

Rett Syndrome

Rett Syndrome is a severe neurological condition which, it was thought, affected only girls. It is named after Dr Andreas Rett who first described the disorder in 1966. Further work in 1999 by Dr Huda Zoghbi and her colleagues found that the symptoms of Rett Syndrome are often the result of a mutation of a gene (MECP2) on the X chromosome. It is not usually passed down from parent to child: it can happen to anyone.

The affected gene produces too much of a binding protein, resulting in severe disabilities on several levels, including cognition, communication, social and motor skills. Genetic testing can now diagnose Rett Syndrome, and it has very recently been discovered in boys. It is predominantly a condition which affects females, however, and this information sheet has been written from that perspective. At present there is no treatment or cure, but advances in genetic manipulation may eventually provide answers.

It has been calculated that of all the diseases which may produce mental disabilities in women, Rett Syndrome is a very close second to Down's Syndrome. It is estimated that it occurs in approximately 1 out of every 10,000 female births (calculated in the USA).

At birth the baby girls appear normal, and are often unaffected until around the age of one year, when development slows. Further regression develops from around the ages of 1-3. It may be sudden or gradual, but parents and professionals will notice loss or withdrawal of skills which had been developing normally, such as **social skills, speech, and motor coordination.**

Parents may report that their little daughter is strangely placid and calm compared to their friends' daughters of the same age, and that she does not reach or grab for things she wants.

Early indications

Many of the main symptoms of Rett Syndrome which develop between the ages of one and four are described as 'autistic-like', such as repetitive hand-flapping, -washing, -wringing and -biting; body rocking and sleep problems. The loss of speech, seen as such a major childhood achievement and an essential in all areas of life, can be very upsetting for parents. Overall growth is slow and there is likely to be poor weight gain. Gross and fine motor skills can be severely affected, resulting in an irregular, stiff-legged gait, loss of fine finger control and little change in facial expressions.

Other common problems could include

- sleep disturbance
- breathing irregularities
- feeding and digestive difficulties (including swallowing, choking, regurgitation)

and later, in approximately 50% of women

- epileptic seizures
- scoliosis (curvature of the spine).

Cerebral palsy or autism may be suggested as a diagnosis by the family doctor during the early years.

The physical disabilities may appear to worsen as the child grows, and some skeletal deformities may eventually develop, particularly in the spine and feet. Behaviour and sleep problems should improve with age, however, and many of the girls who are affected by Rett Syndrome appear quite healthy. Recurrent mood swings are common, from intensely happy to sad, frustrated and angry. Their autistic-like behaviours will increase when they are upset or agitated.

Remember that not everyone will show all of the symptoms mentioned here, and individuals will have a wide difference in the severity of their problems.

The majority of girls with Rett Syndrome will become women with Rett Syndrome, and will need life-long care and attention on all levels to provide them with a stimulating, safe and loving environment in which to live.

Early management hints

- An early diagnosis is the best first step in the care of an individual with Rett Syndrome.
- A multi-disciplinary approach to encourage and stimulate motor use and communication skills is advocated by many experts as an essential part of any home and school programme.

This would include

- Physiotherapy
 - hydrotherapy
 - occupational therapy
 - speech and language therapy
 - play therapy
 - music therapy
- Good nutrition is essential to help counteract the early lack of growth and any skeletal problems
 - Do not underestimate their abilities or their interest in the world around them - many of their disabilities are motor in origin and they physically cannot produce a smile or a nod, or indeed any change in expression. Neither can they offer any voluntary finger / hand movement, nor in some cases, when they are older, will they be able to walk unaided.
 - Investigate and have in place calming techniques - gentle music, singing, soft toys, cuddling, gentle rocking, etc.

Other Useful Contacts

Rett Syndrome Association UK, Langham House West, Mill Street, Luton LU1 2NA. They offer information, advice, practical help, friendship and support to people with Rett Syndrome, their families and carers.

Web: www.rettsyndrome.org.uk

Email: info@rettsyndrome.org.uk

Tel: 01582 798910

Publications

Contact RSA UK for their publication lists. See also their website.

Pathways to Learning in Rett Syndrome. Jackie Lewis, Debbie Wilson. 1998. Price: £18.00 + p&p.

Routledge Education (member of the Taylor Francis Group)

Web: www.routledgeeducation.com

Tel: 020 7017 6000

Fax: 020 7017 6699

Available from the internet bookshop www.amazon.co.uk are many books dealing with Rett Syndrome.

Internet sites

The **Rett Syndrome Association UK's** useful site is at www.rettsyndrome.org.uk

The **Contact a Family pages** on Rett Syndrome are at www.cafamily.org.uk/Direct/r30.html

The **International Rett Syndrome Foundation**, based in the USA, is at www.rettsyndrome.org

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