



Grand staircase, Intercontinental Hotel, New Orleans

**7th WORLD CONGRESS on RETT SYNDROME
JUNE 22-26 2012**

Intercontinental Hotel, New Orleans, USA

**A Report on Proceedings by Bill Callaghan,
President, Rett Syndrome Association of Australia Inc.**

7th World Congress on Rett Syndrome New Orleans, USA, 22-26 June 2012

A brief history of the World Congress

Previous World Congresses have been held in Vienna, Austria (1984, 1988); Antwerp, Belgium (1993); Gothenburg, Sweden (1996); Nagano, Japan (2000) and Paris, France (2008). The Congresses are usually spaced four years apart which means that a considerable period of time has elapsed for research and other developments in the study of Rett syndrome, to have taken place. Where a Congress is held is determined by which national Rett syndrome Association offers to look after the organisation of such. Tasks which need to be undertaken include arranging the venue, formulating the programme, recruiting speakers, promoting publicity, determining registration fees and how they are to be paid, designing and filling information packs, and covering all costs associated with an event of this nature.

Venue and programmes

Although it had been known since 2008 that the 7th World Congress was going to be held in America in 2012, it wasn't until January 2012 that the location was announced by the International Rett Syndrome Foundation (IRSF), the Congress organiser, with the venue being the Intercontinental Hotel in New Orleans. IRSF also announced that the event was to be split into sessions for families (22-24 June) and sessions for medical researchers and scientists (25-26 June). Initial registration costs were US\$265 (which included US\$20 for a ticket to the tribute reception on Saturday evening) and US\$465 for the scientific sessions. **This report only relates to the family programme.**

Information packs

Each attendee received a satchel containing information which included a programme; an attendee directory; a keepsake journal; names of IRSF staff, its board of trustees and family advisory board; an IRSF fact sheet; a DVD entitled 'Portrait of Isabella' which is an account of 3 years in the life of 10 year old Rett syndrome child Isabella Roselli of California; Rett syndrome growth charts in regard to body mass index, head circumference, height and weight; a notepad; biro and necklace (a New Orleans Mardi Gras tradition).

The attendee directory gave name, email address, city, state, country, relationship to Rett syndrome child, child's name and age, and *MECP2* gene mutation. Of the 270 listed names, 33 were from overseas with 4 being from Australia. Approximately, 450 persons had registered for the Congress.



Above: The main auditorium, Intercontinental Hotel, venue for the opening day of the Congress

Respite

Two rooms on the same floor where the day sessions were held, were used for respite for a limited number of children and adults with Rett syndrome. A nominal fee was charged but arrangements had to be made pre-Congress. Carers were recruited from the Louisiana based KPH Pediatric Services.

Facilities

The main auditorium was used for all sessions conducted on the opening day and most of the final day. Its main entrance led out to a room in which the exhibitors were located. This room was also used for serving refreshments and boxed lunches. Three nearby meeting rooms were utilised on Saturday and Sunday for the concurrent sessions.

Opening of the Congress

The Congress was opened by Kathryn Schanen Kissam, IRSF chairperson. IRSF has 3 components, IRSF staff (11 members), a Board of Directors (14 members), and a 10 person Family Advisory Board. Kathryn is also a resident of Louisiana, the US State in which this conference was being held.

At right: Paige Nues (IRSF Director of Family Support) introduces IRSF Chairperson Kathryn Schanen Kissam (at right) who, in turn, opened the 7th World Congress



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Opening of the Congress (cont.)

Kathryn explained why New Orleans was chosen. It is a unique city and Rett syndrome is a unique condition. New Orleans suffered great tragedy and devastation as a result of hurricane Katrina. Rett syndrome is a tragic and devastating condition. New Orleans is resilient as is the Rett syndrome family. Kathryn said that "Our wish is that during your time here, you will indeed chart a course for a better tomorrow for those affected by Rett syndrome". New Orleans, too, has charted a course for recovery leading to it being optimistic about its future.

Sessions

There were 37 speakers for the family programme, of whom only 5 were from overseas, 3 from Australia and 2 from Israel. In the scientific programme, there were 20 speakers, 3 of whom were from overseas, 2 from Scotland and 1 from Germany. The website address for the Congress at the time that this summary was prepared was <http://worldcongress.rettssyndrome.org>

A parental viewpoint

Mickie McCool (USA), whose 14 year old daughter Ellie has Rett syndrome, stressed that an individual with the disorder is a fully functioning person inside and the world needs to be made aware of this. She stated that she openly shares her daughter with the community in which her family live and includes full integration in a mainstream school with a modified curriculum.

An overview of Rett syndrome

Dr Alan Percy was one of the first medical professionals in the USA to be involved with the syndrome, with that involvement now spanning 30 years. He has been a regular presenter at World Congresses and conferences held by IRSF and the former International Rett Syndrome Association (IRSA). In recent times, Alan has been regularly called upon to provide an overview of the like that he gave here. He works at the University of Alabama, Birmingham, as a paediatric neurologist and researcher.

