Rett syndrome

Summary

- Rett syndrome is a severe condition of the nervous system.
- In Australia, Rett syndrome affects one female in 9,000 live female births.
- In general, development appears normal in a child with Rett syndrome until the age of 6 to 18 months.
- The degree of symptoms can vary widely among individuals with Rett syndrome.

What is Rett syndrome?

Rett syndrome is a severe condition of the nervous system. It is almost only seen in females, and affects all body movement.

Rett syndrome may cause speech problems (such as inability to learn to speak, or loss of speech), difficulty walking or loss of the ability to walk, and loss of purposeful hand use.

Females with Rett syndrome need a variety of therapies to help them with movement and communication.

What causes Rett syndrome?

Rett syndrome is caused by a change in the *MECP2* gene that is thought to disrupt the normal function of neurons and other cells in the brain. In most cases, there is no family history of the condition and the genetic change arises randomly in one individual.

How is Rett syndrome diagnosed?

There are two broad types of Rett syndrome:

- 'typical' (or 'classic') Rett syndrome
- 'atypical' (or 'variant') Rett syndrome.

Diagnosing typical Rett syndrome

A diagnosis of typical Rett syndrome requires five symptoms of decline:

- a period of regression (returning to a less developed state) during the first five years of life, followed by recovery or stabilisation
- partial or complete loss of purposeful hand skills (learned hand movements such as grasping, holding or self-feeding)
- partial or complete loss of language skills (speech)
- walking (gait) abnormalities, such as an unsteady walk or an inability to walk
- repetitive hand movements such as hand wringing or hand squeezing, clapping or tapping, mouthing and hand washing or hand rubbing movements.

Diagnosing atypical Rett syndrome

A diagnosis of atypical Rett syndrome requires a period of regression (returning to a less developed state) during the first five years of life, followed by recovery or stabilisation, and at least two of the other four symptoms for a diagnosis of typical Rett syndrome. Other atypical diagnosis factors 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 or feet
- scoliosis (curvature of the spine) or kyphosis.

Genetic testing for Rett syndrome

A genetic test that reveals a change in the *MECP2 gene* is used to confirm a diagnosis that has already been determined or is already suspected by a medical professional. The genetic test on its own is not enough to confirm Rett syndrome, as this change in the *MECP2* gene is also seen in other conditions.

Stages of Rett syndrome

Rett syndrome has four stages:

- **stage 1 – early onset** – between the ages of 6 and 18 months, development slows, developmental milestones may not be met
- **stage 2 – rapid destructive (regression) phase** – loss of acquired skills (such as loss of purposeful hand skills and speech and walking skills). This stage generally occurs between the age of one and four years. Other symptoms of Rett syndrome may occur
- **stage 3 – plateau stage** – abilities stabilise; intensity of symptoms may lessen. This stage can last for years
- **stage 4 – late motor deterioration (loss of movement)** – begins between five and 25 years of age, can last for decades but may not occur in all individuals. Involves reduced movement, muscle weakness and joint stiffness.

Treatment for Rett syndrome

There is no cure for Rett syndrome, but therapy can help slow the progress of movement loss. Therapies include:

- 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)
- other environmental enrichment activities.

Communication with a child with Rett syndrome

Children with Rett syndrome have a keen desire to communicate. Methods for communication may include:

- touch
- pictures and letters
- word boards
- eye gaze, via the use of computer-related devices such as My Tobii and iPad
- switch-operated voice output devices.

Education for a child with Rett syndrome

Most children with Rett syndrome attend specialist schools; some may attend a mixture of specialist and mainstream school, and some just mainstream. They need:

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

Genetic counselling and Rett syndrome

If your child has been diagnosed with Rett syndrome it may be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Rett syndrome and what causes it, how it is inherited, and what a diagnosis means for your child's health and development. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

Support for people and families living with Rett syndrome

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Rett syndrome.

The **Rett Syndrome Association of Australia** also organises gatherings of people and families living with Rett syndrome across Australia.

Where to get help

- **Genetic counsellor**
- **Rett Syndrome Association of Australia** Tel. **0418 561 796**
- **Genetic Support Network of Victoria (GSNV)** Tel. **(03) 8341 6315**
- **Victorian Clinical Genetics Services (VCGS)** Tel. **1300 118 247**

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

Rett Syndrome Association of Australia (RSAA)