InterRett CONNECT

TELETHON KIDS INSTITUTE

www.interrett.org.au

December 2014

NEWSLETTER OF THE INTERRETT STUDY

InterRett Christmas Newsletter 2014

WELCOME

We would like to wish you a very Merry Christmas and a Happy New Year from Helen, Jenny, Nada, Amy, Orla, Nan and Kingsley, the team at the InterRett study. We would also like to wish you and your loved ones a safe, joyous and relaxing holiday season.

Helen and Jenny were privileged and honoured to attend the 13th Rett Syndrome Symposium and Family Conference in Washington DC. It was great meeting some of you on your own turf!

This newsletter provides some information about the progress of InterRett and describes some of our recent studies on spinal fusion, gallbladder function, gastrostomy feeding and communication in girls with Rett Syndrome.

THANK YOU

Again, we have many wonderful people to thank this year who have made the InterRett study possible. Thank you to the families of children and adults with Rett syndrome around the world for your continued interest and support of our research through your participation and commitment to the study. We would like to thank members of the Parent Consumer Reference Group for their previous and recent input to our work - we will continue meeting with this important group on a regular basis so that our work is informed by their experiences. If you would like to join, then please email us at Rett@telethonkids.org.au.

In particular, we would like to acknowledge Rettsyndrome.org for their ongoing funding of this key global research initiative. We also want to thank the wonderful families and supporters who donate to Rettsyndrome.org so that funds are available to support vital research such as ours. Thank you all so much. We are all the more determined to make a difference for those affected by Rett Syndrome.

Finally we would like to thank and acknowledge students who work with us. Anna Urbanowicz and Amanda Jefferson have been working on their PhD and hope to complete during 2015.

INTERRETT FOR 2014 AND BEYOND

We were very privileged to receive a grant from Rettsyndrome.org in 2014 that is enabling us to continue our work with InterRett. Our goals for the current grant will allow InterRett to make the next steps to better serve the Rett syndrome community. They are:

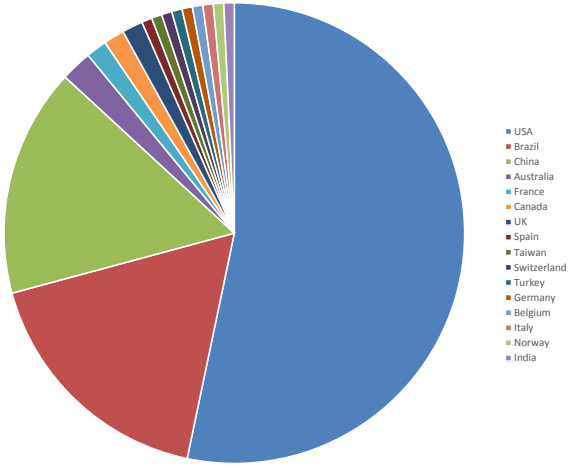
1. To give families a stronger voice in research about Rett syndrome;
2. To expand the data collection and facilitate evidence-based management. In the first instance we are planning new online questionnairs on:
    a. Sleep problems and their management
    b. Respiratory problems and their management;
3. To include focus on families who live in under-represented majority world countries; and
4. To work towards linking our data with that contained in other rare disorder registries.

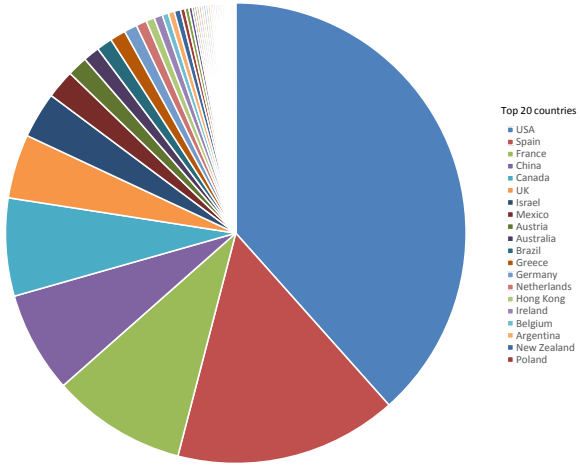
The InterRett Consumer Reference Group recently supported the need for the two new studies on sleep and respiratory problems in Rett syndrome. These are both aspects of health about which we know very little and are unclear as to the best management. Over the next few months, we will develop the questionnaire which can be completed online. We look to forward to working with you to investigate these important topics.

