

TOWARDS A CLINICAL NETWORK

A REPORT ON THE RETT NZ CONFERENCE HELD AT AUCKLAND, 20-22 MAY 2016

The Rett NZ Conference held on 20-22 May 2016 was designed to provide an opportunity for families with women and girls with Rett Syndrome to:

1. Be updated on the latest medical and therapeutic support for women and girls with Rett Syndrome, with lectures from Dr Rosemary Mark and Dr Susanne Davis
2. Learn about the way in which Rett Clinical Networks operate in the US and what lessons these provide for New Zealand
3. Understand the latest research developments from Distinguished Professor Margaret Brimble
4. Participate in individual family consultation sessions with one of NZ's leading Developmental Paediatricians, Dr Rosemary Marks
5. Benefit from individual family sessions with US communications specialist Susan Norwell on non-verbal communications using the latest Tobii devices with Eye Gaze technology to transform the lives of those lacking verbal communications
6. Discuss the ongoing needs of Rett women and girls, especially as they get older and fall outside the oversight of paediatricians and move beyond their school years
7. Reflect on what all these trends and issues mean for women and girls with Rett Syndrome
8. Discuss how a clinical network approach in New Zealand could help provide more comprehensive diagnosis and support
9. Learn from Mr Haemish Crawford, a leading Orthopaedic Specialist and Surgeon, that he would be willing to assist in promoting a Clinical Network style of meeting which would include a range of specialists to provide individual Rett family consultations and advice across a range of disciplines.
10. Facilitate the families meeting each other again and exchanging ideas about the management of Rett Syndrome.

The conference also importantly provided the opportunity for specialist individual family consultations as well as lectures on latest developments and networking opportunities for parents, caregivers and support people, including teachers. It had some elements of a clinical network approach in that a range of specialists attended and provided lectures and individual family advice.

For the first time, separate sessions outside of the Conference proper were held for

1. first, teachers and support people;
2. secondly, paediatricians and specialists interested in Rett Syndrome; and
3. thirdly, speech therapists,

to enable these groups to be updated on the latest research and medical developments as well as advances in Eye Gaze technology and communication teaching techniques for non-verbal students. Audio facilities were provided for paediatricians and specialists from around New Zealand to participate in these sessions.

But it was the intervention of Mr Haemish Crawford which offered a wonderful opportunity for the future. He pointed out the difficulty in a small country of running an ongoing national clinical network but explained that this could be done on an occasional basis when we can gather together the families from across New Zealand and he would undertake to assemble a range of specialists to provide diagnosis and assessment of the variety of challenges encountered by Rett women and girls, from medical problems to treatment options to education issues to support arrangements.

This exciting offer will be taken up by the Rett NZ trustees and planning for such a gathering will commence shortly.

Some of the lessons which emerged from the conference were the following:

1. That the best way to conduct a national clinical network would be on an occasional basis with a concentrated gathering of teachers, paediatricians and other specialists, along with educational and therapeutic support people (and not to attempt to run a clinic on a continuous basis given the small population size and the geographic spread)
2. Participants committed to do this on the next occasion all the families from across NZ can be brought together
3. The Rett NZ Trustees will arrange this with the help of Mr Crawford who has experience of this approach being successful with other disability or special groups
4. This would be a logical extension of the individual family consultations which we organised at this conference in the medical and communications areas
5. The issues of aging and development beyond paediatric care (age 18 or sometimes less) and beyond school support age were substantial challenges which need to be further addressed
6. The separate sessions for each of the specialists, speech therapists and teacher groups were much welcomed and should be organised again at the next conference
7. Eye gaze techniques with the use of Tobii machines is becoming more widespread and is a huge step forward for Rett girls so we must run further training sessions and encourage additional funding initiatives for the machines

For the past three years, Susan Norwell from Chicago, who is an expert in Eye Gaze technology, has been unlocking the communication responses of Rett girls as they choose the symbol or words or letters on the Tobii computer. Now some of the girls are learning to read and write. It was interesting that Susan said some of the older girls who have been in normal schools are able to spell quite well as they have seen the words on the blackboard. The great problem for the girls is the apraxia which means it can take quite some time for them to respond to the questions that have been asked. Eye reading facilitates an increase in confidence and a reduction in frustration for Rett girls. Some of the girls are now able to point to areas of pain and discuss where it is. This has always been one of the problems for parents and children to decipher where the pain is located.

Last conference Distinguished Professor Margaret Brimble spoke to us about designing the New Zealand drug (known as NNZ-2566 or Trofinetide) which has now had a successful FDA Pre-clinical and Phase 1 Trial for Rett women over 18 years and is subject now to further testing in Phase 2, along with FDA trials for Fragile X Syndrome, Traumatic Brain Injury (TBI) and Concussion patients.

At present the drug is being trialled with younger Rett girls at a number of different hospital facilities in the USA. There are also other drugs being trialled under FDA auspices in the US which may help with the agitation and seizures associated with Rett Syndrome.

The parents asked if Professor Brimble could come back again and this year she gave a fascinating lecture on her work including a new melanoma vaccine her laboratory is working on.

About 80 people attended the conference proper including families, caregivers, representatives of Talk Link and some teachers. Participation in non-conference sessions extended to paediatricians and medical specialists, speech therapists, and teachers of children with special needs. The conference charged no fees. Free accommodation and meals were provided to Rett family participants. This was thanks to the generosity of our sponsors, the IHC Foundation and the Deane Endowment Trust. We record our great gratitude to these sponsors. The Chairman made this appreciation abundantly clear at the Conference. With just one exception, all speakers provide their time free of charge which was wonderfully generous.

The speakers at the Conference were:

Dr Rosemary Marks, a senior Developmental Paediatrician at Starship Children's Hospital, and former Chair of the New Zealand Paediatric Society, who has a particular interest in Rett Syndrome.

Dr Suzanne Davis, a Paediatric Neurologist at Starship Paediatric Neurology, who is also very knowledgeable about seizure management and who has experience of Rett Clinics in California.

Mr Haemish Crawford, an Orthopaedic Consultant Surgeon in Auckland, who has provided great help to Rett girls who need complex orthopaedic operations and in particular to resolve scoliosis problems.

Susan Norwell from Chicago USA who is an international expert in Eye Gaze technology and non-verbal communications techniques and who has a special interest in girls with Rett Syndrome. She has visited New Zealand on several occasions including traveling to a range of centres across the country under the auspices of the Rett NZ Trust to assist families with Rett women and girls.

The Trustees of the Rett NZ Trust which organised the Conference are Dugald MacBrayne, Chairman, Doug Laing, Treasurer, Mark Campbell, past Chairman, and Gillian Deane, National Coordinator.