

New Student Research Projects

Two new student research projects are under way this year. Occupational Therapist and PhD student Anna Urbanowicz aims to describe how girls and women with Rett syndrome communicate during daily activities and the factors that influence their ability to do so. Initially, existing questionnaire and video data in the Australian Rett Syndrome Database and additional information collected from interviews with mothers will be used. As part of her study Anna plans to develop a better way of measuring communication ability in Rett syndrome.

Joanne Lee's Masters of Clinical Psychology project will be a more detailed description of early development and regression for girls with Rett syndrome. Joanne will be interviewing mothers of young girls in both the AussieRett and InterRett studies to find more about this period of time in their daughter's lives.

This poem was written by a mother Anna Kohlbach from the perspective of her beautiful daughter Samantha who has Rett syndrome. Thank you Anna for your lovely poem which highlights Sam's courage and strength, attributes many parents can relate to in their own daughters.

I AM SAM

*I am Sam, I can hear, See me
Courage, A quiet courage*

*My Parents say
I like to hear "Clever girl"
Makes me smile
It's all worth while*

*I am Sam, I can hear, See me
Courage, A quiet courage*

*Those small things
You do each day
On your way
Without a thought
Easily taught*

*Years I worked
To raise a cup
To take a drink
On my own
With MY hand*

*Years I worked
To take a fork
From plate to mouth
What a task
What an ask*

*Clever girl
I hear them say
It makes me smile
Oh yeah it's worth while*

*I am Sam, I can hear, See me
Courage, A quiet courage*

*Oh no a step
What's the rules
Stop and think
Hold on tight
I lift my leg
I lean, I step
Where's the top
Does it stop
If the world was flat
There'd be none of that*

*There's so many more
Things you do
Without a thought
Easily taught
I'm yet to learn*

*Take a shower
Relax an hour
Take a walk
On my own
Have a talk
On the phone*

*So much to learn
Years it takes
Just for one
I'll keep working
With quiet courage
On and on
Till I'm done*

*I'll make my mark
On your hearts
I am Sam, Sam I am
With quiet courage
I SEE YOU
I thank you*

By Anna Kohlbach

Keep in touch with AussieRett CONNECT

If you would like to make a contribution to the next AussieRett CONNECT or if you have any questions or comments about this latest edition, please contact:

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Every AussieRett family helps to improve our knowledge and research into Rett syndrome. Please let us know if your contact details change at any time or if you have a new email address. We don't want to lose you!

AussieRett CONNECT

NEWSLETTER OF THE AUSTRALIAN RETT SYNDROME STUDY

September 2011 - ISSUE THIRTEEN



It is hard to believe we have passed the halfway mark of 2011 already. The year is flying by! In our last newsletter we wrote about the National Health and Medical Research grant that is allowing us to continue our research in Rett syndrome. In particular we will be looking at factors affecting the development of spinal curvature and the outcomes for girls and their families following scoliosis and gastrostomy surgery. Many of you have been invited to take part either observing the alignment of your daughter's back or giving us information over the phone about your daughter's scoliosis diagnosis and treatments. Thank you very much for this important information. As the study progresses over the next three years we will be informing you where we are up to and what we are finding.

Background to AussieRett

We have all recently been part of the latest census, where the Australian Government asked for information about ourselves to form a picture of current and developing needs of Australians and to help plan for the future. One of our goals in AussieRett is similar in that we want to analyse the current picture and identify needs for all girls with Rett syndrome and their families in Australia. In order to meet this goal, we need the very valuable and important information you give us about the medical, therapy, transition, behaviour and communication needs of your daughter and yourselves. We are then able to answer questions and communicate the findings to your clinicians, specialists and other researchers globally through publishing in academic journals and presenting at conferences and seminars. Our work also involves collaborations with researchers in Australia and overseas to better understand the genetics of Rett syndrome to contribute to finding a cure. We believe it is extremely important to share our findings with you, through this newsletter, booklets (e.g. Scoliosis Guidelines booklet) and through speaking with you informally or at organised events.

