

Rett University Conference 16-18 May 2014

Auckland New Zealand

Rett New Zealand hosted their bi-annual family conference in Auckland between 16-18 May 2014. The conference was titled Rett University Conference, in recognition of the work being undertaken by the key presenter Susan Norwell.

The conference presented a detailed two day format, with sessions presented as listed below, and as per the attached video links generously provided by the Rett New Zealand Chairman Mr Dugald MacBrayne:

1. Conference Welcome – Mr Dugald MacBrayne – Rett New Zealand Chairman
<http://www.rettsyndrome.org.nz/conference2014.php?pg=presentations>
2. Focused Learning Solutions – Mrs Susan Norwell “Welcome to the Rett University”
3. NNZ2566 – A new drug for traumatic brain injury, Fragile X Syndrome and Rett Syndrome – Distinguished Professor Margaret Brimble CNZM, University of Auckland
4. Question and Answer Session – Dr Rosemary Marks, Paediatrician, Starship Hospital and President NZ Paediatric Society
5. Education Forum – Mr Doug Laing (Rett New Zealand Treasurer), Mrs Susan Norwell, Ms Helen Philpott (Teacher Aide) and Mrs Lisa Holten (Parent/Occupational Therapist)
6. Group Forums – Related to Age Groups of Girls
7. Further Questions and Discussions – Mrs Susan Norwell
8. Orthopaedic Challenges – Mr Haemish Crawford – Orthopaedic Consultant Surgeon, Starship Hospital
9. iPad Apps and Uses – Mrs Susan Norwell

1. Conference Welcome – Mr Dugald MacBrayne

Dugald welcomed all the attendees, comprising numerous NZ families, some teachers and other interested persons to the Conference. He also acknowledged the ongoing support of Roderick and Gillian Deane as Patrons of Rett New Zealand.

Dugald then introduced Mrs Susan Norwell, a previous speaker at Rett New Zealand conferences, describing her as the ‘David Copperfield’ of Rett Syndrome.

2. Focused Learning Solutions – Mrs Susan Norwell

“Welcome to the Rett University” – www.rett-.org, and www.focusedlearningsolutions.com.

Susan has spent the last 2 years focusing on the use of eye gaze devices by girls with Rett Syndrome, specifically related to reading/writing, and has developed on-line courses (4 hours) on reading and use of eye gaze devices for new beginners. She noted the importance of these courses for teachers.

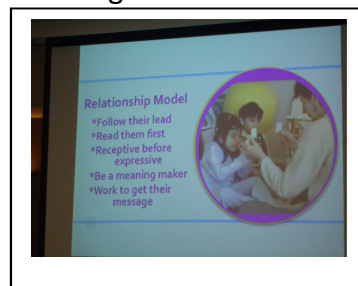
She stressed the importance of talking with girls, and not at them, and to avoid testing them during conversations. Questions should be asked in a facilitative way, and should not be directive or condescending. The conversations should also be interesting, and not boring to the girls!

Susan commented that Rett Syndrome was not a cognitive disorder, but a motor disorder, and it was important for parents and educators to look for ‘how can’, rather than can’t...

In respect of breath holding, Susan noted that it was important not to say to the girls to 'BREATHE' when they were holding their breath, as increased demands can exacerbate the problem, but rather to wait, and to possibly tilt the girl's head backwards to assist.

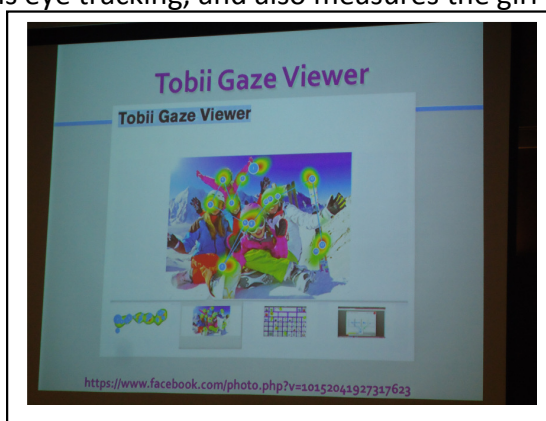
Susan confirmed what parents already know – that the girl's love music and animation (Peppa Pig is a favourite). She described a Relationship Model to assist with learning in order to teach them, being:

- Follow their lead
- Read them first
- Receptive before expressive
- Be a meaning maker
- Work to get their message



Susan noted that when using eye gaze or other communication devices that it was important not to ignore other 'non-verbal' forms of communication, such as nose pointing, or a look for YES and a look away for NO. Eye gaze should be used daily, with use at school important for learning and social interaction with friends. It is also important not to wait for 100% accuracy before responding, but be supportive during the conversation. Susan stressed that use of the eye gaze should be fun, and not just functional.

