Rettsyndrome Ireland
& CDKL5

Rett Syndrome Ireland is an association of families of children and adults with Rett syndrome living in Ireland (North & South).
It is a voluntary organisation and a registered Charity (CHY 16340)

OUR AIMS:

- To provide information and support to each other and to parents & families who have recently received a diagnosis.
- To provide information and raise awareness among service providers, medical practitioners and anyone working with children & adults who have Rett syndrome.
- To raise general public awareness of the condition
- To raise funds for research aimed at finding treatments and hopefully eventually a cure.

Please feel free to contact us by phone or email (details below) if you need any information , assistance or if you just need to talk to someone who can listen and understand – (particularly after diagnosis).

Further information links -

www.rettsyndrome.ie
www.rettuk.org
www.rettsyndrome.org
www.rsrt.org

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Registered in the Republic of Ireland No. 387789 Registered as a Charity: CHY 16340
Directors: Andrew Kehoe, Alan Connolly, Declan McPhilips

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Rett Syndrome Information Leaflet

ABOUT RETT SYNDROME

Rettsyndrome Ireland & CDKL5

What is Rett syndrome?

Rett syndrome is a rare complex genetic neurological disorder resulting in physical and intellectual disabilities. It is usually diagnosed in early childhood and predominantly affects females.
In most cases it is caused at time of conception by a spontaneous mutation of the MECP2 gene on the X chromosome and is rarely inherited.

How can Rett syndrome be recognised?

The age when first symptoms appear and the severity of these symptoms will vary from child to child but a common pattern is for the child to appear healthy at birth and then appear to develop normally for some time thereafter (6-18 months as a guide).
It is at this point that a slowing down or stagnation of development may be noticed, followed by a regression phase where skills attained up to that point are lost partially or completely, in particular communication skills and purposeful hand function. Other signs include stereotypical hand movements (mouthing or ringing of hands), seizure activity, irregular breathing, severe crying/screaming spells and general irritability.

How is Rett syndrome diagnosed?

Rett syndrome is diagnosed clinically by a Neurologist through observation of symptoms & patterns over time and then confirmed in most cases by a genetic test after all other potential conditions have been ruled out.
Diagnosis should be considered where post natal deceleration of head growth is observed.

Other common symptoms and complications -

1) Apraxia – inability or restriction in carrying out fine and gross motor movements.
2) Epilepsy – approx. 65% of patients will be affected at some stage
3) Breathing irregularities – (e.g. breath holding, air swallowing , hyperventilation)
4) Feeding difficulties resulting in poor weight gain – some patients may need to be tube fed.
5) Teeth grinding (Bruxism)
6) Abnormal muscle tone (rigid or floppy)
7) Scoliosis
8) Impaired sleep patterns
9) Small cold hands & feet
10) Abdominal distension
11) Osteoporosis is common
12) Constipation
13) Reflux
14) Trapped wind

As stated previously not all patients will experience all symptoms and the severity of symptoms will vary also.

Therapeutic Interventions -

There is currently no cure for Rett syndrome but the following therapies will be required to assist in the management of some of the symptoms.
1) Hydrotherapy – (swimming)
2) Physiotherapy
3) Occupational Therapy
4) Hippotherapy - (horse riding)
5) Speech & language Therapy
6) Music Therapy

A very useful reference document for parents and medical practitioners unfamiliar with Rett syndrome is the " National Best Practice – Management & Care Guidelines" published by Rett UK and available to download from their website www.rettuk.org