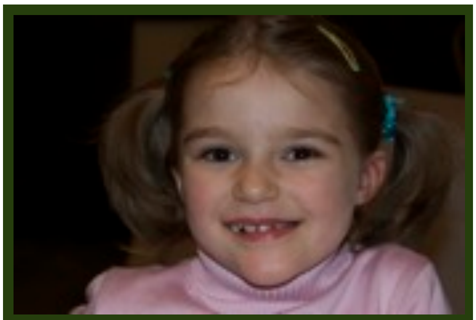
Rettsyndrome Ireland

newsletter - winter 2008

Happy Christmas and best wishes for the Year to all the Families and friends of the Association

Conference '08

Families, teachers and therapists gather in Limerick for a very special event



Ciara Barry from Waterford at the Conference in Limerick

The Association held it's Conference and Family day for 2009 at the Kilmurry Lodge Hotel in Limerick on October 31, last.

There were 150 people registered for the conference. Among those registered were 26 families, most of whom attended with their girls. It was great to see so many of the girls at the event. The other delegates included teachers, special needs assistants, music therapists and music therapy students, occupational therapists, speech therapists, nurses, psychologists, school principals and service managers, physiotherapists and physiotherapy students among others.

As it was Halloween many of the girls turned up in their Halloween costumes and they were really cute. Angie Fitzsimons did a brilliant job of decorating the family room with Halloween decorations and treats for the girls and St. Gabriel's Centre in Limerick supplied Bean Bags and mats for the girls to relax on during the day if they got tired during the conference. Accommodation was provided by the Association on the Thursday and Friday for those families who needed it.

Presentations were made by Meir Lotan, Pyhsiotherapist (Israel), Cochavit Elefant, Music therapist (Israel) and Kathy Hunter (United States) on aspects of caring for someone with Rett Syndrome.

THE RETT SYNDROME ASSOCIATION OF IRELAND

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 it's effects on those affected by it.

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

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Meir Lotan receives a gift from Sarah Kehoe (Dublin) at the Conference in Limerick

Conference Report

__Meir Lotan__, Physiotherapist with the Israeli Rett Syndrome Evaluation Team and __Cochavit Elefant__, Music Therapist, Associate Professor of Music Therapy at the Grieg Academy, Norway, __Kathy Hunter__, founder, International Rett Syndrome Association all spoke at the conference

The main focus of Meir's presentation was the work that he has done with girls with scoliosis. He demonstrated that even in a case of advanced scoliosis that where intensive physiotherapy was available that he was able to halt and even reverse the degree of spinal curvature. However, he emphasized that once the programme was stopped the progress also stopped and the scoliosis progressed.

Cochavit's presentation showed some of her work with girls who were able to communicate using picture and word cards. In some cases the girls were able to communicate at a very sophisticated level, one Israeli girl even managing to communicate in English as a second language. Some of the most powerful video pieces that Cochavit played involved girls making simple yes and no choices using communication cards. She demonstrated how music can be used to engage the girls and prepare them for other activities such as physiotherapy or communication.

Kathy Hunter, Founder and past President of the International Rett Syndrome Association and parent of an adult daughter with RS, attended as a surprise special guest speaker. Kathy gave a short presentation entitled 'Welcome to the Club you never wanted to join' and spoke about the realities of being a parent of a child with Rett Syndrome. She also spoke later in the day about the genetic aspects of RS and also the gastro-intestinal problems a lot of the girls deal with and the importance of the appropriate medical management of these.

At the end of the day, all of the speakers formed a panel and answered questions from the audience. The speakers were also available to speak with parents and answer questions over the course of the day and after the conference.

Cochavit Elefant receives a gift from Emma O'Donoghue (Dublin) at the Conference in Limerick

6th World RS Conference in Paris

The 6th World congress of Rett Syndrome took place in Paris on the weekend of October the 10th last. Most experts working in the field of Rett Syndrome were there presenting their work as well as representatives from Rett Syndrome Parents Associations.

(above) Huda Zoghbi, Baylor College of Medicine, Texas Professor, Departments of Molecular and Human Genetics, Pediatrics, Neurology, and Neuroscience, was the first to identify mutations in the MECP2 gene as the cause of Rett Syndrome.