Susan referred to an alternate, cheaper communication device available being Tobii Gaze Viewer software, at a cost of around \$2,500 compared to around \$18,000 for an Eye Gaze device. This software records voice and performs eye tracking, and also measures the girl's response delays.

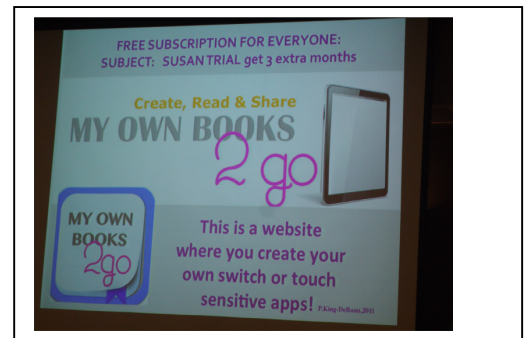
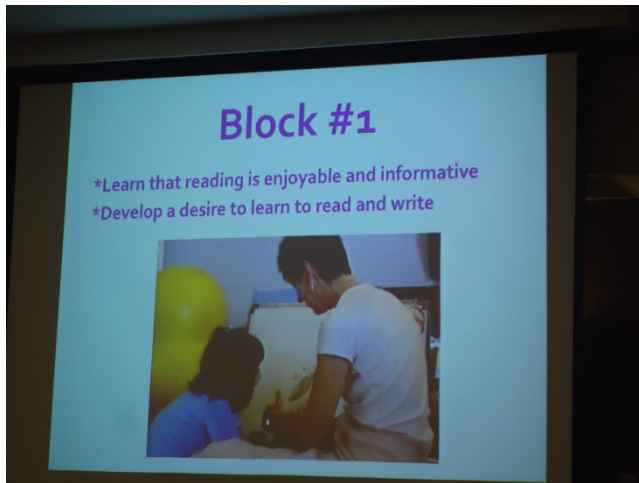


Susan outlined some tips and traps for parents when they have, or are getting an eye gaze, as follows:

- More v less choices on screen, as there can be higher apraxia if given fewer choices.
- Girls learn new pages quickly.
- Should start with a core phrased system.
- MODEL, MODEL, MODEL a word-based language system.

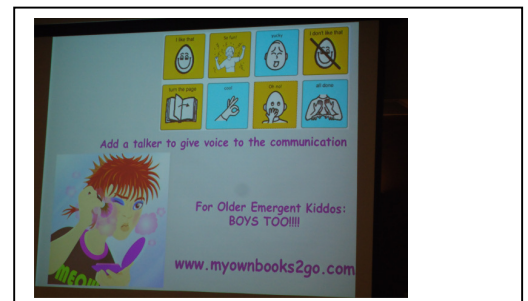
A number of Learning Blocks were discussed, as follows:

BLOCK #1



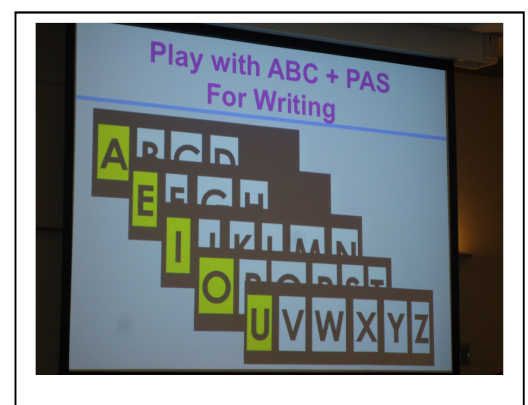
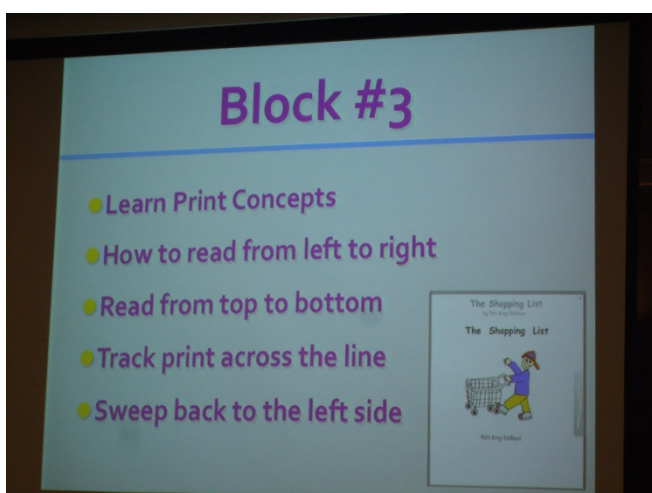
A useful site to review is www.myownbooks2go.com.

BLOCK #2



Susan noted the benefits of Toka Boka Apps.

BLOCK #3



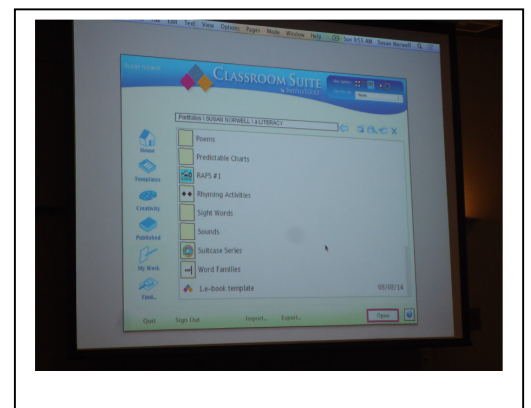
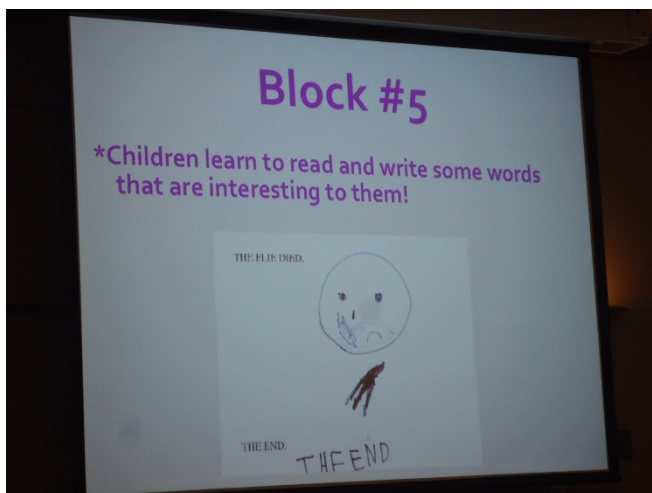
A useful site is www.tarheelreader.org (although a fee will apply).

BLOCK #4



Refer to CLICKER App. Foundations for Reading, and to www.storybots.com that allow girls pictures to be included in video.

BLOCK #5



Susan suggested that it was important for parents in particular to talk to their daughters about Rett Syndrome and apraxia, as they will understand what is being said. They need reassurance that it is not their fault their body doesn't respond.

3. NNZ2566 – A new drug for traumatic brain injury, Fragile X Syndrome and Rett Syndrome – Distinguished Professor Margaret Brimble



Professor Brimble was a winner of the World Women in Science Award from Loreal.

In conjunction with Dr Paul Harris, Professor Brimble was responsible for making NNZ2566.

She noted that the drug had been approved by the United States Food and Drug Administration (USFDA) for Fast Track designation following completion of the current Phase 2 clinical trial, and had been granted Orphan Drug status. NNZ2566 is aimed at Rett Syndrome, Fragile-X and Traumatic Brain Injury, and is working as a neuro-protective agent.

The Phase 2 clinical trial is scheduled for completion in the second half of 2014. 48 female patients have been involved in the trial, with 22 having completed the trial at the time of Professor Brimble's presentation. Should the trial be successful, Professor Brimble noted that more than 30,000 affected girls could be treated for life on a daily basis in order to restore protein into the body. She believed the drug would be suitable for all girls with Rett Syndrome, with a greater dosage for older girls.