He gave the Congress an international atmosphere by playing an audio message of welcome from Swedish researcher Bengt Hagberg. Bengt is the author of the first article to draw world attention to the syndrome, and like Alan, has been involved with the disorder for 30 years.

Alan spoke about the history of Rett syndrome in America and referred to the growth in global research over time. From 1965 to 1999, 600 papers on the condition were published in medical journals but since the year 2000, there have been 1,400. He felt that this rapid pace of basic research promised a bright future for effective therapies.

Of those known individuals in the US who have been diagnosed with 'classical' Rett syndrome, 95% had an MECP2 gene mutation. Eight specific mutations account for 60% of all MECP2 mutations detected. Alan is in charge of a current study which is collecting information on the natural history of the disorder in 1,052 American Rett syndrome individuals. The study is now in its second phase. His data on scoliosis (curvature of the spine) in the syndrome revealed that it is present in 8% of pre-schoolers, 80% of those aged 16 years and under, and 85% of those aged under 26 years. Thirteen per cent of all those with a scoliosis, had had spinal surgery.

His research into life expectancy in Rett syndrome revealed that the odds of living 10 years with the condition were the same as that of the general population living for 10 years. 90% of the Rett syndrome population would still be alive at age 20, 75% at age 30, 65% at age 40, and 50% at age 50 years.

Communication

Apraxia, communication book and techniques

There were a number of sessions which focussed on communication and/or literacy. Linda Burkhart (USA), who is a private consultant and technology integration specialist, is a regular presenter at IRSF conferences. She spoke on the topic of apraxia and communication at the 2010 IRSF conference and this was her topic again.

She reminded the audience that apraxia in Rett syndrome is the inability to carry out a cognitive intent and that it worsens with demand. She displayed a PODD (Pragmatic Organisation Dynamic Display) communication book, an Australian invention created and maintained by Gayle Porter of Melbourne. Linda demonstrated the use of the book with communication switches that she was able to activate by moving her head. She pointed out that one must support the communication intent of the Rett syndrome individual, moving her towards independence over time. Different parts of her body should also be used when attempting to access communication devices. Linda stressed that those directly involved in the communication process in Rett syndrome must not "keep rebooting the system" by asking the same question over and over again.

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Sessions

Communication

Apraxia, communication book and techniques (cont.)

Later on in the conference, Linda gave a two part presentation on 'Light Tech' communication, i.e., methods of communication that usually don't have high degree of difficulty associated with them. Her first session looked at a variety of 'Light Tech' communication strategies involved in teaching yes/no responses, partner-assisted pointing, reading subtle cues, and the provision of an environment for learning a visual language system. PODD communication books were the subject of her second session. In its 'light tech' form, PODD is powered by the partner who attempts to recognise those motor skills the Rett syndrome child has that will assist or enable interaction and social engagement.

Challenges to communication, useful communication assessment tools

'Partnering Families, Teachers and Therapists to Maximise Communication' was the title of the presentation made by Theresa Bartolotta (USA), an Associate Professor of Speech Language Pathology at Seton Hall University in New Jersey. She is also the mother of 22 year old Lisa who has Rett syndrome.

Theresa felt that there isn't a lot of information available to guide therapists in their work with those affected by Rett syndrome. The unique behaviours evident in the disorder also posed a challenge. Even so, the potential for communication is within everyone and the therapist just has to find it. A good start is to get to know the person with the condition very well, a task that ideally should have input from the family. Not only therapists but also families and teachers, need to be aware of the wait time needed by the Rett syndrome individual because of her apraxia. Therapists should also be prepared to create opportunities to communicate utilising a variety of methods. All modalities require evaluation, because not all are going to be effective.

Attention was drawn to two useful tools for assessing and tracking the communication progress. One was the communication matrix (www.communicationmatrix.org) and the other, 'The Inventory of Communication Acts' which was developed in 1996 by Australian Jeff Sigafoos when he worked at the University of Queensland. RSAA has material on the inventory which it received from Jeff. Theresa is in the process of developing communication coaching models that can be used at home, school, and in other communication settings.

Eye gaze tracking technology

Judith Lariviere (USA) is an assistive technology specialist and occupational therapist who spoke about 'ramping up' eye gaze technology to enhance communication and literacy learning for Rett syndrome girls. Her presentation was 'ramped up' as she spoke very fast, not only to the audience but also to those Rett girls with whom she appeared in the videos that she presented. The latter showed her assisting and interacting with them by using a number of different augmentative communication devices. However, Judith's interaction appeared to ignore the technique of allowing Rett syndrome individuals time to both process and act on an instruction.

She also used a lot of interpretation of what a child was trying to communicate, sometimes putting to the child an additional three interpretations within 15 to 20 seconds of making her first interpretation. Those videos that Judith showed of Rett syndrome children using the Tobii eye gaze technology, were filmed from side on and thus it was not possible to see the on screen the choice made by the child.