(below) Adrian Bird, Ph.D., of the University of Edinburgh and Chairman of the RSRF Scientific Advisory Board who was responsible for the landmark study reversing the symptoms of Rett Syndrome (RTT) in a genetic mouse model.

Professor Alan Percy, Paediatrician and Neurologist, opened the conference with an introduction to recent developments in Rett Syndrome. He confirmed that although presence of a mutation in the MECP2 gene confirms the clinical diagnosis of RS, the condition remains a clinically rather than a genetically defined condition. 95% of those with classic RS have the MECP2 mutation. MECP2 mutations may also be seen in children who do not have classic Rett on clinical examination. He also spoke about the most common management problems in RS. These include seizures, in 20% up to 75%, although this is considered to be a over estimate; constipation a problem for 70-85%; bruxism (teeth-grinding) 75-85% at some point in time; drooling 72%; gastro-oesophageal reflux occurring in up to 55% often causing problems with night time awakening. Ineffective chewing and swallowing is also identified as a problem in many. Recently there have been difficulties found in gall bladder function with many girls showing evidence of gall bladder disease, even at an early age. By the age 16 up to 80% will have some degree of scoliosis, approximately 12% requiring surgery. Osteopenia, or reduced mineralization of the bones increases to 40% once the girls are over the age of 30 and bone fractures are common. There is also evidence to show that fracture rates are four times higher in RS than in the general population due to reduced bone density and that remedial measures should be taken, with young girls as well as older.

Dr. Helen Leonard from Interrett spoke about the more unusual forms of RS e.g. preserved speech variant, atypical and late onset. Research from the Australian database classify Rett Syndrome into 26% as ‘more severe’, 56% ‘classical’ and 18% ‘milder’. The diagnostic criteria have now been relaxed to allow children who have had an absence of normal development in the first six months and an absence of head circumference deceleration to also be included. Dr. Leonard gave a presentation on the eight most common mutations. These are: R106W, R133C, R168X, R255X, R2770X, R249X, R309C and T158M.These eight mutations account for approximately 65% of all of those who receive a genetic diagnosis of RS.

Dr. David Roye, Chief Orthopaedic Surgeon at the Paed. Orthopaedics Department of Columbia University Hospital NY gave a presentation on the clinical management of scoliosis. He was joined by Meir Lotan, Physiotherapist. He identified those most at risk as having low muscle tone and never learning to walk. Certain mutations may have a protective factor against developing scoliosis. Regular monitoring before the development of scoliosis is recommended, optimising weight bearing and physical activity opportunities and supported seating are recommended. Surgery is generally recommended in

(left) Robyn Dagg from Co.Wicklow with her Dad Trevor at the Conference in Limerick

(below) Anna Boland from Kilkenny was there too

curves above 40 degrees. Spinal surgery is appropriate for the management of more severe curves and is usually associated with positive physical and functional outcomes. However the decision to operate must take a number of factors into consideration, including bone maturity and general health. Posterior surgery is now recommended in preference to anterior surgery and 'growing systems' are seen as preferable for children under age 8. As this is a complex surgery, Rett Syndrome being a neurodevelopmental disorder, Dr. Roye recommends that surgery is carried out at a hospital where there is specialist anaesthetics and post-operative intensive care available. According to Occupational Therapist Eva-Lena Larsson who works with a spine surgery unit in Sweden the girls generally can sit on the edge of the bed on the day following surgery, and then stand and sit in their own chairs a few days after surgery.

Dr. Alison Kerr, specialist Paediatrician and author in the field of RS, who many of the families will remember from our Neuroscope workshop with Dr. Julu in 2007, introduced a session on sleep and behavioural difficulties. According to Dr. Kerr, approximately 85% of the girls experience agitation on occasion, 67% sadness, 33% have been found to self injure. Sleep disturbance occurs in 73%. Medication should only be used as a last resort. Management of these behaviours should look to ensuring that all possible medical problems are investigated; that the girls are physically comfortable, that they are kept sufficiently active, interested and socially engaged. These problems are believed to decrease with age.

