Welcome to our Parents Panel

We want AussieRett Connect to be informative, interesting and valuable in supporting families who have children with Rett syndrome. We believe it is important that parents are given the opportunity to contribute their ideas to the newsletter. A new ‘Parents Panel’ has been formed for the newsletter. It will be so helpful to be able to consult them and make sure our newsletters are full of the ideas and information that you want to read.

Lisa Potter, Julie Trigg, Heather Jennings and Cathey Stanberg are four parents who have joined our panel. As Lisa commented in her first email to the editor – “There is so much to talk about with other families. Sometimes we live in such isolation with Rett syndrome. We are like sponges when it comes to reading and talking about our girls.” Bill Callaghan, a name many know from the Rett Syndrome Association of Australia will be on hand to share his years of valuable experience with us. But more than anything we welcome ideas from all families who have an interest in Rett syndrome.

A large number of videos have already come back from families across Australia. This is a fantastic achievement as we all appreciate how much hard work has gone in to getting them finished. However as parent, Julie Trigg told us, “It was worth the effort as it will be a great record for us as well as recording Louise’s functional abilities. We have kept a copy to show family and friends, as most of our family videos are of Louise relaxing, not doing her day to day activities.” It’s not too late to send your video back and do what Julie did, save a copy for yourselves. Many schools and relatives have been very supportive in helping families complete their videos. Thank you to everyone who has helped Rett syndrome research by contributing this valuable visual information to what we already know from your questionnaires. Please let us know if you are having any difficulty getting the video done and we will do all we can to help.

By the time you are reading this you will all have received the 2004 follow-up questionnaire in the mail. You will recognise many of the questions from the 2002 questionnaire. Our research has been the first to be able to show that children with Rett syndrome may be affected differently according to which genetic mutation they have. The data you are providing in 2004 will allow us to advance further with this very valuable work. You will also see why we are asking about ‘money, money, money’ if you read the article on Page 2.

If you have access to the Internet it is very easy to complete your questionnaire online. Instructions on how to access and complete the online questionnaire are included with every parent questionnaire. One parent observed recently, “I have found it much easier to fill in online. I find it much less distracting to sit at a keyboard and focus on the screen than to sit with a pen and paper and go through a booklet of questions.” It certainly makes life easier for us. It means our time and effort can be concentrated on the analysis of your data rather than first having to type all the information you send us into the database.
Many of you have heard the names of our research team based at the Telethon Institute for Child Health Research in Perth. You may have even spoken to them on the phone, but we would like to introduce them in a little more detail. This edition we focus on Crystal Laurvick.

Crystal has become a valuable member of the AussieRett team since she joined the project in January 2004 as the clinical coordinator of the study. Awarded a Master of Public Health Degree in 2001, her special interest in ovarian cancer has led to many papers being written and presentations being made at conferences both here in Australia and the USA. Travelling to the USA is not unusual for Crystal as she grew up in Minnesota. After gaining her degree in biology and psychology at Gustavus College in Minnesota, Crystal moved over to Australia in 1999 to continue her studies at the University of Western Australia in Perth.

Living in Western Australia suits this busy lady very well, so well in fact that she took out permanent residency here in 2002. Crystal adds, "I really enjoy the laid-back lifestyle in Australia, Perth in particular, and the friends I have made here have made me feel right at home."

Crystal is getting married in April 2006 and many of her family and friends are making the big trip from Minnesota to Perth for the special occasion. When she is not planning her wedding, she enjoys going for walks and bike rides in Kings Park with her fiancé, Aaron, and getting together with friends over dinner. "A hobby that I have really gotten into is stamping, where I make cards for all occasions using rubber stamps of various designs."

Her work on the Rett Syndrome Study is hectic but fascinating. She has been finalising the 2004 follow-up questionnaire; making DVDs of the videos families have sent us; and developing a database for collecting the clinical assessment information next year. "I really enjoy getting to use my creative side in designing the questionnaires and databases and giving everything that professional touch."

### Presenting Rett Research

Many researchers all over the world are working hard to expand our knowledge and understanding of Rett syndrome. Helen Leonard and her team in Perth are no exception. During 2004 they have given many presentations all over Australia. International conferences give research scientists the opportunity to talk about their work, listen to new research findings and in some cases collaborate on new avenues to explore.

Both Helen and team member Hannah Moore are due to visit the Child Neurology Society meeting in Ottawa. Hannah will present “InterRett: the application of bioinformatics to international Rett syndrome research” and Helen will talk about “Genotype and early development in Rett syndrome: the value of international data”. They will then travel on to the UK to the Rett Syndrome Association of the United Kingdom Family Weekend where Hannah will introduce InterRett and give a demonstration of the database.

### Money, money, money……

Or so the song goes. But where does the money go, and who from, and why? These are questions we are trying to answer with the 2004 follow-up questionnaire. We ask about the services and care that your family and daughter receive because she has Rett syndrome. This will include medical and other health care such as physiotherapy, and other help you receive, such as around the house and garden. We want to build up a complete picture of health service and care needs that include paid and unpaid care. The unpaid care is especially important and an often ignored part of this type of work, but vital to families relying on it to cope. We also ask about medications, equipment and other items that you need to buy. The financial demand of having a child with Rett syndrome and its effect on the family budget has never been looked at before in such detail.