Judith believes that eye gaze technology taps directly into the Rett syndrome individual's incredible use of their eyes to communicate. It's as if their eye function is their index finger for pointing and gives them the ability to directly select what they want to say. She referred to software which may be of assistance, namely, the "Dynamic Communication Book for Girls", more information about which can be found on the following website www.creativecommunication.com It is phrase-based and the panels are modelled on those used for the Tango communication device. A handout of Judith's presentation on eye gaze tracking technology can be found at <http://worldrettcongress.jlariviere.com>



A young Rett syndrome girl trialling Tobii eye tracking technology in the Congress exhibitor display area

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Communication (cont.)

The Ipad as a communication device?

Linda, Judith and Susan, conducted a workshop together on the appropriateness or otherwise of an Ipad, to support communication and meaningful interaction in Rett syndrome. In addition, they made suggestions about which applications (APPs) might be beneficial for communication, therapy, interaction and leisure.

Linda felt that an Ipad is not far enough advanced to be a Rett child's main system for communication as touch access is challenging, symbol systems are limited, and there is only one switch applicator in existence. Susan, on the other hand, saw it as a tool to expand on play and build language. Judith referred to applications which she thought would be useful, such as Sandra Boynton books, TouchChat HD, mathematic applications, and AbiliPad for working with and writing words. Handouts, together with a list of helpful Ipad web sites, can be found at <http://rettworldcongressipad.wikispaces.com/Ipadhelpfulsites>

To truly benefit from this session, attendees were encouraged to bring their own personal Ipad, fully charged with the latest operating system installed and having already downloaded Sonic Pic (an APP which is available on the iTunes App store).

Eye gaze pilot study

Associate Professor Aleksandra Djukic (USA) is the Director of the Centre for Rett Syndrome at the Albert Einstein College of Medicine in New York City. She had already completed a pilot study which compared the eye gaze of a group of medical professionals with that of a group with Rett syndrome. Slides (one of which appears below) were shown which depict visual discrimination, the study of which led her to conclude that eye gaze was virtually the same for both groups. Eye tracking technology was used to determine where the eyes were focussed.



Although the photo at left is faint, it will hopefully still be useful in gaining an appreciation of the pilot study results.

There are 2 segments, each containing a block of 6 square pictures. The segment, at left, is headed 'Medical professionals', the one at right, 'Our girls' i.e., girls with Rett syndrome.

The coloured blots/blobs on each picture indicate the points at which the eyes of each group member were focussed when their eye gaze was tracked. Overall, the difference between the two groups was found to be insignificant. Her findings were published in a 2012 edition of the journal 'Pediatric Neurology'. RSAA has a copy of the article.

In the near future, Aleksandra intends to investigate -

- Whether the eye gaze in Rett syndrome can be trusted?
- What they are looking or seeing. If looking, what are they looking at?
- If they have an understanding of the pictures that they are looking at?
- If they have an understanding about the world in which they live?
- If they are remembering, and if so, how much is being remembered?

Literacy

Susan Norwell (USA) is an educational specialist from North East Illinois University. She has worked with a wide array of students, primarily those with autism spectrum disorders, for more than 20 years. In May 2012, Susan was a guest speaker at Rett New Zealand's biannual conference which was held in Auckland. Unfortunately, I was unable to be at her talk 'Beyond Emergent Literacy - Let's Teach Them to Read' as I was attending another presentation. There were no handouts. From what I can gather, the emphasis of her talk was on the development of a balanced literacy program which teachers could use in their classroom with Rett syndrome students, the main elements of which were word use, guided reading, self-selected reading and writing.

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Therapies

Multi-disciplinary teams, genetic research, drug trials, fish oil

Professor John Christodoulou, head of the NSW Centre for Rett Syndrome Research in Sydney, and one of three Australian speakers at the Congress, referred to the importance of a multidisciplinary team approach which focusses on addressing the physical needs of individuals with Rett syndrome. Do they need occupational therapy to assist with sitting, hand control, communication and/or personal care? Is speech therapy required to assist with feeding and communication? Can physiotherapy play a role in improving mobility, and the functioning of joints and muscle? Is there a need for a dietitian to address issues like constipation and/or nutritional support? Dental health could be a problem because of teeth grinding, drooling or tongue thrusting, so a dentist may need to be a member of the team.

Studies, including that of Mari Kondo from the Howard Florey Institute in Melbourne, which investigated environmental enrichment in Rett mice, suggest that manipulation of brain derived neurotrophic factor (BDNF) levels or of the BDNF signalling pathways, could be a therapeutic option in the future. Environmental stimulation such as music therapy, and being in a rich and colourful setting, may be an important component of early intervention in Rett syndrome.



