

Rett Syndrome Ireland is Official GAA Charity 2012

We're delighted to announce that Rett Syndrome Ireland has been selected by the GAA as an Official Charity for 2012. This is a huge boost for the Association, sufferers and families throughout the country. Being an Official Charity means a substantial donation to the work of the Association as well as support from the GAA in raising awareness of Rett Syndrome and of the work of the Association.

During a small ceremony at Croke Park, GAA headquarters, a cheque for €50,000 was presented to Andrew Kehoe, Chair, RSAI and Declan McPhillips, of the Association.

Uachtarán Chumann Lúthchleas Criostóir Ó Cuana wished the charity every success in its vital work over the next year. He went on to say, "The GAA is glad to be able to help what is a most worthy cause for 2012 and in addition to the tangible benefits of being afforded the status of an official GAA charity, we also hope to be able to work with the Rett Syndrome Association to raise the profile of both the condition

and the excellent work of the charity."

Andrew Kehoe, Chairperson of RSAI said "On behalf of the Association Ireland and the families we represent I would like to express our deepest gratitude to Christy Cooney, Paraic Duffy and the GAA for their extraordinary generosity.

"RSAI is a voluntary organisation and registered charity which relies solely on private donations and fundraising. This will go a long way in helping us to support families affected by Rett Syndrome, to help raise

awareness in general and to assist with on-going research."

Special thanks are also due to Declan McPhillips for all his great work on behalf of the Association to make this support possible.



Uachtarán Chumann Lúthchleas Gael Cristóir Ó Cuana and, Ard Stiurthoir Paraic Duffy at Croke Park with Declan McPhillips and Andrew Kehoe, Rett Syndrome Ireland. Photo: Brian Lawless / SPORTSFILE



Pictured outside Dail Eireann are Paula Connolly, Declan McPhillips, Andrew Kehoe, Stephen Tighe, Jerry Buttimer, TD and Alan Connolly

Rett Syndrome Ireland at the Dail

On International Rare Diseases Day (February 29, 2012) the Rett Syndrome Association of Ireland was invited to present to the Joint Oireachtas Committee on Health and Children, along with other organisations working in the field of Rare Diseases. It was an opportunity to explain to law and policy makers the realities of living with Rett Syndrome in Ireland today.

Below are the remarks made by Chairperson Andrew Kehoe for the occasion.

Andrew began by outlining the nature of Rett and how it affects the girls and their families. He explained the genetic component in the Syndrome, the stages and the various symptoms that can affect the girls. He stressed that even in its mildest form the girls are left profoundly and multiply disabled requiring full time care 24/7 365 days a year for the duration of their lives. He also pointed out that while the prognosis is not fully known and can vary - but there are recorded cases of women in their seventies in Sweden and U.S - the oldest woman RSAI is aware of in Ireland is 42

Despite the myriad of disabilities that come with Rett, Andrew emphasised that the girls are still capable of lifelong learning and of having happy and fulfilled lives.

The main part of Andrew's talk was to describe some of the many problems that we face each day of our lives:

- Length of time it can take to get the first appointment with a Neurologist when initial signs of regression appear - It can take anything from 6 to 12 months.
- The gap between appointments thereafter is too long prolonging the length of time it takes to get a correct diagnosis.
- Misdiagnosis is common and can prolong this process even further
- At diagnosis stage many families are given little or no information (and sometimes incorrect information) about the condition itself but also about how to go about accessing appropriate services.
- Accessing appropriate and adequate services from HSE becomes a constant battle for all families to some extent but can vary dramatically based on geographical location. This includes difficulties in getting everything from incontinence pads to therapies, mobility equipment, communication aids and respite services to name but a few.

- Accessing appropriate school placements and SNA allocation from Dept. of Education is a constant struggle for families and again varies geographically.

- Qualifying for Domiciliary Care allowance, Carers allowance and medical cards is an on-going issue - for those who do receive these payments and rely on them as their sole source of income, they are totally inadequate.

- Respite services are totally inadequate and in many cases not available at all.

- If you do not look after the Carers, the state could end up looking after their relatives full time at a much higher cost.

