**Common Rett Syndrome Symptoms**

Most children with Rett syndrome start to show some of the following symptoms:

1. Loss of purposeful hand movements
2. Loss of speech
3. Balance and coordination problems
4. Breathing problems
5. Social behavioral problems
6. Excessive saliva and drooling
7. Slowing head growth
8. Loss of normal sleep patterns

**Rett Syndrome: When to see a doctor**

Signs of Rett syndrome can be subtle at the early stages, but the following changes in behavior should indicate a need for medical intervention:

1. Slowed growth of child’s head or other parts of her or his body
2. Decreased coordination or mobility
3. Decreasing systocontact or loss of interest in normal play

**What is Rett Syndrome?**

Rett syndrome is a rare, severe form of autism, which affects neurological growth that mostly occurs in females and leads to developmental reversals, especially in the areas of expressive language and hand use.

Most babies with Rett syndrome are generally born after a normal pregnancy and delivery. Then they develop normally at first, but Rett syndrome symptoms appear after approximately 6 to 18 months of age. It affects children of all races and the only known risk factor is having random genetic mutations known to cause the disease. And in rare cases inherited factors may play a role.

Over time, these children have increasing problems with movement, coordination and communication that may affect their ability to use their hands, communicate and walk. Until recently, researchers thought that Rett syndrome affected only females, but they now know that it affects males as well.

Males born with this defective gene do not have a second X chromosome to make up for the problem. Therefore, the defect usually results in miscarriage, stillbirth, or very early death. Because the number of males with Rett syndrome is very small, most statistics and research on the syndrome are specific to females.

**What causes Rett Syndrome?**

Most children with Rett syndrome have a mutation in a particular gene on the X chromosome. Exactly what this gene does, or how its mutation leads to Rett syndrome is not clear. It is believed that the single gene may influence many other genes involved in development.

Although Rett syndrome seems to be genetic, the faulty gene is almost never inherited from the parents. Rather, it is a chance mutation that happens in the baby’s own DNA. No Rett syndrome risk factors have been identified.

**Stages of Rett Syndrome:**

Scientists generally describe four stages of Rett syndrome. Stage I, called early onset, typically begins between 6 and 18 months of age. This stage is often overlooked because symptoms of the disorder may be somewhat vague, and parents and doctors may not notice the subtle slowing of development at first. The infant may begin to show less eye contact and have reduced interest in toys. There may be delays in gross motor skills such as sitting or crawling. Hand-writhing and decreasing head growth may also occur, but not enough to draw attention. This stage usually lasts for a few months but can continue for more than a year.

Stage II, or the rapid destructive stage, usually begins between ages 1 and 4 and may last for weeks or months. Its onset may be rapid or gradual as the child loses purposeful hand skills and spoken language.

Characteristic hand movements such as wringing, washing, clapping, or tapping, as well as repeatedly moving the hands to the mouth often begin during this stage. The child may hold the hands clasped behind the back or held at the sides, with random touching, grasping, and releasing. The movements continue while the child is awake but disappear during sleep. Breathing irregularities, which include episodes of apnea and hyperventilation may occur, although breathing usually improves during sleep.

**Rett Syndrome: A Rare Genetic Disorder that Impacts Brain Development**

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Some children also display autistic-like symptoms, loss of social interaction and communication. Walking may be unsteady and initiating motor movements can be difficult. Slowed head growth is usually noticed during this stage.

Stage III, or the plateau or pseudo-stationary stage, usually begins between ages 2 and 10 and can last for years. Apraxia, motor problems, and seizures are prominent during this stage. However, there may be improvement in behavior, with less irritability, crying, and autistic-like features. A girl in stage III may show more interest in her surroundings and her alertness, attention span, and communication skills may improve. Many girls remain in this stage for most of their lives.

Stage IV, or the late motor deterioration stage, can last for years or decades. Prominent features include reduced mobility, curvature of the spine (scoliosis) and muscle weakness, rigidity, spasticity, and increased muscle tone with abnormal posturing of an arm, leg, or top part of the body. Girls who were previously able to walk may stop walking. Cognition, communication, or hand skills generally do not decline in stage IV. Repetitive hand movements may decrease and eye gaze usually improves.

A pediatric neurologist, clinical geneticist, or developmental pediatrician should be consulted to confirm the clinical diagnosis of Rett syndrome. The physician will use a highly specific set of guidelines that are divided into three types of clinical criteria: essential, supportive, and exclusion. The presence of any of the exclusion criteria negates a diagnosis of classic Rett syndrome.

Examples of essential diagnostic criteria or symptoms include having apparently normal development until between the ages of 6 and 18 months and a normal head circumference at birth followed by a slowing of the rate of head growth with age (between 3 months and 4 years). Other essential diagnostic criteria include severely impaired expressive language, repetitive and stereotypic hand movements, and gait abnormalities, including toe-walking or an unsteady, wide-based, stiff-legged walk.

Supportive criteria are not required for a diagnosis of Rett syndrome but may occur in some individuals. In addition, these symptoms, which vary in severity from child to child, may not be observed in very young girls but may develop with age. A child with supportive criteria but none of the essential criteria does not have Rett syndrome. Supportive criteria include breathing difficulties, electroencephalogram (EEG) abnormalities, seizures, muscle rigidity, spasticity and/or joint contractures which worsen with age, scoliosis. teeth-grinding, small hands and feet in relation to height, growth retardation, decreased body fat and muscle mass (although there may be a tendency toward obesity in some affected adults), abnormal sleep patterns, irritability or agitation, chewing and/or swallowing difficulties, poor circulation of the lower extremities with cold and bluish-red feet and legs, decreased mobility with age, and constipation.

In addition to the essential diagnostic criteria, a number of specific conditions enable physicians to rule out a diagnosis of Rett syndrome. These are referred to as exclusion criteria. Children with any one of the following criteria do not have Rett syndrome: enlargement of body organs or other signs of storage disease, vision loss due to retinal disorder or optic atrophy, abnormally small head at birth (microcephaly), an identifiable metabolic disorder or other inherited degenerative disorder, an acquired neurological disorder resulting from severe infection or head trauma, evidence of growth retardation in utero, or evidence of brain damage acquired after birth.

**Rett syndrome treatment**

There is currently no cure for Rett syndrome. However, these children can be treated for some of the problems associated with the condition. These treatments aim to slow the loss of abilities, improve or preserve movement, and encourage communication and contact.

People with Rett syndrome often benefit from a team approach to care, in which many kinds of health care providers play a role, along with family members. Members of this team may include:

- Physical therapists, who can help patients improve or maintain mobility and balance and reduce misshapen back and limbs
- Occupational therapists, who can help patients improve or maintain use of their hands and reduce stereotypic hand movements
- Speech-language therapists, who can help patients use non-verbal ways of communication and improve social interaction
- Other options, such as medication (such as for constipation or heart problems) or surgery (to correct spine curvature or correct heart defects) are also effective for treating some of the symptoms of Rett syndrome.

How is Rett syndrome diagnosed?

Doctors clinically diagnose Rett syndrome by observing signs and symptoms during the child’s early growth and development, and conducting ongoing evaluations of the child’s physical and neurological status. Scientists have developed a genetic test to complement the clinical diagnosis, which involves searching for the MECP2 mutation on the child’s X chromosome.