinal disorders and bone health, the communication guidelines are being developed using a consensus approach with clinicians, researchers and families. The guidelines will ensure consistent information is provided to families around the world. The guidelines will be completed by the end of 2017.

For more information on this research please email Anna at: [a.urbanowicz@uq.edu.au](mailto:a.urbanowicz@uq.edu.au)

## NEWS

Our scoliosis and gastrointestinal guidelines booklets are available on our InterRett website and can be freely downloaded. You can access them at <http://interrett.org.au/resources/guidelines,-reports-and-books/>

Ms Michelle Stahlhut is a Danish physiotherapist who is completing her PhD looking at physical activity in girls and women with Rett syndrome. She is collaborating with Jenny and Helen and visited Perth this year – this was a great opportunity to work towards how we can ensure optimally active lifestyles in Rett syndrome.



Jenny and Michelle





Helen & Jenny presenting posters at the 2016 Rett syndrome Research Symposium and Family Conference, including one on physical activity. Please download our assessment for 'uptime' and share with your therapist.



[http://aussierett.org.au/media/1397275/Modified-Bouchard-activity-diary\\_Rett-syndrome\\_handout.pdf](http://aussierett.org.au/media/1397275/Modified-Bouchard-activity-diary_Rett-syndrome_handout.pdf)

## PUBLICATIONS IN 2016

Members of the InterRett team have published the following papers in 2016. Snapshots of these papers are available for you to read on our website under 'Our Research', or you can request full copies by emailing our team at: [rett@telethonkids.org.au](mailto:rett@telethonkids.org.au)

1. Leonard H, Cobb S, Downs J. Clinical and Biological Progress over 50 Years in Rett syndrome. *Nature Reviews Neurology*. 2016; doi:10.1038/nrneurol.2016.186
2. Urbanowicz A, Leonard H, Girdler S, Ciccone N, Downs J. Parental perspectives on the communication abilities of their daughters with Rett syndrome. *Developmental Neurorehabilitation*. 2016;19(1):17-25.
3. Urbanowicz A, Downs J, Girdler S, Ciccone N, Leonard H. An exploration of the use of eye gaze and gestures in females with Rett syndrome. *Journal of Speech, Language, and Hearing Research*. Forthcoming 2016.
4. Stahlhut M, Downs J, Leonard H, Bisgaard A M, Nordmark E. Building the repertoire of measures of walking in Rett syndrome. *Disability Rehabilitation*. Forthcoming 2016.
5. Horne T, Leonard H, Stannage K, Downs J. A qualitative investigation of recovery after femoral fracture in Rett syndrome. *Child: Care, Health and Development*. Forthcoming 2016.
6. Downs J, Torode I, Wong K, et al. The Natural History of Scoliosis in Females With Rett Syndrome. *Spine*. 2016;41(10):856-63.
7. Boban S, Wong K, Epstein A, Anderson B, Murphy N, Downs J, Leonard H. Determinants of sleep disturbances in Rett syndrome: Novel findings in relation to genotype. *American Journal of Medical Genetics Part A*. 2016;170(9):2292-300.
8. Downs J, Forbes D, Johnson M, Leonard H. The child with Rett syndrome: how can clinical ethics guide the management of the comorbidities in the child with Rett syndrome? *Journal of Paediatrics and Child Health*. 2016;52(8):809-13.
9. Jefferson A, Leonard H, Siafarikas A, Woodhead H, Fyfe S, et al. Clinical Guidelines for Management of Bone Health in Rett Syndrome Based on Expert Consensus and Available Evidence. *PLoS One*. 2016;11(2):e0146824.

- ★ For access to more of our research, please visit our website: [www.interrett.org.au](http://www.interrett.org.au)
- ★ Find summaries of our recent research publications at: [www.interrett.org.au/our-research](http://www.interrett.org.au/our-research)
- ★ Follow our team on Facebook: [www.facebook.com/interrett](https://www.facebook.com/interrett)

### Keep in touch with InterRett CONNECT

Every InterRett 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!

#### InterRett

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*We wish you a very happy and safe holiday period.*

