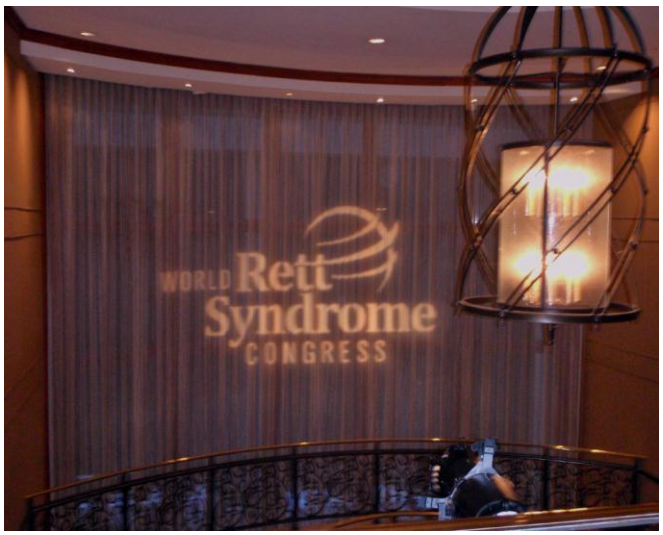
7th World Congress on Rett Syndrome - New Orleans

Previous World Congresses have been held in Vienna (1984, 1988); Antwerp (1993); Gothenburg (1996); Nagano (2000) and Paris (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 usually determined by which national Rett syndrome Association offers to look after the organisation of such. For last year's Congress, the organiser was the International Rett Syndrome Foundation (IRSF) which is in the United States, with the venue being the Intercontinental Hotel in New Orleans. The event was split into sessions for families (22-24 June) and sessions for medical researchers and scientists (25-26 June).



Grand staircase, Intercontinental Hotel, New Orleans

RSAA was represented at the Congress by its President, Bill Callaghan, and it is he who has provided the following summary of the family component of the Congress.

Opening of the Congress

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

She 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 in 2005. Rett syndrome is a tragic and devastating condition. New Orleans is resilient, the Rett syndrome family is resilient. As New Orleans has charted a course for recovery and is optimistic about its future, Kathryn stated 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."

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.

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 is fully integrated into 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 his association 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 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 feel 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. As mentioned earlier, 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 'classic' 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 2nd 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 under 17 years, and 85% of those aged under 26 years. Thirteen per cent of all those with a scoliosis, had had spinal surgery.

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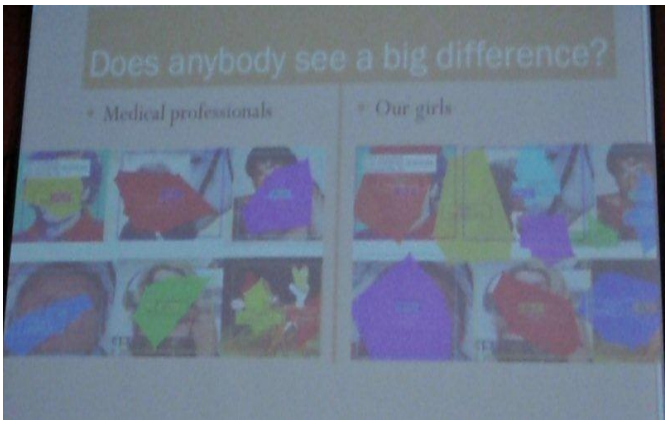
An overview of Rett syndrome (cont.)
His research into life expectancy in Rett syndrome revealed that the odds of living 10 years with the condition were the same as that of the general population living for 10 years. At age 20, 90% of the Rett syndrome population would still be alive, 75% at age 30,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

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 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 above 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', and 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 focused when their eye gaze was tracked. Overall, the difference between the 2 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 work out

- Whether the eye gaze in Rett syndrome can be trusted?
- 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?

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.

Later on in the conference, Linda gave a 2 part presentation on 'Light Tech' communication, i.e., methods of communication that usually don't have a 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 wasn'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.

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Communication (cont.)

Challenges to communication, useful communication assessment tools (cont)

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 marred by several things. For instance, her delivery 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 saw 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 an additional three interpretations to the child within 15 to 20 seconds of making her first interpretation. The videos that Judith showed of Rett syndrome children using the Tobii eye gaze technology, were taken from side on and thus it was not possible to see the on screen 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. Its 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.

Eye gaze tracking technology (cont.)

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 at the Congress exhibitor display area

Literacy programme

Susan Norwell (USA) is an educational specialist from North East Illinois University. She has worked with a many students, primarily those with an autism spectrum disorder, 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.