From your answers we will work out the costs to you, the state and the federal governments, and non-government organisations. We will also be able to identify problem areas and work out where services are most needed. The results can then be used to guide which services are provided and funded, and where they can be placed so that they are most effective. If services are not available the information may be used to advocate for services to be started or expanded.

Sonj Hall and Delia Hendrie from the School of Population Health at The University of Western Australia are responsible for this part of the study. They strongly believe that their work is useless if it doesn’t make a difference to people’s lives and very much hope that this study will help to make sure that the services you and your family need are available and affordable.
Time is just too precious for most of us to be able to spend hours searching the web for relevant websites. Here are just a few that you have found interesting, informative and sometimes just fun.

www.yourshout.org.au The Association for Children with a Disability have recently launched a website for siblings called, ‘Your Shout’


www.incontinence.health.gov.au/caas.htm Several parents have found this site useful. Not all families may have heard of the Continence Aids Assistance Scheme that is offered by the Australian government.

Your answers help everyone

Our research has been the first to be able to show that children with Rett syndrome may be differently affected according to which genetic mutation they have. The data you are providing in 2004 will allow us to advance further with this very valuable work.

- Your answers will help us to better understand how the disorder changes over time in different age groups.
- The resource and cost information collected will identify where improvements could be made in the current services available to your child.
- The information collected this year will be presented in a future report a copy of which will be sent to all families.

WORKS FOR ME ....

We have stories, challenges and day to day life coping with Rett syndrome. We want to tell us just that! But don’t forget that though these ideas work for one…In this edition we are looking at food and diet.

- Poo a day! I have been mixing them with porridge and it seems to have gone from going every three days to almost every day.
- Fibre, calories and vitamins. When liquidised they are just the right consistency for people who have swallowing problems and they are great for constipation!
- Adding diced and small vegetables in with mashed potatoes.
- Peas and sweet corn into Penne pasta tubes. Or mixing in with mashed potatoes.
- Raisins are easy to carry around in your handbag or car. In your handbag and then you can take custards and rice puddings with you wherever you go.

Moveable treats – Small boxes of raisins are easy to carry around in your handbag or car.

Won’t eat vegetables? Try pushing peas and sweet corn into Penne pasta tubes. Or mixing in with mashed potatoes.

Tinned apricots are full of fibre, calories and vitamins. When liquidised they are just the right consistency for people who have swallowing problems and they are great for constipation!

Three prunes a day = One poo a day! I have been mixing them with porridge and it seems to do the trick. My daughter has gone from going every three days to almost every day.

The angel ornaments cost $10 and the lapel pins cost $5 each. There is a small charge for posting and handling. If anyone would like to purchase an Angel, they can go to the web site at www.rett.nesher.com.au, download a form and post it with their credit card details’. Janelle added, “Unfortunately we haven’t come far enough as yet to sell items over the Internet but one day we may get there. I would love it to catch on and maybe other Rett syndrome organizations could sell them too.”

If anyone wants to know more about the Angels and the possibility of selling them please contact Janelle on jlillis@ispdr.net.au
Many of our letters come from parents so it was especially exciting to open one recently, sent by the sister of a girl with Rett syndrome and read her story.

“Hi my name is Rebekah Gale. I’m 12 years old. I have a sister with Rett syndrome, her name is Sarah Gale, and she is 14 years old. I love her very much. I wish she didn’t have Rett syndrome, because then we could go more places and Mum would not have to rush around to get herself and Sarah ready when we were going somewhere, and I could play more games with her.

Our family likes to go to Ticker’s Shack at Temma. There we go to the beach and play in the sand and go swimming. I build sandcastles for Sarah and she knocks them over. I like reading to Sarah and building things with her blocks. She sometimes annoys me while I’m doing something like when I’m sitting watching TV she pulls my hair and chuck’s her toys at me and I get crabby at her.

Sarah’s favourite pastime is sitting on the couch watching football or cricket with Dad. She likes to go 4 wheel driving in Dad’s bus and laughs when we go over bumps. She gives me big hugs all the time. I like to help Mum with Sarah. Sarah loves Bananas in Pyjamas and watches the same episode all the time.

Sometimes I wish she didn’t have Rett syndrome but then when I think about it and if she was different, Sarah and I would fight all the time and I would not be able to push her on her bike or take her for walks. She may annoy me sometimes but I still love her and I always was on such a high that we couldn’t get her to sleep until 3 am!

Our family took Lilly to America in 2001 to learn about the Son-rise program, which is a philosophy on life, and an approach on teaching techniques which we had read about and really loved.

So we keep Lilly happy, we believe that if she is not happy, feeling loved and secure, she will not learn. We break down new tasks for learning into tiny steps and practice all her skills daily in the hope that she will retain them. We entice her to try new things at her own pace and celebrate her for all that she is.”

For anyone who would like to read Son-rise. It is written by Barry Neil Kaufmann and published by HJ Kramer Inc.