

RETT syndrome IRELAND

Charlotte O'Brien with her Dad at the Dublin Presentation



RETT SYNDROME ASSOCIATION OF IRELAND

We are a group of families who get together to share information and offer support to each other. We are mostly parents, who meet three times a year. We keep in touch by circulating our newsletter.

Dublin Presentation Success

Almost 75 people, including 19 girls with Rett Syndrome, attended the Presentation on Brainstem Dysfunction at Crumlin Hospital on February 22 & 23 last. With more than 25 families represented, the event was pronounced a great success by all who were there.

The Presentation, by Dr. Peter Julu and his team of Doctors, ran over both days. It included assessments of some of the girls, plus excellent talks by both Dr. Julu and Dr. Alison Kerr. (More details on both of these talks inside the newsletter.)

Dr. Julu used the Neuroscope, which he has invented, to assess some of the girls, and explained in easy to understand terms the breathing abnormalities that are part of Rett, and how they can be confused with epilepsy. Dr. Kerr spoke about the recent research developments in Rett, especially their recent advances by Dr. Adrian Byrd. There was also a lively question and answer session.

The event was hosted by the Neurophysiology Department at Crumlin Hospital, where Dr. David Webb offered the new Conference Facilities at the hospital, and was personally very helpful and 'hands-on' over the two days. The entire staff made great efforts to ensure that the families were made welcome and comfortable, with Ann Coughlan, Head Neurophysiologist, Dr. Webb's Secretary, Helen Flanagan and Ray Coughlan the head EEG technician working behind the scenes to make the event a success. Even the food was wonderful!

Breathing Patterns in Rett

Dr. Julu's Dublin Presentation

At the recent presentation in Dublin, Dr. Peter Julu and his colleagues, Dr. Stig Hansen and Dr. Flora Apartopolous,, demonstrated the Neuroscope and it's use in RS. It has long been understood that many of the problems experienced by girls with RS arise from abnormal brainstem function. The brainstem is responsible for heart function, breathing and digestion among other things. Over the course of the two days, Dr. Julu's team assessed seven girls. Because of the enormous demand - enough to fill the weekend 3 times over - the girls to be assessed were chosen by drawing names from a hat. Dr. Julu also demonstrated the procedure and explained brainstem dysfunction in very clear terms for the families.

The assessment is non-invasive and similar to an EEG. While the assessment was taking place, it was possible to see all the results together on a screen, through the use of a computer

Breathing abnormalities are thought to be present in all girls with RS even if they are not obvious, and can even be mistaken for epilepsy. Dr. Julu described 4 main categories of breathing abnormality in RS, and each of these categories has several sub-types of breathing, involving thirteen different types of breathing in all. The main categories are:

- Apneustic breathing
- Forceful breathing
- Valsalva's breathing and
- Feeble breathing

Girls with Rett Syndrome usually show an abnormality from one Category, but can also show several sub-types.

Dr. Julu was in contact with the Association at the beginning of April to let us know that he is still analyzing the Neuroscope tests carried out in February and that he will be forwarding the results once they have been completed.

Dr. Peter Julu, Maria Keegan, Nurse Specialist and Dr. David Webb, Crumlin Hospital at the Neuroscope demonstration in Dublin



Right: Sarah Keogh is assessed with the Neuroscope

Below Right: Lauren Rankin at the Dublin Presentation



stop and there may be symptoms like epileptic stiffness of the limbs. There may then be brainstem shutdown (the skin turns white and the body becomes floppy). Others may turn blue in the face, starting with the lips. This breathing category does not respond to drug treatment.

Valsalva's type of breathing

This is forceful breathing leading to an increase of pressure in the lungs and the chest. This reduces blood flowing back into the heart and brainstem reacts very violently. This kind of breathing can result in unsteadiness, dizziness and wandering eyeballs. Blood pressure can be affected too. Sometimes the symptoms can be confused with epilepsy. This breathing category does not respond to drug treatment.

Feeble Breathing

This happens with girls who have the habit of very shallow breathing. The movements of the chest and the abdomen are so small that it's easy to think that she has stopped breathing altogether. Sometimes the person does stop breathing briefly. The Rett person can turn blue in the face (starting with the lips). Feeble breathers may also have "Brainstem Storms". Whether this type of breathing responds to drug treatment is an area of current research.