World Congress presentation by Professor John Christodoulou, head of the NSW Centre for Rett Syndrome Research

Another area for potential treatments in the syndrome lies with the genetics of the disorder, namely, at the *MECP2* gene level. It may be possible to activate the normal *MECP2* gene or repair what has been damaged. Thirty per cent of *MECP2* mutations in Rett syndrome are nonsense mutations which can cause a shortened version of the *MeCP2* protein to be made and upset its stability. There are some drugs available that can 'trick' cells into making a more or less normal version of that protein.

Human and mice studies in the past have involved substances like L-carnitine, creatine, folate-betaine, naltrexone, choline, ampakines, HDAC6 inhibitors and sarizotan. Some improvements were observed for most of these treatments but none benefited all the persons or mice who were being administered a particular drug. Currently, there are several overseas drug trials underway. Walter Kaufman from the Boston Children's Hospital (USA) is investigating Insulin Growth Factor 1 (IGF1) as a means of treatment. Josette Mancini from the Assistance Publique Hospital in Marseille (France) is recruiting persons with Rett syndrome to pilot study the use of Desipramine, while Sakkubai Naidu is presently in phase 2 of assessing the use of Dextromethorphan in the disorder.

Claudio De Felice from Siena, Italy, who trialled the use of fish oil by 20 Rett syndrome girls who have an *MECP2* mutation and are in stage 1 of the disorder, reported improvement in their motor abilities, hand use, nonverbal communication and breathing. The questions that his pilot study raises such as what is the optimum dosage, how long does one use this treatment, and its impact on organs, warrants more detailed study using the Rett syndrome mouse models. A copy of De Felice's findings on his fish oil trial, is held by RSAA.

John's presentation is available on DVD and can be purchased from the World Congress web site, the address of which appeared on page 2.

Physical therapy

Meir Lotan, an Israeli physiotherapist, has been actively involved with Rett syndrome since the mid 1990s. He began his presentation by emphasising that a physical intervention program should be in place now for persons with the disorder so that they are in their best physical shape when the cure for Rett syndrome is found. Whatever the cure is, it will not remove the physical problems that the Rett child or adult already has.

Meir described an intensive physical program used with a 3 year old Rett syndrome child and with whom he and members of the child's family, all played a part. Just prior to beginning this intervention, the child had no hand function, could not move independently, and often screamed. During the program, her father supported and interacted with her while she was in a standing frame, her brother would put her on all fours as well as helping her walk, and with the child sitting on a therapeutic cushion, her sister would read to her and help her play with toys. Each session would be for half an hour and when appropriate, the family would show her cartoons. Six months after starting this program, the child no longer screamed, she walked with help, stood independently and drank from a cup unaided.

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Therapies

Physical therapy (cont.)

Physical intervention and the evaluation of such should be carried out each day of the week according to Meir. He said that Rett syndrome girls are awake for 100 hours per week. His physical intervention program uses just 5% of that time. It has 5 components which are done twice daily, each lasting half an hour. It consists of physiotherapy, occupational therapy, hydrotherapy, speech and music therapies. If the child is unwell, concentrate on passive activities such as using pillows to work against scoliosis by strengthening her back muscles. Continuity is essential so when she gets well, resume the daily intervention or what Meir referred to as the "24/7 activity program".

It was interesting to watch a video of Meir working with the girls to improve their proprioception, the malfunctioning of which can negatively affect their scoliosis. While massaging and moving their heads and shoulders, he would sing to them. He is a firm believer that scoliosis can be physically managed in Rett syndrome, that physical fitness can be improved by such means as use of a treadmill, and that walking can be maintained or regained. All of these outcomes, he has achieved.

Music therapy

Another presenter from Israel was Cochavit Elefant who is a music therapist and co-founder of the Israeli National Rett Syndrome Association. She reinforced what is already known in Rett syndrome that music promotes and motivates the desire of the girls to interact and communicate, to express themselves, and develop cognitive, sensorimotor and physical skills.

Occupational therapy

Aside from physiotherapy, music and speech therapies, the only other session specific to therapies was that presented by Carrie Luse (USA) who is an occupational therapist. She views this form of intervention as a means of increasing the independence of the Rett syndrome girl, a vital element of which is easy access. Access involves a number of things, namely, use of a body part to perform an activity; modification or adaptation of the environment to make tasks easier; conducting activities that are within her reach; the use of technology including switches; and the utilisation of objects that not only appeal to her but are shaped in a way that she can use them. To perform each of these tasks properly, the occupational therapist must get to know the girl, find out what she likes and dislikes, and what makes her anxious.

There are a number of barriers in Rett syndrome which impede independence. Self-injurious behaviour is one. If she pulls her hair, she should be assessed for arm splints or a swim cap; if the skin of her hand (s) is sore or raw, due to biting, mouthing or hitting herself, the use of elbow or hand splints should be looked at. The repetitive hand movements are another barrier. Consideration should be given to inhibiting her use of her non-dominant hand during functional activities such as eating or interacting with someone. The environment in which she is in at a particular time, be it home, school, outdoors, car or respite, can actually inhibit her freedom of movement because of things like noise, temperature or seating. These inhibitions can be remedied quickly. Her freedom of movement may also be impaired because of sensorimotor deficiencies that can be present in Rett syndrome such as depth misperception. As a result, there may be a need to reassure her, guide her, or change the environment.

