Joyeux Noël! 圣诞节快乐! Feliz Navidad! Wesołych Świąt! Frohe Weihnachten!

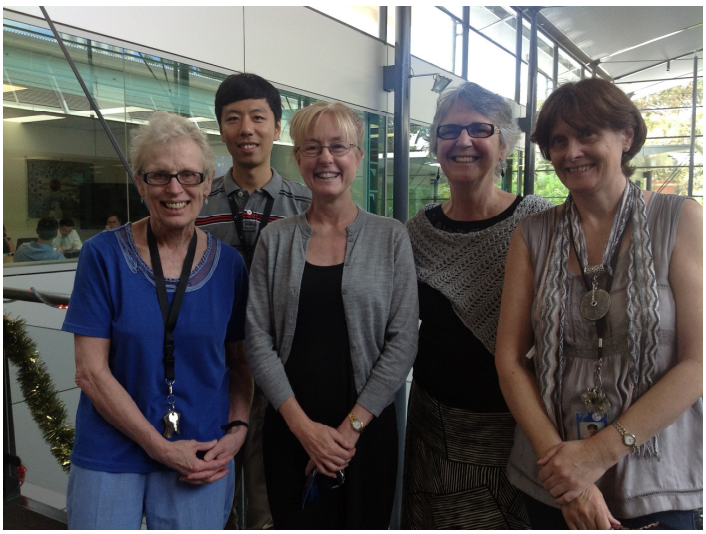
MERRY CHRISTMAS FROM INTERRETT!

A REPOSITORY OF INFORMATION ON OVER 2,500 CASES BUILT WITH INPUT FROM OVER 40 DIFFERENT COUNTRIES!!!

InterRett – 10 Year Celebration!!

This year the InterRett project, which is funded by the International Rett Syndrome Foundation (IRSF, previously IRSA), entered its 10th year. To celebrate we published a flyer outlining our many achievements. You can download a copy from our website: http://www.interrett.org.au/media/392270/interrett_10_year_flyer.pdf

Meet the Team



The InterRett project is managed by the Australian Rett syndrome study team. From left: Medical Director Dr Helen Leonard, Nan Hu, Dr Jenny Downs, Nada Murphy and Alison Anderson; Heidi Meyer and Ami Bebbington (in absentia).

PhD student Nan Hu is new to the team and is helping Chinese families to participate in InterRett. In 2013 Nan aims to provide these families with translations of recent research and the most up-to-date information on the management and care of individuals with Rett syndrome.

Plans for 2013

1. **An update of the current questionnaire:** The InterRett questionnaire was designed by a panel of clinicians in 2002. We would now like to update the questionnaire based on experience and knowledge gained over the last ten years.
2. **Age specific questionnaires**: As individuals who have Rett syndrome become older their needs change and the research questions that need to be answered also change. We therefore would like to develop a short questionnaire that is specific to the needs of older individuals.
3. **The collection of additional information from families who have already participated:** The InterRett project was originally designed to collect information at only one time point. Many of the early participants have indicated that they would be happy to provide updated information on their family member with Rett syndrome. The collection of information over time is very powerful for research and provides a more comprehensive picture of how symptoms change as individuals become older. In 2013 we aim to expand and modify the InterRett database to accommodate the collection of follow-up information from existing participants.

IRSF INTERNATIONAL RETT SYNDROME FOUNDATION

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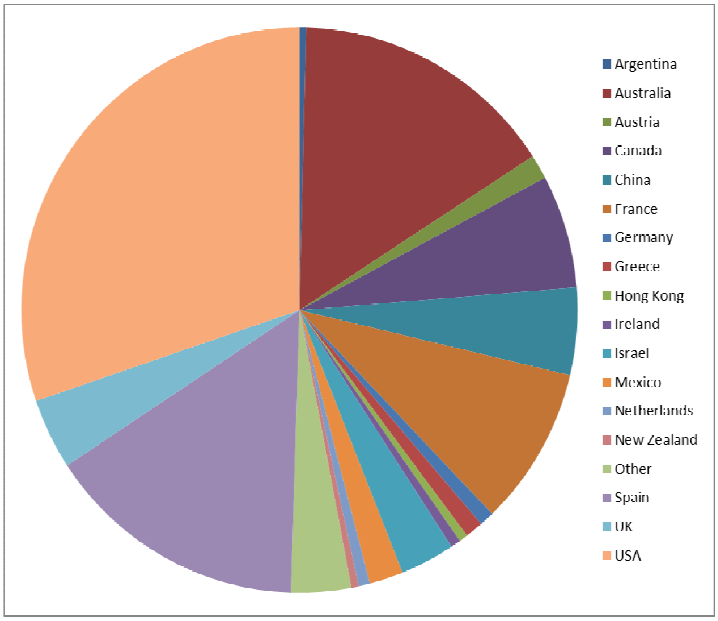
4. **The translation of information into knowledge – what would you like to know?** The InterRett database is a valuable resource that can be harnessed to address research questions relating to a wide range of topics. We continually look for ways in which this resource can be utilised. In 2013 we would like to invite you to have input into this process. Please email us at rett@ichr.uwa.edu.au and let us know what issues are of the upmost concern or interest to you.

