



In the Media

A story on Rett Syndrome was included on the ABC 7.30 report in Western Australia on 31 May 2013. It featured the stories of two WA families and their personal experiences with Rett syndrome. Helen and Jenny were also interviewed. This piece helped to raise awareness in the community. Here is the link.

<http://www.abc.net.au/news/2013-05-31/fighting-for-their-childs-future/4727062>

2013 Publications

The following papers and book chapters written by members of the AussieRett team have been published or accepted for publication in 2013. 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@icmr.uwa.edu.au.

1. Andrews, J., Leonard, H., Hammond, G. C., Girdler, S., Rajapaksa, R., Bathgate, K., et al. Forthcoming 2013. Community participation for girls and women living with rett syndrome. Disability and Rehabilitation.
2. Baikie, G., Ravikumara, M., Downs, J., Naseem, N., Wong, K., Percy, A., et al. Forthcoming 2013. Guidance in the management of gastroesophageal reflux disease, constipation and abdominal bloating in rett syndrome. Journal of Pediatric Gastroenterology and Nutrition.
3. Bao, X., Downs, J., Wong, K., Williams, S. & Leonard, H. 2013. Using a large international sample to investigate epilepsy in rett syndrome. Developmental Medicine and Child Neurology, 55, 553-8.
4. Downs, J., Parkinson, S., Ranelli, S., Leonard, H., Diener, P. & Lotan, M. Forthcoming 2013. Perspectives on hand function in girls and women with rett syndrome. Dev Neurorehabil.
5. Downs J, Leonard H. Longitudinal and population-based approaches to study the lifelong trajectories of children with neurodevelopmental conditions. IN Ronen GM, Rosenbaum PL, editors. Life Quality Outcomes in Children and Young People with Neurological and Developmental Conditions: Concepts, Evidence and Practise. London: MacKeith Press: 2013. p.329-43.
6. Freilinger, M., Bohm, M., Lanator, I., Vergesslich-Rothschild, K., Huber, W., Anderson, A., et al. Forthcoming 2013. Prevalence, clinical investigation and management of gallbladder disease in rett syndrome. Developmental Medicine and Child Neurology.
7. Knight, O., Bebbington, A., Siafarikas, A., Woodhead, H., Girdler, S. & Leonard, H. 2013. Pubertal trajectory in females with rett syndrome: A population-based study. Brain and Development, 35, 912-20.
8. Lee, J., Leonard, H., Piek, J. & Downs, J. 2013. Early development and regression in rett syndrome. Clinical Genetics, 84, 527-576.
9. Leonard, H., Anderson, A., Bebbington, A., Bao, X., Wong, K., Williams, S., et al. 2013. Resourceful and creative methods are necessary to research rare disorders. Developmental Medicine and Child Neurology, 55, 870-1.
10. Leonard, H., Ravikumar, M., Baikie, G., Naseem, N., Ellaway, C., Percy, A., et al. 2013. Assessment and management of nutrition and growth in rett syndrome. Journal of Pediatric Gastroenterology and Nutrition, 57, 451-460.
11. Lim, F., Downs, J., Li, J., Bao, X.-H. & Leonard, H. 2013. Caring for a child with severe intellectual disability in china: The example of rett syndrome. Disability and Rehabilitation, 35, 343-351.
12. Walker, E. Forthcoming 2013. Community participation: Conversations with parent-carers of young women with rett syndrome. Journal of Intellectual & Developmental Disability



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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AussieRett CONNECT



www.aussierett.org.au

December 2013 - ISSUE SIXTEEN

NEWSLETTER OF THE AUSTRALIAN RETT SYNDROME STUDY

AussieRett Christmas Newsletter 2013

Welcome

Merry Christmas and a Happy New Year from Helen, Jenny and all the team at the Australian Rett syndrome study. We hope that this time of year brings joy, rest and relaxation as you spend time with family and friends. This newsletter features a short report on the recent European Rett Syndrome Conference in Maastricht and the development of our clinical guidelines for gastrointestinal issues in Rett syndrome.

Thank you

There are many amazing people who have made our work at the Australian Rett Syndrome Study both possible and enjoyable over the course of 2013. Firstly we would like to give you, the families and children and adults with Rett syndrome, our heartfelt thanks for your continued support of our research through your ongoing participation. We thank the members of the Consumer Reference Group for their important contribution to Rett syndrome research. Thank you also to all the clinicians who support our work. Finally we would like to thank and acknowledge students who have worked with us in the area of Rett syndrome - Anna Urbanowicz is continuing with her PhD studies on communication and Caitlin Marr has just finished an Honours project on recovery after spinal fusion. Thank you again!

Family Stories

Meet Ruby Balkwell who has just turned 10 years of age. Participation in physical activities in the community is good for health and wellbeing which Ruby knows. This summer she will be spending a lot of time swimming and in her favourite swing. Looking forward to the summer holidays!



Shenzhen, China

Helen and Jenny visited Shenzhen in the south of China twice during 2013 to talk with clinicians at the Shenzhen Children's Hospital and the families from the Shenzhen Rett Syndrome Family Association. This was a wonderful opportunity to make links with those affected by Rett syndrome in our region. They are also keen to collaborate in a small study in which we will assess the effects of early intervention on four young girls with Rett syndrome. We are hoping that the pilot study will begin in early 2014.