Health issues

Nutrition, feeding and gastrointestinal problems

Particular health issues in Rett syndrome were discussed in a number of sessions. Good nutrition can be compounded by gastrointestinal problems such as reflux, constipation, air swallowing, and feeding problems like poor chewing, a limited ability to self-feed, mouthing and drooling. Poor nutrition causes problems with weight, bone density and body growth.

Measures to assist nutrition include high protein/high caloric diets, modification of food texture, proper posturing, good hydration, nutritional supplements, specialised feeding programs such as tube feedings, as well as proper management of issues such as reflux and constipation. American recommendations were presented on selected vitamin dosages by age. Daily calcium amounts were 700 milligrams (mg) for those aged under 4 years, 1000 mg for 4 to 8 year olds, 1300 mg for 9 to 18 year olds and 1000 mg for those aged 19 years and over. Calcium absorption is promoted by vitamin D, the recommended US daily intake of which was 15 micrograms for persons aged from 1 to 70 years.

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Health issues (cont.)

Scoliosis and other orthopaedic issues

I didn't attend this session but evidently the speakers discussed the prevalence of, and risk factors for, scoliosis; how it is diagnosed and monitored; and ways that it is managed, be it by physical activity, postural care or surgery. An overview of hip instability and the development of foot deformities were also presented. Jenny Downs, a physiotherapist and chief investigator with the Australian Rett Syndrome Study, was one of the speakers.

As at mid-October 2012, there were 15 sessions from the World Congress available on DVD which can be purchased from the Congress web site (see page 2 for its address). The presentation on 'Scoliosis and Orthopaedic Issues' is one of those, as is the following session on seizures.

Seizures

Daniel Glaze (USA) has been the medical director of the Blue Bird Rett Centre at the Texas Children's Hospital, Houston, for 25 years. His talk was on seizures, a topic that he has spoken about at many Rett syndrome conferences. I did not attend either of Daniel's presentations. What I can gather is that he explained what a seizure and epilepsy are; the relationship of seizures to age and to *MECP2* gene mutation; severity of seizure; the need for an electroencephalogram (EEG); treatments; and non-seizure events that appear to be seizures.

Sleep problems

Daniel also spoke on the management of sleep problems in Rett syndrome. He is the primary investigator for the sleep component of a current American study, the 'Natural History of Rett Syndrome'. Steps to improve sleep were to be described as too were causes of sleep disruption, sleep-related breathing problems, behavioural management and medications.

Emotions and behaviours

The presentation on emotions and behaviours in Rett syndrome was given by Sarojini Budden (USA), a developmental paediatrician with 27 years of experience with the disorder. Sarojini grouped emotions and behaviours by age. Irritability, crying, poor sleep, social withdrawal, loss of language and hand use, are characteristic of the 18 month to 3 year old age group. Screaming, hair pulling, hitting, biting, anxiety, pacing, inattentiveness and hyperactivity, are significant in the 5 to 10 year group. For those aged 11 to 20 years, moodiness, sleeplessness, unexplained crying, loss of interest in previously enjoyed activities and signs resembling depression, are prominent.

Puberty to menopause

I'm unable to provide first hand information on the session given by Jane Lane (USA) on her topic of the maturing woman with Rett syndrome as I was not present. I am aware that she referred to findings from the American study 'Natural History of Rett Syndrome' in relation to puberty and menarche, the management of menstruation; as well as the role that hormones play in seizures. She has spoken on this topic before at an IRSF conference.

Education

Only one Congress session centred on schooling and that was about one individual student with Rett syndrome, 13 year old Ellie McCool (USA). Her mother Mickie, and teacher Jennifer Ethridge, described the success that they had had in integrating Ellie into a mainstream suburban Missouri school using a modified curriculum. I was not present, however, a DVD of this talk is available for purchase from the World Congress web site.

Parent and family support

Chat sessions

Immediately following the last speaker on day one, groups of attendees got together in various meeting rooms to participate in what were termed 'Crackerbarrel Breakout' sessions. These were informal discussions on the topic of 'Conference Hopes and Expectations', with who was in each group being determined either by the age of the family's Rett syndrome child or the relationship one had with the child.

At the conclusion of the programme on day 2, another round of informal talks were held but just for three groups, fathers only, grandparents only, and siblings (open to anyone). I was present at the 'Dad's only' segment, which involved a panel of 4 American fathers with Rett syndrome daughters of differing ages. Issues raised and discussed included marriage, divorce, guilt, management of a daughter's personal hygiene, and employment.

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Parent and family support (cont.)