Our next questionnaire

In the next few weeks you will be receiving in the mail the 2011 Follow-Up questionnaire which you can either complete with pen and paper, online, or by telephone. If you are new to AussieRett, we send out questionnaires to families of girls with Rett syndrome approximately every two years.

This is an internationally unique study of girls and their families as it is longitudinal, that is; by collecting information over time (since 1996), we are assembling a very special picture of the development and needs of your daughters as they grow and mature which is not collected in any other population with Rett syndrome anywhere else in the world.

Like a plasma screen television with more 'pixels' in the picture, we have a stronger research voice, families and the health and medical community are better informed about Rett syndrome, and clinicians can provide a better service.

If you have completed one of these questionnaires before, thank you, we greatly appreciate all your valuable

information. We understand that filling in questionnaires takes time, so

we have some ideas to help make it easier for you.

- If you have a computer with Internet access you may wish to complete your survey online. You will be able to save sections and return to it so you don't need to complete it all at once.



- If you would find it easier to talk through the answers one of the AussieRett team members can help you complete the questionnaire over the phone.
- You are also welcome to contact a member of the team via our email address or phone numbers (details on back page) if you are unsure of any question or need any help with completing the online or paper versions.

We thank you in anticipation of your continued support, without it, our research and the benefit it has for families would not be possible. If you have recently changed your postal address or know you are about to, please send us an email or call one of the team to let us know of your new address, thank you.

Would you like a copy of your paper or online questionnaire?

In the past, some parents have requested a copy of the questionnaire they completed online to pass onto medical specialists or to keep as a record of their daughter's health and development. If you would appreciate a copy of your completed questionnaire please contact Katherine on (08) 9489 7774 or aussierett@ichr.uwa.edu.au and Katherine will post it to you.

Diagnosis of Rett syndrome in China

One of our students, Janice Lim, together with Dr Jenny Downs, Jianghong Li (Curtin University and TICHR) and Xin-Hua Bao (Peking University First Hospital) and Dr Helen Leonard have recently had a paper titled 'Barriers to diagnosis of a rare neurological disorder in China – lived experiences of Rett syndrome families' accepted for publication. This study involved discussing the process of

diagnosis of Rett syndrome with 14 families in different regions of China. We found the families experienced considerable delays in diagnosis involving visits to multiple health professionals. There were frustrations with the health care system with parents encountering long waiting lists, high costs in having to travel to receive specialist care and short consultation times. Families were also concerned about the lack of information about Rett syndrome and its management available in China. There may be many parallels with experiences of families in developed countries when seeking a diagnosis for their daughter. This prompted the journal editor to ask us if an Australian family would be interested in writing an article relaying their journey.

Marissa Knott who talked on this topic at the Family Conference that we held

in Sydney in 2005 has very kindly agreed to do this. Thank you Marissa.

New book on Rett syndrome

Over the course of this year and next, international clinicians and researchers with expertise in Rett syndrome have been putting together a new book on Rett syndrome published by MacKeith Press. Dr Helen Leonard is coordinating the section on clinical care as well as contributing to a chapter that looks at the influences of the specific type of mutation on clinical presentation. Dr Jenny Downs is helping to write a chapter on orthopaedic issues and another on movement in Rett syndrome. We hope that this book will be an important resource for clinicians as well as families.



LEFT TO RIGHT: Steph Fehr, Ami Bebbington, Helen Leonard, Katherine Bathgate, Jenny Downs, Kingsley Wong and Janice Lim.

Team Update

The AussieRett team here at the Telethon Institute for Child Health Research is in the photo beside, some of whom you may have met or spoken to on the phone and some will be new faces. The newest member of the team, Katherine Bathgate has started as the Project Coordinator and is taking over many of the tasks previously performed by Carol Philippe who is retiring after nine years with the study. Katherine is a dietitian and has a particular interest in nutrition and disability, she also has a son with a disability so can empathise with the realities of life with a family member with a disability. Dr Helen Leonard is the medical director and one of the Chief Investigators in the team. A recent online search revealed Helen as one of the foremost researchers in Rett syndrome world wide. Dr Jenny Downs is a physiotherapist, and also a Chief Investigator in the team. Dr Kingsley Wong is a Research Officer working with Helen and Jenny on the NHRMC study. We also have two students with special projects – Anna Urbanowicz is doing a PhD study on communication and Joanne Lee is doing a Master of Clinical Psychology study investigating early development and the regression period.

