NEWS

The Rett Syndrome Association of Australia is a national not-for-profit group who, via a membership fee of $25 per family annually, were able to host and facilitate our very first National Rett Syndrome Conference. We hope to do it again. Contact Charlotte Fitzpatrick the RSAA Secretary on cdfitz73@gmail.com or 0427422788 or upload membership forms and lots of other information on our website www.rettaustralia.com. Watch this space for conference speaker summaries, photos and our video tribute.

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

Lily Klingner and Sarah Bromhead-Dunn happy to play at the RSAA Geelong conference

For access to more of our research, please visit our website: http://www.aussierett.org.au
Find summaries of our recent research publications at: http://aussierett.org.au/our-research/research-snapshots
Follow our team on Facebook: http://www.facebook.com/aussierett

We wish you a very happy and safe holiday period.

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:

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PUBLICATIONS IN 2015

The following papers written by members of the AussieRett team have been published or accepted for publication in 2015. 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@telethonkids.org.au.

1. Epstein A, Leonard H, Davis E, Williams K, Reddihough D, Murphy N, Whitehouse A, Downs J. Conceptualizing a quality of life framework for girls with Rett syndrome, American Journal of Medical Genetics Part A (in press).
2. Downs J, Torode I, Wong K, Ellaway C, Elliott EJ, Christodoulou J, Jacoby P, Thomson MR, Izatt MT, Askin GN, McPhee BI, Bridge C, Cundy P, Leonard H. Spinal fusion in early onset severe scoliosis improves survival in the child with Rett syndrome. Developmental Medicine and Child Neurology 2015 11 Dec | DOI: 10.1111/dmcn.12984.
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 (in press).
4. Lor L, Hill K, Jacoby P, Leonard H, Downs J. A validation study of the Bouchard activity record that extends the concept of uptime to Rett syndrome. Developmental Medicine and Child Neurology. [Epub ahead of print].
5. Marr C, Leonard H, Torode I, Downs J. Spinal fusion in girls with Rett syndrome: postoperative recovery and family experiences. Child: Health, Care and Development. 2015 Mar 9. doi: 10.1111/cch.12243. [Epub ahead of print]
6. Jefferson A, Fyfe S, Downs J, Woodhead H, Jacoby P, Leonard H. Longitudinal bone mineral content and density in Rett syndrome and their contributing factors. Bone. 2015;74:191-8. doi: 10.1016/j.bone.2015.01.023.
7. Downs J, Leonard H, Jacoby P, Brisco L, Baikie G, Hill K. Rett syndrome: establishing a novel outcome measure for walking in an era of clinical trials for rare disorders, Disability and Rehabilitation 2015;37(21):1992-1996. doi: 10.3109/09638288.2014.993436
8. Downs J, Wong K, Ravikumara M, Ellaway C, Elliott E, Christodoulou J, Jacoby P, Leonard H. Experience of gastrostomy using a quality care framework: the example of Rett syndrome, Medicine 2014 Dec;93(28):e328. doi: 10.1097/MD.0000000000000328.
9. Urbanowicz A, Downs J, Girdler S, Ciccone N, Leonard H. Aspects of speech-language abilities are influenced by MECP2 mutation type in girls with Rett syndrome, American Journal of Medical Genetics Part A 2015;167(2):354-62. doi: 10.1002/ajmg.a.36871.
10. Wong K, Leonard H, Jacoby P, Ellaway C, Downs J. The trajectories of sleep disturbances in Rett syndrome. Journal of Sleep Research. 2015;24(2):223-33. doi: 10.1111/jsr.12240.

AussieRett CONNECT

TELETHON KIDS INSTITUTE

www.aussierett.org.au

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NEWSLETTER OF THE AUSTRALIAN RETT SYNDROME STUDY

WELCOME

We would like to wish you all Seasons Greetings and a Happy New Year from Helen, Jenny, Amy and the team at the Australian Rett Syndrome Study. Wishing you and your loved ones a safe, joyous, and relaxing festive season.

A Big Thank You!

Thank you to the families of children and adults with Rett syndrome, for your ongoing support of our research through your participation and commitment to the AussieRett study. We would like to thank all of the members of the Parent Consumer Reference Group for their input and contribution to Rett syndrome research. We will continue meeting with this important group on a regular basis so that our work is informed by their experiences. If you are a parent who is interested in joining our meetings, then please email us at: aussierett@telethonkids.org.au

We have had some wonderful donations from the Rett syndrome community and beyond over 2015. Thank you to all, and a special thank you to the Diffey family and Clare's Angels for their amazing contribution from the New Year Ball in Dubbo, the Brown family, the Dirnbauer family for Run for the Kids, and the amazing HBF runners in Western Australia.