Sleep problems are common, with many girls experiencing night-time awakenings often with spells of laughing. Increased day time sleeping seems to be a issue for many as they get older and can interfere with daytime activities. Judy Wine, Speech Therapist from the Israeli RS Center gave a presentation on communication. She says that clinical experience has shown that girls with RS have a rich inner world. Repeatedly, people caring for and working with these girls say that they give clues that they understand the world around them. They have strong emotional responses to situations and will choose to participate depending on their perceptions and relationships with the persons involved. The girls fundamental physical disabilities combined with a delayed reaction time of up to two minutes on occasion, interfere with their ability to indicate what they understand. This can lead those not familiar with the girls to believe that their ability to understand is less. Augmentative and Assistive Communication (AAC) is seen as essential for RS girls. It provides the tools to enable social participation, interpersonal interaction, language and cognitive development, development of feelings of self worth and confidence, increased motivation and reduced frustration. AAC strategies can range from picture cards to computers and other communication devices.

Nigel Livingston from CanAssist at the University of Victoria, BC gave a presentation on some devices developed by his department to help people with special needs to communicate. One of their main devices involves an eye-tracking system, involving an infrared camera and LEDs to detect movement of the pupil of the eye which is then displayed on a computer screen. These devices can also be used to activate toys. Some of the research at the University has involved helping girls make choices using these devices. More information on the devices can be seen at their website http://www.uvatt.org/

For more papers from the Congress, there's a link on our website.

We would like to thank Fr. Patrick Fitzgerald of the Parish of St. Joseph's, Paris for his hospitality in accommodating committee members who attended the congress.

New Members for Association Committee

The Annual General Meeting of the Rett Syndrome Association was held at the end of the Family Day Conference in Limerick on October the 31st. We are pleased to welcome two new members, Nathalie Shaw from Dublin and Kevin Barry from Waterford.

The committee for the next year is Angie Fitzsimons, Treasurer, Kevin Barry is Secretary and Noreen Harrington is Chairperson. Other committee members are Joe Lawlor, Deirdre Horan and Susan Boland. We are grateful to all members for staying on the Committee for another year.

Thank You

Andrew Kehoe has stepped down from the committee after four years as Secretary. Andrew manned one of our phones, organised printing and was responsible for all the mailings and many other duties in his time on the committee. Also stepping down is Neil O'Donoghue. We thank them both and wish them all the best.

Rett Syndrome Europe (RSE)

The RS Association of Ireland is now a member of the Rett Syndrome Europe network. The purpose of the network is to represent the interest of people with Rett syndrome and their families, especially in the following areas:

- To make Rett syndrome better known to the public, professionals, carers and those who are directly concerned in all European countries.
- To improve the communication within the European Rett community.
- To promote, as a representative European organisation, the interests of people with Rett syndrome and their families.
- To expand the RSE to all European countries and to assist, if necessary, in the creation of national associations.
- To promote research into Rett syndrome.

These aims are to be especially attained by:

- Co-operation with Rett syndrome associations within Europe and Worldwide.
- Co-operation with other relevant international and national institutions.
- Co-operation with other social groups.
- Exerting influence on decisions concerning legislation in relation to medical, health, and education in professional and social fields.

Membership is open to Rett syndrome associations from European States, as well as to Rett syndrome subgroups of umbrella organizations. Rett Syndrome Europe is a non-profit making organisation. Details relating to the organization may be seen at the RSE website: http://www.rettsyndrome.eu/index.html

Most European Rett Syndrome Associations are now members of the organization. The network meets once a year. As most of the representatives were attending the World Congress in Paris, the 2008 annual meeting of the RSE was held during the Congress proceedings.

(above) Robyn Scully (Co.Kildare)

(below) Shauna McPhillips (Co.Monaghan)

Christmas Cards

Following the great success of the Rett Syndrome Association Ireland Christmas cards, the Association have cards for sale again this year. Packs contain 12 cards and cost €7 plus €1.50 postage per pack. Contact the association for details. The e-mail address is on the front page.

Fundraising

Thank you to Nathalie Shaw, mother of Ellie, who together with Christina King and Liz Cloonan did the women's marathon earlier in the year. They raised €1875 for Rett Syndrome Ireland. Donations were also made to the Association of €100 by Janet Sutton, €170 by Martina Davoren, €170 by Niamh and Niall Rankin and €100 by Kathleen Burns. Thank you to all for your generosity.