If you would like more information on breathing abnormalities in Rett Syndrome and the use of the Neuroscope, the company that produces the Neuroscope has a website that explains it with video clips giving examples of the different breathing patterns. You can see it at www.medifitinstruments.com.

Apneustic Breathing

This involves breathing in but failing to breathe out regularly. There are three types of breathing abnormalities in this category. They are long breath holds, repeated short breath holds and extended inhaling. This breathing category responds to drug treatment.

Forceful Breathing

This occurs when there is forceful inhaling and exhaling of air. Breathing may



The Association hopes to be involved in further neuroscope assessments, so anyone thinking of going abroad to have their child assessed should get in contact with us in advance in case there are any further developments in the meantime.



Above:: Laura Dowling at the Presentation in Dublin

Left:: Dr. Alison Kerr

Dr. Alison Kerr answers questions on Major breakthrough in Retts Research

Dr. Alison Kerr, paediatric neurologist, and long time expert in the area of RS very kindly agreed to attend on the Saturday. Dr. Kerr has been a consultant to the Rett Syndrome in the UK for years and has worked with hundreds of girls with RS. As she is now retired and making an attempt to do less work, she did not give a formal presentation. However, she spoke about the recent breakthrough in the area of genetic research in RS.

In early February of this year, Dr. Adrian Byrd, who conducts research into the genetic aspects of RS in Edinburgh, made a major breakthrough, which made headlines throughout the United States and the Great Britain. Dr. Byrd explains RS as coming about as a result of one of the X chromosomes being 'switched off' leading to the symptoms we see in our girls. In his work with laboratory mice, he succeeded in switching the X chromosome back on in mice that had been genetically

engineered to have Rett Syndrome. The results were quite dramatic in that most of the Rett symptoms disappeared even in the older subjects.

Dr. Kerr emphasised that while this is a major breakthrough, it could take a long time before the success of this research can be reproduced in humans. She also stressed that while it promised progress with the physical aspects of RS, we simply did not know what effect, if any, it might have on learning disability.

Dr. Kerr also did a Question and Answer session on RS in general. We were delighted that she agreed to attend.



Left:: Clara Barry at the Presentation in Dublin

Budget 2007 Health Service Funding for Disabilities

In the budget for 2007 €41 million was announced to fund disability services. This is to include 255 new residential places, 85 new respite places and 535 new day places. From funding that was made available in 2006 193 new residential places, 66 respite places and 433 day places were created. 112 residential places, 9 respite places and 35 day places were upgraded. The umbrella organisation Inclusion Ireland has expressed concern that this funding was decided in terms of places rather than being based on individual need. They believe that funding for services for disabled people should be planned according to waiting list information and the precise need of the individual.

€15 million is being allocated to the introduction of the second part of the Disability Act which involves the Assessment of Need process for the under 5s. A further sum of €5 is being allocated to voluntary service providers to go towards core funding deficits.

Disability Allowance

The Department of Social and Family Affairs has announced that the Disability Allowance will be increased by €20 to €185 a week, from January 2007. This will now also extend to those living in residential institutions. Previously, the first €20,000 of a person's capital was means tested for this allowance. This has now been extended to €50,000.

Carers Allowance

From September 2007 a "half rate carers' allowance" is available to those already entitled to a Carers as well as another social welfare benefit. Up to now carers were only entitled to one social welfare benefit and for this reason often decided not to avail of the carers' allowance in order to receive a more beneficial allowance. For those under 66 the allowance will increase by €20 to €200 a week and for those over 66 by €18 to €218.

Tax Credits

From 2007 the tax credit for a disabled child will be increased from €1,500 to €3,000.

Respite Care Grant

From June 2007 the Respite Care Grant will increase from €1,200 to €1,500.

Bereavement grant

From May 2007 families may now claim a bereavement grant in the event of a death a disabled family member. This was not available previously on the grounds that the person had not paid PRSI contributions due to not being in employment. The grant has also been increased from €635 to €850.