The title of her talk in New Orleans was 'Beyond Emergent Literacy - Let's Teach Them to Read'. 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, both guided reading and self-selected reading, and writing.

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 the 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 *rettworldcongressipad.wikispaces.com/Ipadhelpfulsites*

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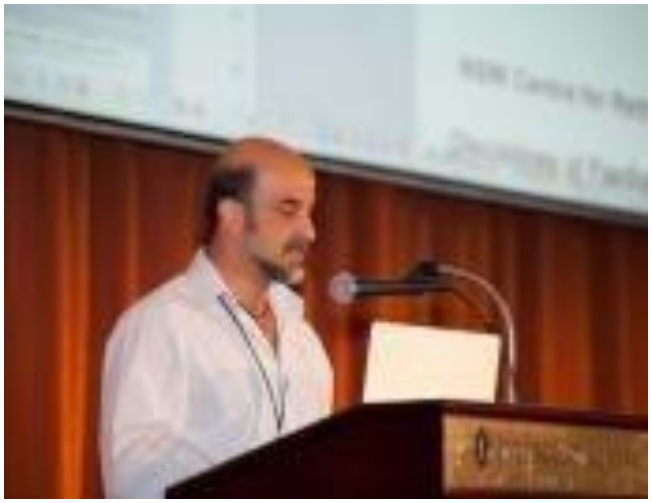
Therapies, potential treatments

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.



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

Research, including that of Mari Kondo from the Howard Florey Institute in Melbourne, which investigated environmental enrichment in Rett mice, suggests 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.

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 thus 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 Rett mice who were being administered a particular drug.

Currently, there are several overseas drug trials underway. Walter Kaufman from the Boston Children's Hospital is investigating Insulin Growth Factor 1 (IGF1) as a means of treatment. Josette Mancini from the Assistance Publique Hospital in Marseille is recruiting persons with Rett syndrome to pilot study the use of Desipramine, while Sakkubai Naidu in Baltimore is presently in phase two 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 Rett syndrome mouse models. A copy of De Felice's findings on his fish oil study, 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 is *http://worldcongress.rettsyndrome.org*

Physical therapy

Meir Lotan, a 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 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.

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, potential treatments (cont.)
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 7 hours 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".

A video of Meir working with the girls to improve their proprioception, the malfunctioning of which can negatively affect their scoliosis, was shown. While massaging and moving their heads and shoulders, he would sing to them. He is a firm believer that scoliosis in Rett syndrome can be physically managed, that physical fitness can be improved by such means as using 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 well known in Rett syndrome that music promotes and motivates the desire of the girls to interact and communicate, to express themselves, and to 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,

Occupational therapy (cont.)
due to biting or mouthing or hitting herself, the use of elbow or hand splints should be investigated. 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 like 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. Proper management of issues such as reflux and constipation will also aid nutrition. 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.

Scoliosis and orthopaedic issues
Discussion centred upon 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.

At present, there were 15 sessions from the World Congress available on DVD. These can be purchased from the Congress web site *worldcongress.rettsyndrome.org* The presentation on 'Scoliosis and Orthopaedic Issues' is one of those, as is the following session on seizures.

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Heath issues (cont.)
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. He explained what seizures and epilepsy are; the relationship of seizures to age and to MECP2 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 described such as behavioural management and medications. One cause of sleep disruption is a sleep-related breathing problem.

Emotions and behaviours
Sarojini Budden (USA) is a developmental paediatrician with 27 years experience with the disorder. She 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
Jane Lane (USA) spoke about the maturing woman with Rett syndrome, referring to findings from the American Natural History of Rett Syndrome study in regard to puberty and menarche, the management of menstruation as well as the role that hormones play in seizures. She has spoken on this topic at a previous IRSF conference.

Education
Only one Congress session centred on schooling and that was about an 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.

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 3 groups, fathers only, grandparents only, and siblings (open to anyone). The 'Dad's only' segment 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.

Caring for the adult with Rett syndrome
Three parents with an adult Rett syndrome daughter spoke of the care that they provide and the plans that they have for their future care. 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. The content was more relevant to an American audience.

Scientific topics
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 featuring data obtained from the former have made significant contributions to understanding its genetic characteristics, how common is the syndrome, the natural history of the disorder, the degree of functional ability, and medical complications that may arise.

Rett syndrome conferences
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Scientific topics (cont.)
Rett syndrome databases (cont.)

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

Helen's presentation can also be purchased on DVD from the World Congress web site.

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 Boston Children's Hospital. 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 all irregular in Rett syndrome.

Brain
When the molecules that control how a brain forms do not function properly, disorders such as Rett syndrome occur. The session on 'The Neurobiology of Rett Syndrome' was presented by Jennifer Larimore (USA), an independent research investigator.

