

Rett syndrome

Rett syndrome severely affects speech and movement. People with the syndrome are nearly always girls. Mutations in the gene MECP2, which is located on the X chromosome at Xq28, are a cause of Rett syndrome.

A variety of physical effects

People are diagnosed with Rett syndrome if they display a certain set of symptoms in their first three or four years. These include:

- Period of normal early development.
- Slowed head growth.
- Severe impairment of expressive language.
- Loss of purposeful hand use, followed by repetitive hand movements such as clapping, tapping and wringing.
- Shakiness of the upper body - this may extend to the legs and arms.
- Unsteady walk (if they can walk). They walk with stiff legs and feet wide apart.

Other effects of Rett syndrome

Other effects of Rett syndrome that may be present but are not necessary for diagnosis include:

- Breath holding, hyperventilation and air swallowing
- Spinal curvature, rigid (inflexible) muscles and contracted joints
- Seizures (fits)
- Bluish-red feet and legs because of poor circulation
- Teeth grinding and difficulty swallowing.

Four stages of Rett syndrome

Rett syndrome progresses through four stages. These are:

- **Early onset** (between six to 18 months).
- **Rapid destructive phase** - this 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.

How to help a child with Rett syndrome

Therapies

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

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

- Touch
- Pictures and letters
- Word boards
- Using eye gaze
- Switch-operated voice output devices.

Education

- People 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 about 1 in 15,000 females aged 5 to 18 years
- Development is usually 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)

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