www.retthelp.info

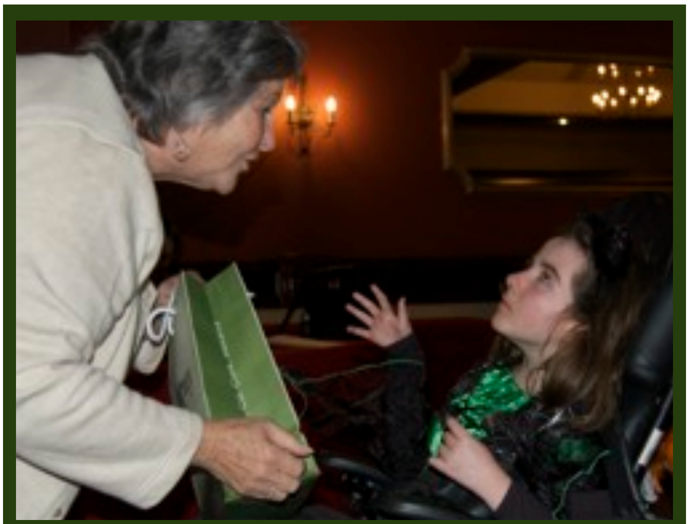
Kathy Hunter, Founder and President (retired) of the International Rett Syndrome Association, and author of the Rett Syndrome Handbook, has launched her own website at www.retthelp.info. Kathy will take questions there, and help if she can. If she can't help then she'll bring them to experts in the Rett Syndrome field on your behalf. Check it out.

NEUROSCOPE ASSESSMENTS

Following the success of the workshop presentation on autonomic dysfunction in Rett Syndrome and assessments using the Neuroscope in Spring 2007 at Our Lady's Hospital in Crumlin, Dr. Julu returned again to carry out two days of assessments on some of the girls. After the first assessments in 2007, the Association invited families to contact Dr. Webb at the Neurology department in Crumlin if they were interested in having their girls assessed if we were able to arrange for Dr. Julu to come back with his colleagues, Dr. Hansen and Dr. Apartopoulos, to do some further assessments. All of those who contacted Dr. Webb were approached by the Association to participate in the assessments which took place in November. Eight girls were assessed on the weekend of the 7th of November. All costs were paid by the Association. The Association would like to thank Dr. Webb, Consultant Neurologist, Ann Coughlan, Chief Neurophysiologist and the Neurology department for making their facilities available to the Association and Dr. Julu for the assessments.

InterRett

Interrett : the largest database on Rett Syndrome internationally is the Interrett with up to 1600 girls included to date. It is the database of the International Association and is based in Australia and headed up by Dr. Helen Leonard. Those families who gave their consent for contact for research purposes at the conference in October should expect a call from Dr. Leonard's team some time in the future to ask if they would like to take part in the Interrett research project. This involves completing a number of questionnaires, either online or a paper version on your daughter's condition and development to date.



Katie Devlin (Co. Clare) makes a presentation to Kathy Hunter at the Conference in Limerick

Inclusion Ireland: recent News:

Some comment from recent updates to the Inclusion Ireland website: www.inclusionireland.ie

Inclusion Ireland welcomes Social and Family Affairs Minister Mary Hanafin's decision not to implement Budget measures that would have raised the qualifying age for disability allowance from 16 to 18. Minister Hanafin said existing arrangements will continue pending a full review.

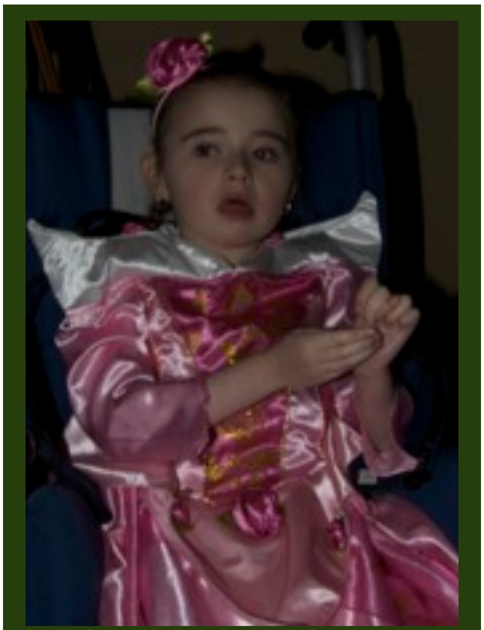
Inclusion Ireland CEO Deirdre Carroll says "the move to cut the Disability Allowance without putting in place an allowance to offset the true cost of disability was a retrograde step made in haste". Inclusion Ireland had a joint meeting with Down Syndrome Ireland and Minister Hanafin on Tuesday last (28th October).