Caring for the adult with Rett syndrome

I didn't attend this presentation which was given by 3 parents who have an adult Rett syndrome family member. They spoke of the care that they provide and the plans that they have for the future care of their daughters. A DVD of this session can be purchased via the World Congress web site.

Planning for the future

Mary Ehlert (USA) has her own business called 'Protected Tomorrows' which helps families plan a safe and fulfilling life for their family member with special needs. Her plan addresses issues such as how to manage the transition of the disabled family member from residing with the family to living away from home, how to maximise government benefits, and coming to grips with legal and financial situations. I would suggest that the content was more relevant to an American audience rather than an international one.

Scientific topics



World Congress presentation made by Helen Leonard, Director of the Australian Rett Study and Manager of InterRett, an International Rett Syndrome Study.

Rett syndrome databases

Helen Leonard, Director of the Australian Rett Syndrome Study, began by stating that families want answers to questions like what causes the disorder, what lies ahead, will she walk, will she get epilepsy, what is the best therapy, what is the best type of school to attend, what is the most suitable form of respite care, and many more.

Information collected and stored on databases, such as the Australian Rett Syndrome Study and InterRett, can assist in answering these questions. Publications using data obtained from the former have made significant contributions to understanding how common the syndrome is, functional ability, genetic characteristics, natural history and medical complications.

Helen's presentation can be purchased on DVD from the World Congress website.

Disease classifications

An extremely small audience of 5 attended this session which was devoted to classifications used to catalogue or code health disorders. It was presented by Walter Kaufman (USA), Director of the Rett Program at the Children's Hospital, Boston. The 4th and 5th editions of the Diagnostic and Statistical Manual of Mental Disorders (DSM-4 and DSM-5) classify Rett syndrome with autism spectrum disorders. Debate still continues as to whether this is the correct category for the condition.

The World Health Organisation produces and maintains the International Classification of Diseases and Related Health Problems (ICD) to classify health disorders. The most recent revision, the 10th, does not have a unique code for Rett syndrome.

Malfunction of the autonomic nervous system

The session on the importance of problems associated with the autonomic nervous system in Rett syndrome, which was given by Jeffrey Neul (USA) from Baylor College of Medicine, Houston, is available for purchase on DVD from the World Congress web site. The autonomic nervous system controls basic bodily functions that are not consciously controlled, like breathing, temperature regulation and heart rhythm, the functioning of which can be irregular in Rett syndrome. Potential ways to treat these problems were to be put forward.

Brain

When the molecules that control how a brain forms do not function properly, disorders such as Rett syndrome occur. I wasn't present at the session which was entitled 'From Mind to Molecules: The Neurobiology of Rett Syndrome' delivered by Jennifer Larimore (USA), an independent research investigator. From what I can gather, her talk referred to the basic concepts of neurobiology and how these can relate to the dysfunction occurring both in the entire brain and in individual brain cells, in a patient with Rett syndrome.

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Scientific topics (cont)

Genetics

Another presentation that I could not attend was 'Genetics: Beyond the Basics' given by Steve Skinner (USA), a clinical geneticist from South Carolina. He provided an insight into the interpretation of *MECP2* gene mutations and postulated on what role genetic knowledge might play in future treatments. A DVD of Steve's talk can be obtained via the Congress website.

MeCP2 protein, *MECP2* gene mutations

Anna Kalashnikova (USA) from Colorado State University studies how the *MeCP2* protein works and this was her topic at the Congress. I was attending another talk so I cannot comment on what she presented. However, this session can be purchased on DVD from the Congress web site.



Huda Zoghbi (USA) began her career as a paediatric neurologist, but a chance encounter with a young Rett syndrome child caused her to shift from clinical practice to genetics research. In 1999, she was head of the team at Baylor College of Medicine who discovered that mutations in the *MECP2* gene were the main cause of Rett syndrome. Huda was the keynote speaker at the World Congress and talked about the *MECP2* gene and the protein that it produces.

Studies involving Rett syndrome mice have improved our knowledge of *MeCP2* protein. Even though the ability of the *MECP2* gene to produce that protein has been impaired, protein is still being made but at lower levels than it should be. Huda said that it was better to have some *MeCP2* protein than to have none at all.

Mutations in the *MECP2* gene cause Rett syndrome and several neuro-developmental conditions, such as cognitive disorders, autism, juvenile-onset schizophrenia, and encephalopathy with early death. Mice lacking *MeCP2* protein from GABA-releasing neurons (nerve cells) reproduce numerous Rett syndrome and autistic features, including repetitive behaviours. Loss of the protein from a subset of forebrain GABAergic neurons also results in many features of Rett syndrome. Enhancing GABA (y-aminobutyric acid) signalling, using drugs to increase GABA levels at the synapse (the minute spacing that separates one nerve cell from another), might help Rett patients, she suggested.