Thank you Carol

Our wonderful research assistant Carol Phillipe is retiring in September after 9 years with the Rett syndrome study. Carol has done a remarkable job working with families and all members of the team and she will be missed. On behalf of all the families and the staff and students you have worked with and supported over the years, THANK YOU Carol, we wish you all the very best in your retirement.



to be involved in the planning please let Katherine know on aussierett@ichr.uwa.edu.au. We would particularly appreciate families from Brisbane who can assist with local knowledge being part of the Committee as well as families from other states.

Family Stories and Photos

Do you have any special stories or photos of your daughter you would like to share with other families? We would love to feature the special moments or even the little moments with your daughter that make you smile. Please feel welcome to email your story and a photograph to the Editor on aussierett@ichr.uwa.edu.au for inclusion in future newsletters.

Watch this Space!

The Telethon Institute for Child Health Research is working on a new website and we are working on a new website for AussieRett. The new website will feature information on our research written in non-scientific language, publications we have produced to download and other information for families and clinicians. Further details will be available in the next newsletter.

Published Articles from Our Research

Using the information you have generously given us, we have now published nearly one hundred articles on Rett syndrome over the last twelve years. These are all listed on our website. Below are included those which have been published so far this year. If you would like to know more about any of these studies please contact Dr Helen Leonard on 0419 956

946 or email us at aussierett@ichr.uwa.edu.au

1. Urbanowicz A, Downs J, Bebbington A, Jacoby P, Girdler S, Leonard H. Use of equipment and respite services and caregiver health among Australian families living with Rett syndrome. *Research in Autism Spectrum Disorders*. 2011; 5(2):722-32.

2. Young D, Bebbington A, de Klerk N, Bower C, Nagarajan L, Leonard H. The relationship between MECP2 mutation type and health status and service use trajectories over time in a Rett syndrome population. *Research in Autism Spectrum Disorders*. 2011; 5(1):442-9.

3. Jefferson AL, Woodhead HJ, Fyfe S, Briody J, Bebbington A, Strauss BJ, Jacoby P, Leonard, H. Bone mineral content and density in Rett syndrome and their contributing factors. *Pediatric Research*. 2011; 69(4):293-8.

4. Downs J, Bebbington A, Kaufmann WE, Leonard H. Longitudinal hand function in Rett syndrome. *Journal of Child Neurology*. 2011; 26(3):334-40.

5. Hendrie D, Bebbington A, Bower C, Leonard H. Measuring use and cost of health sector and related care in a population of girls and young women with Rett syndrome. *Research in Autism Spectrum Disorders*. 2011; 5:901-9.

6. Fehr, S, Bebbington, A, Ellaway C, Rowe P, Leonard H, Downs J. Altered attainment of developmental milestones influences the age of diagnosis of Rett syndrome. *Journal of Child Neurology*. 2011; 26(8):980-7.

7. Foley KR, Downs J, Bebbington A, Jacoby P, Girdler S, Kaufmann WE, Leonard H. Changes in gross motor abilities of girls and women with Rett syndrome over a 3-to 4-year period. *Journal of Child Neurology* (in press).

Rett syndrome in the News

On Wednesday 10th July the SBS World News ran a story about the National Disability Insurance Scheme and Katrina Clark and her daughter Georgia were featured. Thank you Katrina and Georgia for highlighting the need for this scheme for all people with a disability, their families and carers.

Possible Family Conference Coming

We are exploring the possibility of a family conference next May in Brisbane. The timing and location has been chosen to coincide with the International Child Neurology Congress (May 27-June 1) as international experts will be presenting at and attending the congress and we will invite them to also share their work with families. We have discussed this idea with the Consumer Reference Group members who are supportive. If you think this is a good idea and would like