- Inadequate facilities in our hospitals to cater for the needs of disabled people - e.g. hoists for lifting, showing tables and changing areas.

- Waiting times for critical operations e.g. scoliosis - some girls have been waiting up to 2 years while in constant pain and at risk of death in some cases.

- Lack of service provision for girls once they reach 18 - school leaving age. Many end up at home full time until such time as their parents/family can no longer look after them - this is not acceptable.

- No independent inspection or regulation of service providers to the intellectually disabled - this is of grave concern to all parents but particularly when a child/adult has no means of communication.

All of the above problems (and the list is not exhaustive) are experienced by the majority of families on an on-going basis throughout the lives of their disabled relative and compound what is already a very stressful and pressurised situation - that of looking after a totally dependant person full time for all of their life.

As yet there is no cure for Rett Syndrome, but the above additional issues/problems could be cured if there was a real will on behalf of politicians and Government to do so.

At the height of the boom when there was plenty of money around, families still had to fight for all of the above services - what chance have they now unless the rights and dignity of disabled people are finally put before budgets and resource allocations.

Why has Ireland still not ratified the UN Convention on the Rights of Persons with Disabilities which it signed up to in 2007?

As the parents of disabled children we can only draw one conclusion - it is not a priority



Pictured at the recent *Talk Sport for Shauna* event were Peadar Mc Mahon (chairman organising committee) Joe Brolly (Sunday Game) Declan McPhillips, Mick Quinn (Irish Rugby player), John Delaney (CEO FAI) Paraic Duffy (Director General GAA), Jarlaith Burns (BBC Sport /TNAG)

With them are Sarah Kehoe, Shauna McPhillips and Alex Connolly

Mother's Day Out

Next June 15/16 sees a new opportunity for meeting and sharing with others who are also affected by Rett Syndrome as Carers or Family Members. The Mother's Weekend is at the Sheraton Hotel in Athlone. Organised by Committee Members Cathy Cleary and Paula Connolly and it's an opportunity for the Mum's to get together in a casual and informal way - and to get a well-earned break.

Any time that the Members get together then there is a chance for us to learn from each other. The Association is delighted to support the initiative.

Rett Syndrome Research Trust – Donation

We are delighted to announce that this year Rett Syndrome Ireland has made a donation of €4,500 to the Rett Syndrome Research Trust UK (www.reverse Rett.org.uk) to aid in research into Rett Syndrome. To date RSRT UK has funded 12 different research projects into the primary approaches to reversing Rett.

Access & Mobility 2012

The Annual Access and Mobility exhibition is on at the CityWest Convention Centre on May 25 and 26. The event showcases products and services for people with mobility issues - everything from Chairs to Stairlifts and bathroom aids and appliances. Admission is free. More on www.accessandmobility.ie

World Rett Syndrome Congress

The 7th World Rett Syndrome Congress is on in New Orleans, June 22-26. We have no one going this year but we'll be reading up on it online at worldcongress.rett syndrome.org

Congratulations

Congratulations are due to Deirdre Horan who has been named as Carer of the Year for the Munster area by the Carer's Association. Deirdre will be off to Dublin for the National Final at the end of the month.

Carer's Association

The Carer's Association website is well worth a look at www.carersireland.com. It's full of good advice and information on all aspects of being a Family Carer. They have a FreeFone line and drop in centers all across the country.

THE RETT SYNDROME ASSOCIATION OF IRELAND

PATRONS:

Paraic Duffy, Director General,
GAA

Miriam O'Callaghan, Broadcaster

Dr. David Webb, Consultant
Paediatric Neurologist

We are an association of parents and families of people with Rett Syndrome. We came together to provide support and information:

to each other

to parents and families who have received a new diagnosis

to raise awareness among medical professionals and others who work with people who have Rett Syndrome

to raise public awareness of the condition and its effects on those affected by it.

to encourage and assist research

Contact Us:

by Post:
% St. Gabriel's Centre,
Springfield Drive, Dooradoyle
Limerick

e-mail: info@rettsyndrome.ie

Web: www.rett syndrome.ie

by Phone: 087 6152183 / 087
6061993

The Irish Rett Syndrome Association is a registered Charity: Chy 16340

About CDKL5

CDKL5 is a rare X-linked genetic disorder that results in early onset, difficult to control seizures, and severe neuro-developmental impairment. There are fewer than 200 cases worldwide, however, more and more children are being diagnosed as awareness of CDKL5 spreads.