Huda Zoghbi, immediately after presenting the World Congress keynote speech at the conclusion of the conference

(y-aminobutyric acid) signalling, using drugs to increase GABA levels at the synapse (the minute spacing that separates one nerve cell from another), might help Rett patients, she suggested.

Huda stressed that *MeCP2* protein is critical for the maintenance of neurological function, and that the introduction of a small amount of that protein, might make a key difference in how Rett syndrome manifests itself as an affected child matures. This possibility needs to, and will be, explored. Huda's presentation is available for purchase on DVD from the World Congress web site.

At the conclusion of her talk, I introduced myself to her. I thanked her for what she and her fellow researchers had contributed to the knowledge of the syndrome, particularly the discovery of the Rett gene back in 1999. I told her that within 30 minutes of the announcement being made in Houston, RSAA was aware of it, and that within 7 days, all those Australian families with whom the Association had direct contact, had been informed.

Tribute reception



On the Saturday evening in the main auditorium, a large number of Congress attendees paid tribute to those with Rett syndrome. Parents, when registering for the Congress, were invited to email photographs of their Rett syndrome daughters if they wanted them to be included in a musical video tribute. The function provided an opportunity for those hailing from Australia, to get together informally

Left to right: John Christodoulou (Rett researcher from Sydney and speaker at the Congress), Phil Creswell (from Hobart and father of 2 year old Rett syndrome daughter, Maria) and Bill Callaghan (RSAA President), catch up at the tribute reception.

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Exhibitors

There were two exhibition rooms on the same floor where the talks were presented. One room was occupied by organisations involved in Rett syndrome research and direct health care, and the other, by those with products which might be of assistance in managing the condition.

Research and direct health care

It was difficult to gain an appreciation of the number of organisations involved whose speciality was research or direct health care. There were at least six tables, but they were not always manned nor did they have material on them which one could peruse. To its credit, the table manned by the Australian Rett Syndrome Study always had someone present to assist with enquiries, not only about the Study but also InterRett. Rett Syndrome Europe had fliers promoting the 3rd European Rett Syndrome Congress which is to be held in Maastricht, The Netherlands, in October 2013. There was also material available relating to the Rett Syndrome Clinic at the University of Alabama, Birmingham, and the Children's Hospital at Montefiore, located in the Bronx, New York City. The Blue Bird Circle Rett Centre at the Texas Children's Hospital, in Houston, was another American organisation to be represented. The exhibitions relating to research and direct health care were mostly applicable to the US.

The Greenwood Genetic Centre's mobile science laboratory, also known as the 'gene machine', was a bus equipped with a state of the art genetics laboratory. The vehicle was parked outside the Congress hotel about 40 metres from the main entrance. The Greenwood Genetic Centre, which is located in South Carolina, uses the laboratory to further genetic education. Congress attendees had the opportunity to see a demonstration of testing for *MECP2* gene mutations.

Product exhibitors

Tobii is a Swedish company which was formed in Stockholm in 2001. It offers a range of augmentative alternative communication devices which incorporate eye tracking/eye control, text to speech conversion, communication boards, and accessories such as switches, mounts and headmice. A picture of a Rett syndrome girl visiting the Tobii exhibit, appears on page 3.

As this company's products were often referred to in those sessions on communication, it was fitting that it was an exhibitor. It was a popular presentation. There always seemed to be an adequate supply of product information, plus two or three Tobii representatives were there throughout the day to answer queries. Contact details were supplied for Australia, Asia, Europe, and not just North America.

Another exhibitor whose focus was on communication was 'Talk to Me Technologies' which produce speech generating devices as well as the 'Intelligaze Communication System' which includes eye gaze tracker technology. Aside from website and email addresses, only an American phone number was given in the advertising material. The same thing applied to those exhibits relating to compression garments, innovation suit therapy and arm braces, a single brochure for each being all that I could obtain.

Availability of Rett syndrome resources

Session DVDs

As has been already been referred to in this report, selected Congress presentations are available for purchase from its web site <http://worldcongress.rettssyndrome.org> in blocks of 2, 3 or 4 sessions, ranging in cost from US\$20 to US\$40 per block. As at October 2012, 15 of a possible 35 presentations were available from this source. Given the confidentiality restrictions that applied to the scientific programme, none of those sessions will be available for public release.

Publications

The table which contained publications on the syndrome included several copies of the 2nd edition of 'The Rett Syndrome Handbook' and 'Raindrops and Sunshine', both authored by Kathy Hunter, Founder and President of the International Rett Syndrome Association (which became IRSF in 2007).

Handouts

Only a small number of speakers made available handouts of their talks, 2 of which can be accessed via the Congress web site. RSAA has copies of handouts provided by John Christodoulou (Therapies for Rett Syndrome), Suzanne Geerts (Nutrition), Kathleen Motil (Gastrointestinal issues) and Carrie Luse (Occupational Therapy).