INTERRETT CONTINUES TO GROW

With the wonderful contribution of families and clinicians around the world InterRett has continued to grow providing an increasingly powerful database for Rett syndrome research. Overall, since the inception of InterRett, data has been provided on over 2500 girls and women with the largest proportions coming from the US, Spain, France, Canada, UK and Israel. During 2014, the greatest numbers of family participants have been from the US, Brazil and China. Thank you to everybody but especially to Silvana Santos who has put so much work into encouraging Brazilian families to participate and Shuang Liu (Sheila) from Shenzhen who has helped so much in encouraging Chinese families to participate. We very much value your time and effort.

Some countries are not visible on the graphs below because the numbers of participating families are very small but the countries are many. They include: Belarus, Bolivia, Bulgaria, Chile, Colombia, Costa Rica, Croatia, Cyprus, Denmark, Dominican Republic, Finland, Honduras, Hungary, India, Iran, Italy, Japan, Luxembourg, Macedonia, Malta, Norway, Peru, Portugal, Puerto Rico, Russia, Slovenia, South Africa, Sweden, Switzerland, Taiwan, The Netherlands, Turkey, United Arab Emirates, Uruguay and Venezuela. It is truly wonderful to capture experiences around the world and we value all participation.





The InterRett family questionnaire is currently available in online or paper versions in English, Spanish, German, Italian, Mandarin, Dutch, Polish, French and Portuguese. We would like to continue translations into other languages.

Search our database

Please visit the graph generating tool on our website where you create graphs based on the information provided in the InterRett project by families and clinicians around the world. We welcome your feedback. The link is: https://interrett.ichr.uwa.edu.au//output/

SOME AUSTRALIAN NEWS

Wendy Macklin is the grandmother of a beautiful three year old girl with Rett syndrome. Wendy wrote a wonderful piece called "A grandmother's story" describing her thoughts and perspectives in relation to her little granddaughter growing and developing but also being diagnosed with Rett syndrome. Her piece was published in the West Australian newspaper in March 2014. Here is the link:
http://aussierett.org.au/latest-news/

Jenny and Helen also wrote a piece in The Conversation giving an overview about scoliosis. Here is the link:
http://theconversation.com/explainer-everything-you-need-to-know-about-scoliosis-28409

DISORDERS RELATED TO RETT SYNDROME

International CDKL5 Disorder Database

The International CDKL5 Disorder Database is continuing to collect information. The aim of the new CDKL5 registry project is to collect information that is specific to this disorder from a larger number of families and their clinicians. This enhanced data repository will allow a more comprehensive profile of the clinical features that, in turn, will inform both clinical management and basic science research into cause and cure. All families with a child who has a mutation in the CDKL5 gene are invited to participate in the new database. Please visit http://cdkl5.childhealthresearch.org.au to register.

MECP2 duplication database

MECP2 duplication syndrome is a rare neurodevelopmental disorder that, in contrast to Rett syndrome, mostly affects boys. We also have plans in the months ahead to develop a separate MECP2 duplication registry database. In the meantime we welcome families with a child with the MECP2 duplication syndrome to also participate in InterRett. For families of those with MECP2 Duplication or Rett syndrome who have not yet participated in InterRett please go to: https://interrett.ichr.uwa.edu.au//irett_quest/registration/fam_registration.php.

QUALITY OF LIFE

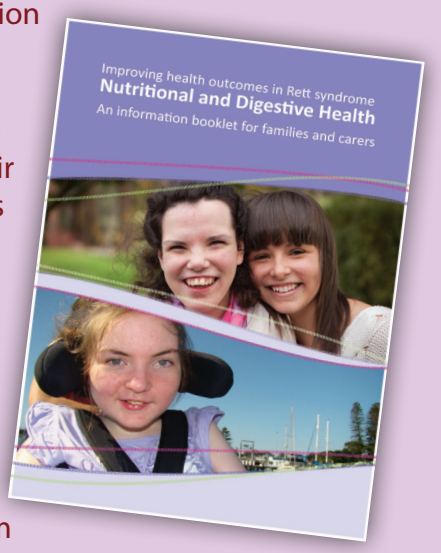
We have interviewd the families of twenty-two girls with Rett syndrome and asked themto talk about their daughter's life experiences (e.g. social inclusion) - to identify aspects of their lives that are most important. Parents shared many beautiful, deeply heartfelt stories of different life events that continue to help foster what they feel is a good quality of life for their daughter. These interviews are allowing us to develop a state-of-the-art measure of quality of life. This measure will help in identifying your daughter's needs and also the value of services that she receives. It will also be an important outcome measure for future clinical trials.