CDKL5 stands for cyclin-dependent kinase-like 5, and is located on the X chromosome. The X chromosome is one of the sex chromosomes; females have two X's and males have one X and one Y chromosome. The letters are an abbreviation of the scientific name of the gene which describes what it does.

The CDKL5 gene provides instructions for making a protein that is essential for normal brain development. Although little is known about the protein's function, it may play a role in regulating the activity of other genes, including the MECP2 gene (of Rett Syndrome). The CDKL5 protein acts as a kinase - an enzyme that changes the activity of other proteins by adding oxygen and phosphate atoms (a phosphate group) at specific positions. Researchers have not yet determined which proteins are targeted by the CDKL5 protein.

Most children affected by CDKL5 suffer from seizures that begin in the first few months of life. Most cannot walk, talk or feed themselves, and many are confined wheelchairs, dependent on others for everything. Many also suffer with scoliosis, visual impairment, sensory issues and various gastrointestinal difficulties

CDKL5 mutations have been found in children diagnosed with Infantile Spasms, Rett Syndrome, and Autism among others. However, it is important to note that scientists and doctors do not know what causes CDKL5 mutations, or the full spectrum of CDKL5 disorders at this time. It is likely that there are many people affected by CDKL5 who have mild symptoms and no seizures. With continued research and awareness of CDKL5, we hope to build a more comprehensive understanding of the spectrum of this disorder, and begin the search for a desperately needed cure.

Because of the connection with Rett Syndrome the Association is delighted to include people with CDKL5 in our activities.

Wills and Trusts – a Reminder

A couple of years ago John Costello, a consultant with Beauchamps Solicitors came to talk to us at the Family Day. He gave an entertaining and informative chat on the subject of Wills, and planning for the long term for our disabled children.

It's not always easy to think that far ahead, but it's worth reminding us of some of the things that he advised.

John emphasised that it's very important to make a Will, and also gave us some pointers on some of the key issues. For instance, if you leave money or property to a disabled person, that person will have *means*, and could then be *Means-tested*, and so lose out of benefits and services. So he suggested a way to leave resources for a Disabled Relative, while not affecting their means.

He introduced us to the Concept of **Discretionary Trusts**. Under this system you would will your estate to Trust, run by a couple of relatives or friends. They would then disburse the funds to the Disabled Person as needed for any matters that might arise. The advantage to this system is that the Trust does not affect the disabled person's means, and therefore their entitlement to services and support from the State.

John also pointed out that the requirements for a person in full-time care can be quite small, so that was another thing to take into account when making your decisions.



RTE Presenter Marty Morrissey at the recent *Talk Sport for Shauna* event.

Rett Syndrome Clinics

The Association has a long-standing ambition to develop Clinics where Professionals with special knowledge of Rett can review your daughter and make suggestions for her further care and development. The U.K. association has been running these Clinics for several years now with some success. They have agreed to help us by mentoring a team here. Now we need to create that team, and we need your help to do it.

We're looking for suggestions of people to approach to join the Irish team. We need a

- Paediatrician,
- Neurologist,
- Physio, a
- Speech and Language therapist and
- someone with expertise in Learning Disability in Adults.

Is there someone in your daughter's care team that you would recommend in one of these fields? Someone you think is interested and committed? If so, drop us a quick email on info@rettsyndrome, or call on one of the numbers.

Changes at the Association

There have been some changes to the Committee of the Association in the past few months. Deirdre Horan has stepped aside after the death of her Husband Dave, Rachel's Dad. All of us in Rett Land send her our very best wishes.

Emmet Harten has stepped aside from the Committee as he is currently unable to make the time commitments to the work that he would like.

We all thank both of them for their efforts on behalf of the Association and wish them the best for the future.



GAA Stars pose with the Sam Maguire and the McCarthy Cups and some very important young ladies at the Family Day in Kilkenny last year.

This year the Family Day will be in Athlone. Details coming soon...