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Meeting of representatives from Rett syndrome Associations

Once the congress had concluded, a meeting was chaired by Stephen Bajardi, IRSF Executive Director, with those attendees representing Rett syndrome Associations in Australia, Canada, Denmark, Germany, Norway, Russia, Sweden, The Netherlands and the United States. There were also four people from Mexico who were there to seek help in forming an Association, only one of whom could speak English. Neither of the two Russian Association women could speak English.

Those attending the meeting are pictured below.



Attendees at the meeting which was held at the end of the Congress for representatives of Rett syndrome Associations - Front row (left to right): Claudia Petzold (Rett Syndrome fundraising group, Germany), Stephen Bajardi (CEO, IRSF), Terry Boyd (President, Ontario Rett Syndrome Association), Female with black top (Not known) and Martine Gaudy (Secretary, Rett Syndrome Europe).

Second row (left to right): Female (Mexico), Female (Mexico), Male with dark beard (Mexico), Bill Callaghan (President, Rett Syndrome Association of Australia), Olga Timutsa (President, Rett Syndrome Russia), Female in red (Rett Syndrome Russia), Anna Davidsson-Karnevi (Swedish Rett Syndrome Association), Winnie Nordberg-Pedersen (President, Danish Rett Syndrome Association), Marielle Van Den Berg (Chairman, Dutch Rett Syndrome Association), Thomas Bertrand (Rett Syndrome Europe), Hilde Friis (Rett Syndrome Association of Norway) and Ingrid Harding (President, Girl Power 2 Cure, USA, and Rett Syndrome Research Trust board member).

Back row (left to right): Male with grey beard (Mexico), Shari Hamelin (Ontario Rett Syndrome Association), Male (Danish Rett Syndrome Association), Male (Danish Rett Syndrome Association), Male (Danish Rett Syndrome Association) and Dr Pavel Belichenko (Rett Syndrome Russia).

Lack of translational support, the busy scheduling with concurrent sessions, and the absence of a welcoming reception, were other aspects of the Congress that were raised at the meeting. It was also suggested that an agenda for this meeting should have been prepared pre-Congress and included an item regarding the facilitation of communication between national Rett syndrome Associations.

Thomas Bertrand (Rett Syndrome Europe) informed the meeting about RareConnect, which is a website set up to globally connect those suffering from, or involved with, a rare disease. It is meant to be a safe place where affected families and individuals can share experiences, find helpful information and resources. There is a section specifically devoted to Rett syndrome. RareConnect was created by EURORDIS (European Rare Disease Organisation) and NORD (National Organisation for Rare Disease) and has translational capabilities. Its website address is <http://www.rareconnect.org>

**7th World Congress on Rett Syndrome
New Orleans, USA, 22-26 June 2012**

Kathy Hunter, Founder and Former President of the International Rett Syndrome Association (IRSA)

On the day that the scientific programme began, my wife and I caught an early train to Washington DC. I had no misgivings about missing this part of the Congress. Given the confidentiality restriction in place for all attendees, I would not have been able to report on what was presented.

Soon after our arrival in Washington DC, I rang Kathy Hunter (IRSA Founder and President) and author of 'The Rett Syndrome Handbook' who lived nearby in Maryland. Subsequently, she, her husband Scott, and their 37 year old Rett syndrome daughter Stacie, came to our hotel and spent four hours with us. As Kathy had not wished to attend the Congress, I told her how I thought it went.

Conclusion

Congress sessions

Speakers from overseas were conspicuous by their absence in both programmes. As such, I felt that the programme was limited in what it could offer in global terms.

Australian representation

Sue Hallenstein and her daughter Lucie (from Melbourne), together with Phil Creswell (Hobart), were the other Australian Rett syndrome parents who made the trip to New Orleans for the Congress. No easy task, what with having to leave their families behind, time off work, and the considerable cost involved. I thank them for the huge effort they made in being there.

The other Australians attending, Helen Leonard, Jenny Downs, Alison Anderson, Stephanie Fehr and Anna Urbanowicz, from the Australian Rett Syndrome Study (Perth), and John Christodoulou from the NSW Centre for Rett Syndrome Research (Sydney), are all involved in Rett syndrome research and deserve a pat on the back for their successful contributions to the conference.

A World Congress in Australia?

New Orleans was an unusual choice to locate the Congress because of its heat and humidity, history of hurricanes and for those coming from overseas, the distance and cost. I feel that the latter may have contributed to the small European attendance. Would Europeans and Americans come to Australia in significant numbers to make a World Congress held here an international event of consequence?

The organisation of past World Congresses on Rett Syndrome has always had considerable involvement of the national Rett syndrome Association. It is expected that that organisation promotes it, finds the venue, pays for such as well as for the airfares and accommodation of overseas speakers, provides input into the programme, arranges social activities, etc. Depending on resources, planning and implementation would take from one to two years.

The next World Congress will take place in Moscow on 13-17 May 2016.

Bill Callaghan

President

Rett Syndrome Association of Australia

12 October 2012.