Family Conference, Geelong 19- 22 November, 2015

Written by Caroline Fitzpatrick, mum to Charlotte and Georgia, RSAA secretary

Pathways Journeys Rett Syndrome Geelong Novotel Families Professionals Siblings Grandparents Access Independence Trapped Hope Love Joy Peace Relief Eyes Surgery Breathing Eating Therapy Local National International Alternative Augmentative Communication AAC Pegs Toileting Eye Gaze Equipment Temperature Controlled Palliative Harpist Playschool Singalongs Kids Creche Tribute Bill Kathy Mice Trusts Scoliosis Neurology John Meir Liz Mary Sally Tracey Helen Jenny Yvonne Glen Allen NDIS Trusts Future Planning Caring Relationships Grief Loss Perspective. Mortality Diagnosis Butterflies X Chromosome Pooping Laughing Smiling Crying Hugging Anxiety Eye Pointing Fighting Spirit Strength Grit Dancing Drinking Eating Bonding Knowing Feeling Community Heard Forever Cure

A brainstorm to try and pinpoint the amazing two and a half day conference that the committee and community I'm part of hosted almost a month ago. Obviously hard and difficult to isolate the most amazing experience heard, felt and learnt during my stint. Though nowhere near as difficult as the drive and determination and patience to speak and operate your mind with a body that refuses to follow your lead. A bitter sweet part of our time together and as Kathy Hunter put it, not a group you would have on your bucket list to join, but a group pulling us all together with a very common thread. Despite knowing our girls have a diagnosis of Rett Syndrome, it's about what we as parents, researchers, siblings, experts and therapists can do about it, and if we are doing it, can we do it better? And can we keep doing it better and keep evolving our attitudes and behaviours to match our evolving child/sibling/grandchild/client with a Rett Syndrome diagnosis so they can choose and live and lead a life they desire?

Helen and Jenny with Jayne Bowden in Geelong

First Australian National Rett Conference
The Rett Syndrome Journey; Pathways to Follow
November 19th—21st 2015
Novotel Geelong Victoria

Rett Syndrome Association of Australia

Other conference reports

In May of this year, Helen attended the **2015 IASSIDD Americas Regional Congress in Honolulu** where she had the opportunity to present on our quality of life and scoliosis studies. One particular highlight was the opportunity to meet with Michelle Lam and her daughter Brittany, both of whom she had met eleven years previously at a Canadian Rett syndrome conference. Michelle and Brittany extended wonderful hospitality which was a highlight of Helen's visit to Hawaii.

Helen and Brittany in Honolulu

At the end of October Helen was invited to speak at the **4th European Congress held in Rome**. In order to cater for families, therapists, educators and teachers as well as researchers and clinicians there were parallel streams running for most of the congress.

- The basic science stream included presentations from several top scientists from the US, Italy and Scotland. Topics included the repression of long genes by MECP2, the role of glial cells in Rett syndrome, the potential for gene therapy and for reactivation of the X chromosome.
- In the clinical stream, Helen talked about how our knowledge about Rett syndrome has changed over time and what we know about adulthood in Rett syndrome. Her talk was followed by presentations on transition, sleep and epilepsy. On the second day, a number of talks were given on breathing, heart and nutritional management in Rett syndrome. The Italian specialists frequently sourced their information from the clinical research papers published by our group.
- There were also sessions on Neuromotor Rehabilitation (Dr Meir Lotan from Israel spoke), Cognition and Communication, and Music Therapy. Helen gave a presentation highlighting Jenny's work developing new measures to assess gross motor function and evaluate walking activity.
- The final day included an update on the clinical trials currently in progress, where Dr Daniel Glaze spoke optimistically about the trial of NNZ-2566 (the drug now known as trofinetide), the next phase of which will again take place in the US. The results of some other trials were less positive. However, preliminary results from a French study investigating the use of bisphosphonate on bone density were promising.