InterRett continues to grow

With the wonderful support of families around the world the InterRett project database continues to grow providing an increasingly powerful database for Rett syndrome research. On behalf of the whole team we would like to ***thank you all*** for your valuable contribution to further research into Rett syndrome



The *Other* category in the graph on the left includes countries with less than 10 cases in the database: Belgium, Bolivia, Brazil, Bulgaria, Chile, Colombia, Costa Rica, Cyprus, Denmark, Finland, Honduras, Hungary, India, Iran, Italy, Japan, Luxemburg, Macedonia, Malta, The Netherlands, Norway, Peru, Poland, Portugal, Puerto Rica, Russia, Slovenia, South Africa, Sweden, Switzerland, Taiwan, Turkey, Ukraine, United Arab Emirates and Uruguay.

The InterRett family questionnaire is now available in online or paper version in English, Spanish, German, Italian, Mandarin, Dutch, Polish and French with a Hungarian version under development. This year the project welcomed our first participant from Russia. Families from Russia and the Ukraine consented to participate at a Rett family conference day held in Moscow on the 15th of September, 2012.

Scoliosis guidelines

Our booklet about the management of scoliosis in Rett syndrome continues to be a valuable resource for families. Copies can be downloaded from our website (http://www.interrett.org.au/resources/guidelines,-reports-and-books.aspx). The booklet is currently available in English, Spanish and Hungarian with a French version under final review. If you would like the booklet in any other language and can assist with the translation please contact Jenny Downs at jdowns@ichr.uwa.edu.au

Launch of International *CDKL5* Disorder Database

The *CDKL5* Disorder Database is up and running! This disorder is caused by mutations within the *CDKL5* gene. In 2012 we used InterRett data to compare the occurrence of Rett-like features in those with *MECP2* and *CDKL5* mutations. We found that most individuals with a *CDKL5* mutation had severe developmental delay from birth and seizure onset before the age of 3 months and some had similar facial features. A period of regression was less common in those with a *CDKL5* mutation compared to those with a *MECP2* mutation. The results of the study were published in the European Journal of Human Genetics. 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.