"Careful analysis of the impact of increasing the qualifying age is needed before any changes to the Disability Allowance are made. Inclusion Ireland's budget submission called for a cost of disability payment, entitlement to a medical card for all children with a disability up to the age of 18, an increase in the domiciliary care allowance to €400 a month and an increase in the respite care grant to €3,000 per annum.

"These changes should have been introduced before the qualifying age for Disability Allowance was raised. Inclusion Ireland looks forward to working with Minister Hanafin on a review of the Disability Allowance, on behalf of the families we represent."

Standards In Residential Institutions

Inclusion Ireland will call for the implementation of standards in residential institutions for people with an intellectual disability when it presents to the Oireachtas Health Committee on Tuesday.



Ellie Shaw (Dublin) at the Conference in Limerick

Residential institutions for people with disabilities are not inspected and there are no national care standards applicable. In December 2007 a report was published into the abuse of adults and children with an intellectual disability in a service in Galway for 33 years. There are still no standards in place.

What do we mean by standards? For example, children without a disability who are in care have their homes independently inspected against national standards under the Children's Acts. Children with an intellectual disability have no such protection.

Jessica Flynn from Co. Cork at the Conference in Limerick

The Health Information Quality Authority (HIQA) has produced standards for residential settings for people with an intellectual disability, which are due to be implemented in early 2009. HIQA will also present to the Committee on Tuesday.

Inclusion Ireland is extremely worried that in light of a decision to postpone implementation of standards for services catering for older people, standards for services for people with an intellectual disability will now also be put on hold.

Inclusion Ireland CEO Deirdre Carroll will say on Tuesday that we have been waiting long enough for standards to come into place and we cannot wait any longer. Inclusion Ireland has been campaigning for over twenty years for standards to be implemented. Standards have now been developed and gone out for public consultation. To not implement them at this stage, would be a massive waste of taxpayer's money. To implement them on a voluntary basis would not be effective, and would ensure that the most vulnerable in our society remain at risk.

Delays in implementing special education legislation

Delays in implementing education legislation for children with special needs is a seriously retrograde step that will have a major impact on their development says Inclusion Ireland. Bigger class sizes will also mean less attention for children with special needs. Inclusion Ireland CEO Deirdre Carroll says:

"It is very regrettable that there will be further delays in rolling out the Education for Special Educational Needs Act 2004* (EPSEN). This is a very important piece of legislation that gives children with special needs the right to attend mainstream schools with appropriate supports. It is another part of the Government's much lauded Disability Strategy being put on the long finger. Inclusion Ireland has repeatedly called for funding for people with a disability to be ringfenced.

"EPSEN also allows for Individual Education Plans. Individual Education Plans allow for appropriate, focused education supports to be put in place. Children with special needs may regress significantly if intervention is not made from an early stage.

"Just 18 months ago in the 2007 Programme for Government we were told that EPSEN would be completely rolled out. Now we are being told that not only is EPSEN being halted, but class sizes are set to get bigger. Bigger class sizes mean less attention for children with special needs. Yet another retrograde step."

(above) Megan Cleary (Dublin)

(below) Ciara Collins (Co. Galway)

The National Council for Special Education set up by EPSEN, was charged with drawing up a plan for implementing EPSEN. This plan was sent to the Department of Education in 2006 and set out the resources needed, and a timetable for all sections of the Act to be completed by 2010. To date there has been no response from the Department of Education.

Women's Mini Marathon

I you feel like running for Rett the Flora Women's Mini Marathon is scheduled for June 1, next. You can raise money for the Association by participating in the Mini Marathon. You need to enter on an official entry form. You can get these in the Evening Herald every Thursday and Saturday from Feb 26th next, or online at www.florawomensminimarathon.ie from the same date.