The Congress audience was a mix of families, clinicians and researchers including laboratory based scientists. The families were predominantly Italian, and English/Italian translation was available for all sessions. There were also families from France, Belgium, the Netherlands and several Eastern European countries. We made available our guideline booklets and these disappeared like hotcakes! It was very gratifying for Helen to hear about the important role our Australian research outputs and publications are playing in Eastern European countries where there is little Rett syndrome expertise available.

Helen in Rome

Shenzhen, China Update

Jenny and Helen received funding in 2015 from Rettsyndrome.org to conduct a study that assesses early intervention for girls younger than six years old. There have been several studies that have shown that mice with a MECP2 mutation develop better motor skills if their environment is enriched, possibly due to the extra physical activity which then increases production of Brain Derived Neurotrophic Factor (BDNF). BDNF is an important protein for nerve cell growth and maturity. We need to examine carefully the precise benefits of intensive early intervention for children with Rett syndrome. This study involves a collaboration between Telethon Kids Institute and a team at the Rett Syndrome Comprehensive Research Institute in Shenzhen, China. We are just now starting to recruit for this study and we very much look forward to sharing our findings in our next newsletter.

RECENT RESEARCH

Uptime

Uptime is the time that a person spends weight bearing in either standing and walking. The duration of uptime is associated with health benefits – natural stretch to muscles and tendons, the maintenance or improvement in muscle strength or fitness, and different perspectives by being up and about. We have recently modified a diary card measure for physical activity to measure uptime in Rett syndrome.

We found that the amount of uptime indicated the number of steps taken, particularly for those who could walk on their own. Measuring uptime therefore provides insights into physical activity and sedentary behaviour. Using our measure could facilitate the planning of suitable day activities and programs in response to your daughter's goals. We have made an assessment form that is available for you to download from our website at http://rett.telethonkids.org.au/media/1397275/Modified-Bouchard-activity-diary_Rett-syndrome_handout.pdf . Please share with your therapists and teachers who may want to measure your daughter's uptime and help to structure her day to increase her uptime.

Bone health research

Since we undertook our first Xray and questionnaire study in 1996, our research group has had an interest in bone health in Rett syndrome. Ours was the first study to demonstrate that girls and women with Rett syndrome were at increased risk of fracture and to start exploring what factors might influence this risk. Over a decade later and thanks to all the families filling out successive questionnaires, we had accumulated a bank of longitudinal data. This allowed us to look in much more detail at the influence of the type of genetic mutation, early motor development, the presence of epilepsy and the anti-epileptic medication regime on the risk of fractures. Simultaneously we also commenced a study where we measured bone density at two time points. This allowed us to look at factors including the individual's mobility status, which not only influenced fracture risk but that also impacted on bone density and its change over time in Rett syndrome.

What we have done over the past two years and as part of Amanda Jefferson's PhD is bring together all of the evidence from our own research findings as well as that from other researchers around the world, to develop a set of guidelines for clinicians and families to follow to optimize bone health in Rett syndrome. This work has been done in collaboration with a distinguished international panel of specialists in Rett syndrome, endocrinology and other relevant disciplines who provided their input through a web-based system. We have now submitted these guidelines for publication and look forward to having them available to guide clinical practice and family decisions in relation to bone health.

Over the course of 2014 and 2015, Honours student Thomas Horne has completed a research project with us. Extending our work in relation to bone health, he conducted interviews with parents whose daughter had experienced a fracture of the femur during the past five years. Those who could walk independently regained this approximately six months after the fracture. The interviews also provided information on how a fracture was recognized, insights into the emotional journeys that the parent caregivers experienced and advice on how families in the future would best manage. Thomas' work is now being prepared for publication, which we hope will help clinicians to better understand the complex issues in relation to femoral fracture in Rett syndrome.

Scoliosis

By putting our database information together with hospital records, we have this year been able to conduct very detailed analyses about scoliosis. Our latest analysis shows that scoliosis occurs in approximately 75% of girls with Rett syndrome by the age of 15 years and its occurrence is strongly influenced by genotype. If a scoliosis develops, it tends to increase with age. This is especially so in girls who are unable to walk and are at risk of developing a more severe scoliosis. Approximately two thirds with a severe scoliosis in Australia have had a spinal fusion, on average when they were 13 years old. Whilst parents had found the decision to consent to spinal surgery very stressful, having had a spinal fusion was associated with their daughter having better health and wellbeing. For general information about scoliosis in Rett syndrome, our guidelines are available at http://interrett.org.au/resources/guidelines,-reports-and-books/.