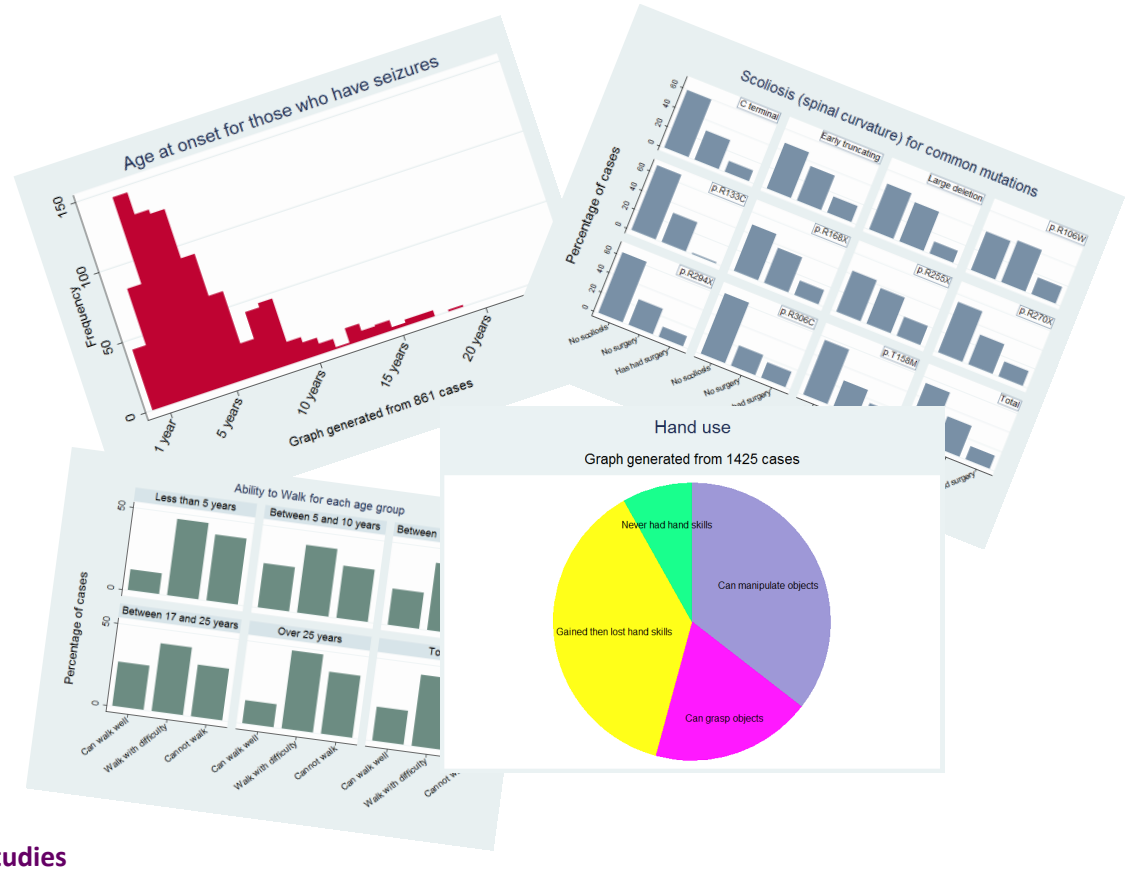
All families with a child who has a mutation in the *CDKL5* gene are invited to participate in the new database. Please visit http://cdkl5.childhealthresearch.org.au to register.

MECP2 duplication research

MECP2 duplication syndrome is a rare neurodevelopmental disorder that, in contrast to Rett syndrome, mostly affects boys. In Australia, the Van Wright Foundation was launched in March 2012 to raise awareness of and funds for research into *MECP2* duplication syndrome. A proportion of these funds will be earmarked for a research project headed by Dr Helen Leonard at the Australian Rett Syndrome study. Our aim is to develop a *MECP2* duplication registry based on the same successful framework that supports InterRett and the new *CDKL5* registry.

Search our database

In 2012 we updated the graph generating tool on our website. Please check it out (https://interrett.ichr.uwa.edu.au//output/) and give us your feedback. We aim to add more graphs during 2013.



Recent Studies

Update on the early development study

Since our 2011 gazette, psychologist Joanne Lee has completed interviews with mothers of 14 young girls with Rett syndrome. During the interviews, the mothers talked about their daughter's early developmental progress and how the signs of Rett syndrome developed. Some girls experienced a clear regression of speech and hand skills and for others, this was much more subtle. Most of the girls also experienced a period of social withdrawal and inconsolable crying, and the crying persisted for a much longer time period than did the social withdrawal. For some girls, balance was harder after regression which affected how the girls were able to sit, stand or walk. These discussions included a wealth of information about regression and highlighted how difficult a time this is for families. The narrative from this project will be very useful for clinicians to raise

awareness of Rett syndrome and for better understanding of how Rett syndrome evolves. Thank you again to those who participated.

Update on the 2011 Epilepsy study

Data from the InterRett database has shown that epilepsy is a common problem for girls and women with Rett syndrome. Epilepsy is particularly prevalent in those aged 7 to 17 years although seizure control in general is slightly better in adulthood. We have also shown that the occurrence of epilepsy may be associated with certain mutations on the *MECP2* gene. Of the common mutation types, girls and women with a p.T158M, p.R255X or a large deletion were more likely to develop epilepsy. For many girls and women, epilepsy was difficult to manage and approximately one third of those with epilepsy continued to have seizures despite taking two or more epilepsy medications. This information highlights the large burden of epilepsy for those with Rett syndrome and their families and also the importance of research into epilepsy treatment for those with persistent epilepsy.

For more research snapshots visit our website: http://interrett.org.au/our-research/research-snapshots.aspx

Merry Christmas & A Happy New Year From the InterRett Team!