Thanks to all those who helped organise and make our conference a success. A particular thanks to

Ben Devlin
Ronan McMahon
Keith O'Donoghue
Cian Callaghan
Ursula Callaghan
Rosemary Butler

Dick Whelan
St. Gabriels' Centre, Limerick
Simon Gilbertson (Dept. Of Music Therapy, UL)
Kilmurray Lodge Hotel
Angie Fitzsimons
All the Committee

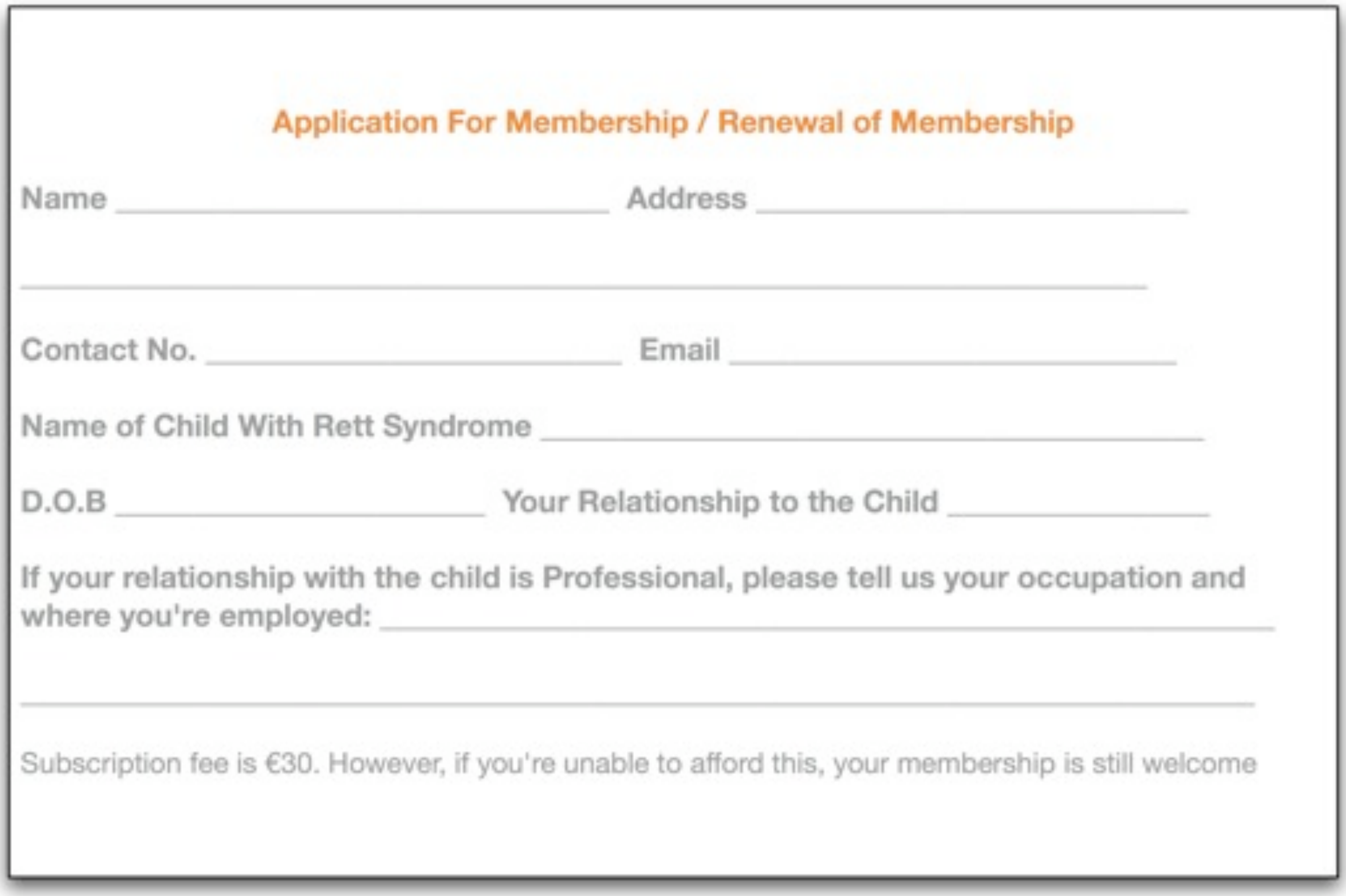
Please support Our Association

Membership costs €30, just fill in the form below and return it, along with your cheque to

The Irish Rett Syndrome Association
℅ the Bishop's Palace
Church St.,
King's Island, Limerick

If you cannot afford to pay, your membership is still very welcome, just fill out the form and send it in.

You can contact Angie on 087 6152183 or Noreen on 087 6152335



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: ____________________

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