Helping Out



Above:: Angela Dowling at the Presentation in Dublin

Tax relief on donations

Any PAYE taxpayer making a donation of €250 or more can increase the value of their donation by up to 72% by completing a tax relief for donations to eligible charities and approved bodies. This means that when donating €250 this becomes worth €181 extra. Unfortunately, fund-raised events don't qualify. In the event that we receive a personal donation of €250 and greater we will contact you with the relevant form.

Ireland signs UN Convention Rights for Persons with Disabilities

‘The existing human rights system was meant to promote and protect the rights of persons with disabilities, but the existing standards and mechanisms have in fact failed to provide adequate protection to the specific cases of persons with disabilities. It is clearly time for the UN to remedy this shortcoming’

(United Nations Human Rights Commissioner, Louise Arbor)

From the 30th of March 2007 the Convention was signed by all participating States including Ireland. This means that Ireland has agreed to ‘engage itself in carrying out laws, policies and administrative measures to secure the rights of disabled people. This includes that the ‘physical and mental integrity of persons with disabilities be protected’.

It also includes a requirement for Ireland to ‘identify and eliminate obstacles and barriers to ensure that disabled people can access their environment, transportation, public facilities, services, information and communications technology’.

It also refers to personal mobility to be fostered by facilitating affordable personal mobility, training in mobility skills, access to mobility aids, devices, assistive technologies and live assistance’.



Countries are also required to recognize the right to an adequate standard of living, services and assistance for disability related. Ireland’s policy on social inclusion recognises that disability can result in disadvantage and poorer standards of living and the National Action Plan for Social Inclusion advocates providing assistance to those living with disability as a result of disadvantage suffered by disabled people and their families.

‘Persons with disabilities have the right to the highest attainable standard of health without discrimination on the basis of disability’. The Convention also states that ‘to enable persons with disability to attain maximum ability, participating countries are to provide comprehensive habilitation and rehabilitation services in the areas of health and education’ Disappointingly, Ireland did not sign the Optional Protocol which allows an individual, or group of individuals to make a communication to the UN in the event that there is the question of such a group considering itself to be the subject of a violation of the Convention by the relevant member State.

Above Right::
Teresa Davoren at the
Presentation
in Dublin

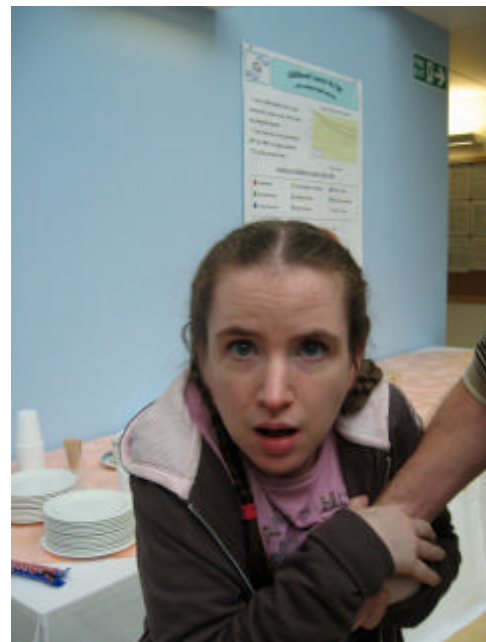
Right: Rachel
Horan at the
Presentation
in Dublin



More pictures of our girls



Clockwise from top left:
Laura Finucane, Ciara O'Loughlin, Aveen Cassin and
Emma O'Donoghue.



You can contact us at
The Rett Syndrome Association of Ireland
c/o The Bishops Palace
Church St.,
Limerick

Remember to let us know if your address or contact details have changed!

Fund Raising for the Association Chilly Swim!



On a very cold Christmas morning last year my big brother Richard and my little brother Emmet and some friends did a swim for Rett Syndrome and raised €3,310. This is me with my two brothers and the cheque.

Olga Geoghegan

Application For Membership / Renewal of Membership

Name _____ Address _____

Contact No. _____ Email _____

Name of Child With Rett Syndrome _____

D.O.B _____ Your Relationship to the Child _____

If your relationship with the child is Professional, please tell us your occupation and where you're employed: _____

Subscription fee is €30. However, if you're unable to afford this, your membership is still welcome

www.rettsyndromeireland.com

You can also contact us at

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