Rett syndrome

Rett syndrome is a severe genetic disorder of the nervous system caused, in most cases, by a mutation in the MECP2 gene. The disorder is usually only seen in girls and affects all body movement. It causes loss of speech and hand use. Girls with Rett syndrome need therapy to help them with movement and communication.

Diagnosis of Rett syndrome

First and foremost, the diagnosis of Rett syndrome is based on an individual meeting a defined set of symptoms or criteria. The most recent revision to the diagnostic criteria was made in 2010. A genetic test which reveals a mutation in the MECP2 gene, confirms the diagnosis already determined or suspected by a medical professional.

Types of Rett syndrome

The two classifications of Rett syndrome are 'typical' or 'classical' Rett syndrome and 'atypical' or 'variant' Rett syndrome.

Typical Rett syndrome

The five criteria that must all be met for a person to receive a diagnosis of 'typical' or 'classical' Rett syndrome are:

- a period of regression during the first five years of life, followed by recovery or stabilisation
- partial or complete loss of acquired purposeful hand skills
- partial or complete loss of acquired spoken language (loss of acquired language is based on best acquired language skill. For example, if a child learned to babble and then lost that ability, it would be considered a loss of acquired language)
- gait abnormalities, such as an impaired ability to coordinate walking or an absence of the ability to walk
- stereotypic repetitive hand movements such as hand wringing/hand squeezing, clapping/tapping, mouthing and hand washing/hand rubbing.

Atypical Rett syndrome

Certain criteria must be met for a person to be given a diagnosis of 'atypical' or 'variant' Rett syndrome. These include having a period of regression followed by recovery or stabilization, at least two of the four essential symptoms required for a diagnosis of 'typical' or 'classical' Rett syndrome, and at least five of the common 11 symptoms, which include:

- breathing disturbances while awake
- inappropriate laughing or screaming spells
- bruxism (teeth grinding) while awake
- diminished response to pain
- sleep disturbances
- intense eye communication 'eye pointing'
- abnormal muscle tone
- growth retardation
- small hands and feet
- Cold or bluish hands and/or feet
- scoliosis (curvature of the spine) or kyphosis.

Stages of Rett syndrome

Rett syndrome progresses through four stages. These are:

- early onset (between six to 18 months)
- rapid destructive phase – which occurs between the age of one and four years
- plateau stage – the symptoms get no worse or their intensity lessens. This stage can last for years
- late motor deterioration (loss of movement) – this starts between five and 25 years of age and can last for decades.

Treatment for Rett syndrome

Therapy can help slow the progress of movement loss. Therapy includes:

- physiotherapy to prevent deformities of the joints and to improve movement
- occupational therapy to improve hand use
- horseback riding
- music therapy
- hydrotherapy (exercise in water).

Communication with a child with Rett syndrome

Children with Rett syndrome have a keen desire to communicate. They can communicate through:

- touch
- pictures and letters
- word boards
- eye gaze (including use of computer-related devices such as My Tobii and IPad)
- switch-operated voice output devices.

Education for a child with Rett syndrome

Children with Rett syndrome attend all types of schools. They need:

- early exposure to toys and music
- age-appropriate activities
- a school environment that provides strong motivation.

Where to get help

- Rett Syndrome Association of Australia Inc. Tel. 0418 561 796

Things to remember

- In Australia, Rett syndrome affects one female in 9,000 female live births.
- Development appears to be normal until the age of 6 to 18 months.
- It is hard to know how much a person with Rett syndrome understands or how intelligent they are, because of their communication problems.
- Rett syndrome wasn't recognised until 1983.

This page has been produced in consultation with, and approved by:

Rett Syndrome Association of Australia (RSAA)