Rett Syndrome

Definition:

- Rett syndrome is a rare genetic disorder that affects the way the brain develops. It occurs almost exclusively in girls.
- Most babies with Rett syndrome develop normally at first, but symptoms start to surface after 6 months of age. Over time, children with Rett syndrome have increasing problems with movement, coordination and communication that may affect their ability to use their hands, communicate and walk.
- Although there's no cure for Rett syndrome, potential treatments are being studied.
- Current Rett syndrome treatment focuses on improving movement and communication and providing care and support for affected children and their families.

Symptoms

The most pronounced changes generally occur at 12 to 18 months of age and occur over a period of weeks or months.

- **Slowed growth:** Brain growth slows after birth. Small head and delayed growth in all other body parts.
- **Loss of normal movement and coordination:** Usually starts between 12 to 18 months of age. The first signs often include a decrease of hand control and a decreasing ability to crawl or walk normally.
- **Loss of communication and thinking abilities:** Lose the ability to speak and to communicate in other ways. May become uninterested in other people, toys and their surroundings. May be rapid or gradual
- **Abnormal hand movements:** Hand wringing, squeezing, clapping, tapping or rubbing.
- **Unusual eye movements.** Blinking or closing one eye at a time.
- **Breathing problems:** These include breath-holding (apnea), abnormally rapid breathing (hyperventilation), and forceful exhalation air or saliva. These problems tend to occur during waking hours, but not during sleep.
- **Irritability:** Become increasingly agitated and irritable as they get older. Periods of crying or screaming may begin suddenly and last for hours. May become calmer between the ages of 2 and 10 years old.
- **Abnormal behaviors:** These may include sudden, odd facial expressions and long bouts of laughter, screaming that occurs for no apparent reason, hand licking, and grasping of hair or clothing.
- **Seizures:** 50% or more develop seizures. Symptoms vary from person to person.
- **Abnormal curvature of the spine (scoliosis):** Typically begins between 8-11 years of age.
- **Irregular heartbeat (arrhythmia)**
- **Constipation.**

Tests and diagnosis

Diagnosing Rett syndrome involves careful observation of your child's growth and development and answering questions about her or his medical and family history.

The official criteria required for a diagnosis of Rett syndrome include:

- Apparently normal development for the first five months after birth
- Normal head circumference at birth, followed by a slowing of the rate of head growth between the ages of 5 months and 4 years
- Severely reduced language skills
- Loss of hand skills and development of repetitive hand movements between the ages of 5 months and 30 months
- Loss of interaction with others (though this often improves later)
- An unsteady walk or poorly controlled torso movements
- Severely impaired ability to communicate and move normally
- May have other signs and symptoms

Treatments and drugs

Requires a team approach, including regular medical care; physical, occupational and speech therapy; and academic, social and job training services. The need for support doesn't end as children become older — it's usually necessary throughout life.

Treatments that can help children and adults with Rett syndrome include:

- **Medications.** They can't cure Rett syndrome, but may help control some of the symptoms associated with the disorder, such as seizures and muscle stiffness.
- **Physical and speech therapy.**
- **Nutritional support.** Proper nutrition is extremely important for both normal growth and for improved mental and social abilities. May need a high-fat, high-calorie diet. Others may need to be fed through a tube placed in the nose (nasogastric tube) or directly in the stomach (gastrostomy)