As the MECP2 gene is deficient, intervention is intended by use of a chemical to stimulate synaptic maturation. Professor Brimble believes that neurons and synapses will improve over time with drug usage.

Mrs Gillian Deane said she has read social media where parents were very pleased with the girl's progress whilst they were on the drug however, the symptoms came back when the treatment stopped. Professor Brimble had said the drug would be needed to be taken every day. Some of the parents wanted the drug to be fast tracked to be able to use it again.

4. Dr Rosemary Marks, Paediatrician

The session was largely New Zealand focused, and referred to the establishment of a Specialist Rett Syndrome clinic in New Zealand. It was noted that NZ Specialists often conduct clinics by video conference to remote locations, with the families, clinicians and girls all in attendance by video.

5. Education Forum

Mrs Helen Philpott spoke to her experiences as a Teacher's Aide who commenced at kindergarten working with Doug Laing's granddaughter, and then spending the next 8 years working with her. She noted that she worked in a regular school, and highlighted the importance of exercise as part of the regular routine. She supported swimming twice per week, plus exercise every day.

Mrs Lisa Holten spoke about her experiences having recently obtained approval for an eye gaze device. Prior to that, she used 'pretend' paper-based documents to replicate eye gaze symbols whilst awaiting approval.

6. Age Group Breakouts

Having attended the 0-5 year's age group, a number of different matters were discussed including:

- The use of brown sugar and water as a therapy for constipation (1 tsp per 100 mls), or kiwi fruit. Dehydration (evidenced by a dry nappy) also contributes to constipation.
- A number of positive experiences were highlighted about girls attending regular schools. In particular, the ability to interact with kids talking and playing was noted as important, and the added benefit of teaching empathy to the other kids.
- Kathy Hunter's Handbook was noted as being important for parents with newly diagnosed girls.
- The problem of blocked ears was discussed, with a lack of chewing motion considered a possible cause. The use of wet cotton wool in the ears during bath time can assist.

8. Orthopaedic Challenges – Mr Haemish Crawford

Mr Crawford noted the major issues facing girls as being:

- Joint Contractures
- Fractures
- Hip joint dislocation
- Scoliosis

Joint Contractures:

- Ankles are most common (36%)
- Hip issues can occur based on the angle of the hip bone
- Treatments include stretching, Botox and surgical release (rare)

Fractures (14%):

- Usually leg fractures (Vitamin D is important)
- Osteopenia (soft bones)
- Minimal force can cause fractures

Scoliosis (45%):

- On average noticed at 7 years of age
- Curvature of the spine, rotation, pelvic tilt
- Progressive and tight for girls with Rett Syndrome
- A muscular imbalance, with a neurological cause
- The treatment goal is to restore the normal sagittal position
- Scoliosis can restrict lung function, and be very painful
- Surgery is generally required when the scoliosis reaches 40-60 degrees
- At this stage, the surgery is to fuse spinal T2 to the pelvis
- Surgery aims to correct the deformity, and to prevent further growth
- Surgery can assist to reduce chest infections and pneumonia
- Surgery can also help improve eating/nutrition
- In a number of cases for younger girls, expandable rods are being used that can be lengthened every 6 months or so
- Also using magnetic rods that can be expanded outside the skin as day surgery



The question posed by Mr Crawford is does spinal fusion improve the quality of life for treated girls?

- Narayan Study 2006 of 84 patients (Cerebral Palsy) showed a 92% satisfaction rate
- Mercado et al (Spine 2007) analysed 244 papers, and recommended surgery
- 96% of parents recommend surgery, and 84% of therapists

Mr Crawford believes that further quality of life research is needed so that the risks and benefits can be adequately assessed.

The following was included on the back page of the conference agenda:

Do not ask your children to strive for ordinary lives.

Such striving may seem admirable, but it is a way of foolishness.

Help them instead to find the wonder and the marvel of an ordinary life.

Show them the joy of tasting tomatoes, apples and pears.

Show them how to cry when pets and people die.

Show them the infinite pleasure in the touch of a hand.

And make the ordinary come alive for them.

The extraordinary will take care of itself.

Phil Creswell