Brain (cont.)
She 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.

Genetics
Steve Skinner (USA), a clinical geneticist from South Carolina, 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 also 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. This session too 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.

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

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.

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Scientific topics (cont.)
MeCP2 protein, MECP2 gene mutations (cont.)
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 neurodevelopmental 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 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.

Exhibitors
Research and direct health care
The exhibitions relating to research and direct health care were mostly applicable to the US with material available about the Rett Syndrome Clinic at the University of Alabama (Birmingham), the Children's Hospital at Montefiore, (New York City), and the Blue Bird Circle Rett Centre at the Texas Children's Hospital (Houston). Members of the Australian Rett Syndrome Study manned a table and were always present to assist with enquiries, not only about the Study but also InterRett.

Tribute reception
On the Saturday evening in the main auditorium, a large number of Congress attendees paid tribute to those with Rett syndrome. An event of this nature is a regular feature of the annual IRSF conference. The speakers included Martine Gaudy (Secretary, Rett Syndrome Europe) who was the only non-American to address the reception.

The reception also provided an opportunity for some of those 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

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, cost and/or taking time off work.

Other Australians attending were, 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. They are all involved in Rett syndrome research and deserve a pat on the back for their successful contributions to the conference.

Session DVDs
As has been already often been referred to, selected Congress presentations are available for purchase from its web site *worldcongress.rettsyndrome.org* in sets of 2, 3 or 4 sessions, ranging in cost from US$20 to US$40 per set. At present, there were 15 of a possible 35 presentations available from this source. Given the confidentiality restrictions that applied to the scientific programme, none of those sessions are available.

Kathy Hunter
Kathy Hunter, founder of the former International Rett Syndrome Association in the early 1980s and its President until 2007 when it was absorbed to became the International Rett Syndrome Foundation, and author of 'The Rett Syndrome Handbook', has done more for the disorder than anyone else on the planet.

Bill's trip to America would not have been complete without contacting her. She did not attend the World Congress (first one that she has not been present at) so when he reached Washington DC, he rang her. As a result, Kathy, her husband Scott, and their 37 year old

Rett syndrome conferences
Kathy Hunter (cont.)
Rett syndrome daughter Stacie, came in from nearby Maryland and spent an afternoon with Bill and his wife, Rosalie. The main topic of conversation was the New Orleans conference.

Meeting of representatives from Rett Associations
At the conclusion of the congress, a meeting was held for those attendees representing Rett syndrome Associations from overseas and the US.

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).

Thomas Bertrand (Rett Syndrome Europe) informed the meeting about RareConnect, a web site 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 web site address is w*ww.rareconnect.org*

Future World Congresses
The 7th World Congress on Rett Syndrome was the third such event attended by RSAA President Bill Callaghan, having previously represented the Association at those held in Belgium (1993) and Sweden (1996).

RSAA is grateful that IRSF was prepared to take on the task of organising this Congress and, with the help of sponsors, the funding of it. Aside from the financing, it involved arranging the venue, programme content, carers, speakers, printing, information packs, catering, promotion, registration and staffing. However, when compared to the previous Congresses that Bill attended, he felt that, despite the enormous effort that went into staging it, the Congress in New Orleans lacked a significant international focus.

Very few attendees (40 out of 450) came from outside the US. Of the 37 speakers in the family programme, 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.

Opting for New Orleans as the host city may have contributed to the lack of overseas interest, given it is hot and humid there in late June, its history of hurricanes (in fact, one formed off the coast while the Congress was in progress but headed to Florida and not Louisiana), and the extra travel and cost after reaching America.

If a time comes when serious consideration is given by RSAA or another Australian body to hosting a World Congress, then thought should be given to whether or not those from overseas will come, given the distance to Australia, and the time and costs involved.

Bill also advised RSAA that the uniqueness of the World Congresses that he had been to previously, was having scientist and family together at each presentation. This didn't happen in New Orleans as the programme was split into, family and science. The sad thing about the latter was that what was said remains secret, as attendees were required to sign a document swearing to treat what they heard or saw presented, as confidential, which to Bill's knowledge, is a first for a Rett syndrome World Congress. Thus, a third of what was presented in New Orleans cannot be reported upon.

8th World Congress on Rett Syndrome
The January 2013 edition of the Belgian Rett Syndrome Association newsletter reports that the next World Congress will be held in Moscow in 2016.

3rd European Rett Syndrome Congress
'Research Update and Preventative Management' are the themes for the 3rd European Rett Syndrome Congress which is to be held in Maastricht, The Netherlands, on 17-19 October this year.