SCOLIOSIS

Some girls develop a severe scoliosis and spinal fusion may be recommended as management. This can be a difficult time for families when making the decision about whether to proceed with the surgery. In our 2011 questionnaire, we included questions on how families felt about the different aspects of the hospital management. The majority of families were very satisfied with their daughter's surgery and hospital care, especially when the staff were experienced and proactive and took family perspectives into account when making decisions. Many families were pleased with their daughter's straighter posture, her health and comfort, and greater ease of sitting, dressing and transfers. Most families said that they would consent to spinal fusion if faced with the same situation again.

GASTROSTOMY

Approximately one quarter of families in the Australian Rett syndrome population currently report using a gastrostomy. A gastrostomy can be used for all feeds, some feeds, just for medications or for venting excess air from the stomach. In our 2011 questionnaire, we included questions about families' satisfaction with gastrostomy surgery and the hospital care. Again, many families expressed their satisfaction with the gastrostomy procedure. They also reported improvements in their daughter's health and reduced family stress and burden during daily care activities.



The booklet on the management of gastrointestinal disorders is available on our website (http://aussierett.org.au/resources/guidelines,-reports-and-books/).

GALL BLADDER

We used information in our Australian database together with information from our international database InterRett to investigate gall bladder disorders in Rett syndrome. Whilst gall bladder problems are relatively rare, they occur more commonly in Rett syndrome than in girls of similar ages in the general population. This may be because the muscles in the gall bladder are weaker and there is less efficient emptying. Gallbladder disease should be considered as a possible cause of abdominal pain in Rett Syndrome.

Shenzhen, China

Laboratory studies have found that mice with a *MECP2* mutation develop better motor skills if their environment is enriched, possibly due to the physical activity which then causes the brain to produce Brain Derived Neurotrophic Factor (BDNF) proteins. BDNF is important for nerve cell growth and maturity. For Rett syndrome, therapies such as physiotherapy should be more effective when implemented intensively at an early age due to brain plasticity and also potentially due to more production of BDNF proteins. However, available research cannot tell us the precise benefits of intensive early interventions.

Jenny and Helen have developed relationships with families and clinicians in Shenzhen, China and worked with them earlier this year to run a pilot study testing an intensive therapy program. A pilot study is a practice study to learn how best to manage all the different aspects that need to be properly run in a larger study. Four young girls participated in the 7 week pilot program and each made some gains in relation to their motor abilities and their general wellbeing. This is encouraging and sets the scene for a larger study.

We wish you a very happy and safe holiday period.

Keep in touch with InterRett CONNECT

Do we have your latest email address and telephone number? If you changed your contact details, please let us know at rett@telethonkids.org.au.

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2014 Publications

The following papers written by members of the InterRett team have been published or accepted for publication in 2014. 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 rett@telethonkids.org.au.

1. 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 (in press).
2. Urbanowicz A, Downs J, Girdler S, Ciccone N, Leonard H. Language abilities of girls with Rett syndrome are influenced by MECP2 mutation type, American Journal of Medical Genetics Part A (in press).
3. Wong K, Leonard H, Jacoby P, Ellaway C, Downs J. The trajectories of sleep disturbances in Rett syndrome. Journal of Sleep Research. 2014, 14 Sept. [Epub ahead of print]
4. Anderson A, Wong K, Jacoby P, Downs J, Leonard H. Twenty years of surveillance in Rett syndrome: What does this tell us? Orphanet Journal of Rare Diseases. 2014;9:87.
5. Downs J, Torode I, Ellaway C, Jacoby P, Bunting C, Wong K, Christodoulou J, Leonard H. Family satisfaction following spinal fusion in Rett syndrome, Developmental Neurorehabilitation. 2014; April 11. [Epub ahead of print]
6. Urbanowicz A, Leonard H, Girdler S, Ciccione N, Downs J. The perspectives of mothers on the communication abilities of their daughter with Rett syndrome, Developmental Neurorehabilitation. 2014, 24 Feb. [Epub ahead of print]
7. Freilinger M, Bohm M, Lanator I, Vergesslich-Rothschild K, Huber W-D, Anderson A, Wong K, Baikie G, Ravikumara M, Downs J, Leonard H. Prevalence, clinical investigation and management of gallbladder disease in Rett syndrome, Developmental Medicine and Child Neurology. 2014;56(8):756-762. .
8. Baikie G, Madhur R, Downs J, Nasseem N, Wong K, Percy A, Lane J, Weiss B, Ellaway C, Bathgate K, Leonard H. Guidance in the management of gastroesophageal reflux, constipation and abdominal bloating in Rett syndrome, Journal of Pediatric Gastroenterology and Nutrition. 2014;58(2):244-51.
9. Andrews J, Leonard H, Hammond G, Girdler S, Rajapaksa R, Bathgate K, Downs J. Community participation for girls and women living with Rett syndrome. Disability and Rehabilitation. 2014;36(11):894-899.