GI GUIDELINES AND BOOKLET

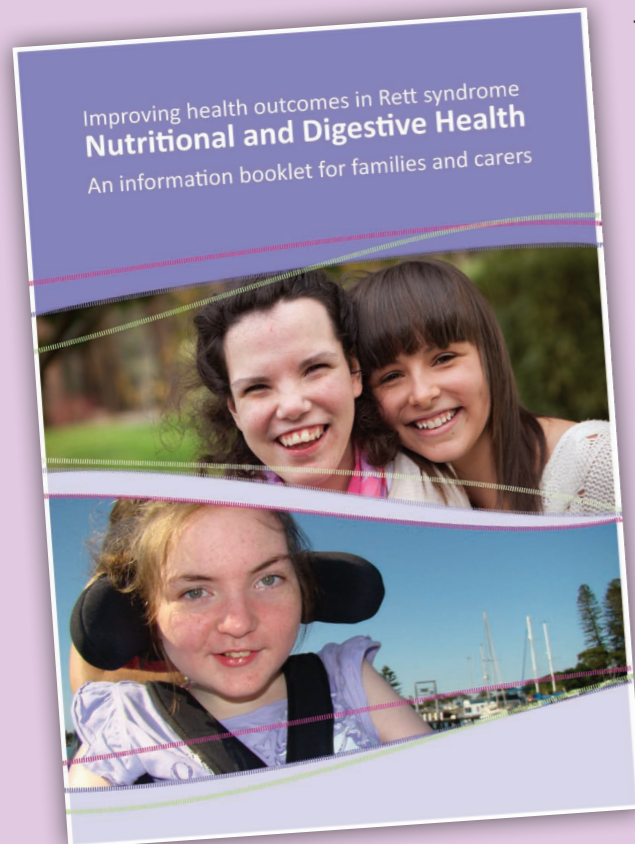
We have now developed clinical guidelines for the management of gastrointestinal disorders in Rett syndrome. We investigated the areas of growth and use of gastrostomy, reflux, constipation, abdominal bloating and gall bladder disease. To do this, we reviewed the research literature, and then consulted with a panel of international experts from the disciplines of gastroenterology, child neurology, developmental paediatrics, clinical genetics, dietetics and speech therapy. The panel provided feedback as to how nutrition and digestive health should be assessed and improved in girls and women with Rett syndrome.

The gastrointestinal guidelines have been published as three papers in the scientific literature. These papers will be practical resources for doctors and allied health professionals who may have limited experience with Rett syndrome. Details of these papers are listed in the section "2013 PUBLICATIONS"

The family and carer booklet enclosed is a ready reference on the best ways of improving the nutritional and digestive health of girls and women with Rett syndrome. Inside the booklet are two summary leaflets which you could give to your clinician if you want.

For the booklet, we also asked families and carers to share their personal stories of how they had dealt with any nutritional issues, feeding difficulties and/or gastrointestinal problems affecting their daughter. Together with the beautiful photos, we hope that the booklet will be a resource for you in everyday life and provide knowledge to assist in dealings with health and medical practitioners.

Electronic copies of the gastrointestinal booklet and the two leaflets can be found at <http://rett.childhealthresearch.org.au/resources/guidelines,-reports-and-books.aspx>.



3rd European Rett Syndrome Conference, October 17-19, Maastricht, the Netherlands

Following last year's World Rett Syndrome Congress held in New Orleans, the 3rd European Rett Syndrome Conference was held in the Netherlands over 3 days. Dr Helen Leonard and Dr Jenny Downs were both keynote speakers. Dr Alison Anderson and PhD students Anna Urbanowicz and Stephanie Fehr also presented.

The conference was mainly about Rett syndrome, but also included presentations on the CDKL5 and the MECP2 Duplication disorders. The conference was wide ranging in its topics but there was a focus on communication, autonomic problems and ageing. Here are just some of the highlights:

In Rett syndrome, we hear a great deal about the *MECP2* gene and its influences. *Dr Willem Voncken* from the Netherlands discussed epigenetics which is the study of how environmental factors can influence how a gene works. For example, both queen bees and worker bees have the same DNA but they develop and function differently according to whether or not they eat royal jelly. The specific structure of our genes is important but there is growing awareness of how other factors in the cells or in the wider environment can influence the work of a gene. Research in this area could be helpful for Rett syndrome.

Ingegerd Witt Engerstrom from Sweden gave an overview of points to consider during clinical management. She discussed how the brainstem had important influences on breathing difficulties and coordination of movement. One of her recommendations was to include *regular and joyful activities* within daily routines.

Monica Coenraads from the Rett Syndrome Research Trust, USA described how we were living in a golden era for understanding rare disorders and that our growing biological understanding is opening doors for treatments in the future. She gave an overview of their very comprehensive research program. The five broad themes of the types of treatments being investigated included:

1. methods of improving the proportion of active *MECP2* that do not have the mutation (reducing unfavourable X inactivation);
2. working on gene therapies where healthy genes are delivered via vectors such as viruses;
3. boosting MeCP2 levels - testing drug compounds that might for some mutations change the folding of the proteins in the cells and make the MeCP2 function better;
4. testing drugs that might influence other genes and result in better function. A clinical trial is being planned to test statins in improving function in Rett syndrome; and
5. testing drugs that can impact the proteins that *MECP2* would be influencing.

It was encouraging to hear of the systematic research that is investigating these potential treatments. However, it is important to remember that the biological pathways are very complex, there is still much work to be done, and researchers need to be very sure that any new treatments are safe and do not cause and do not cause new problems.

Professor Walter Kaufmann from Boston Children's Hospital gave an update of the progress of the IGF2 clinical trial. The phase 1 clinical trial has been completed with 12 girls participating and the drug was thought to be safe and well tolerated. They are now in the early stages of a phase 2 clinical trial and are in the process of enrolling patients.