

# **Rett's Disorder**

## **Common Names for Disorder:**

Rett's Syndrome, RS

## **What is Rett's Disorder?**

Rett syndrome is a childhood neurodevelopmental disorder characterized by normal early development followed by loss of purposeful use of the hands, distinctive hand movements, slowed brain and head growth, gait abnormalities, seizures, and mental retardation. It affects females almost exclusively.

It also causes problems in brain function that are responsible for cognitive, sensory, emotional, motor and autonomic function. These can include learning, speech, sensory sensations, mood, movement, breathing, cardiac function, and even chewing, swallowing, and digestion.

## **How is Rett's Diagnosed?**

Doctors 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. Rett syndrome is most often misdiagnosed as autism, cerebral palsy, or nonspecific developmental delay. However, since it is known that the MECP2 mutation is also seen in other disorders, the presence of MECP2 mutation in itself is not enough for the diagnosis of Rett syndrome. Diagnosis requires either the presence of the mutation (a molecular diagnosis) or fulfillment of the diagnostic criteria (a clinical diagnosis, based on observable signs and symptoms) or both. The simple blood test used to make a clinical diagnosis involves searching for the MECP2 mutation on the child's X chromosome. Given what is known about the genes involved in Rett syndrome, such tests are able to confirm a clinical diagnosis in up to 80 percent of all cases.

## **When and what symptoms develop?**

The course of Rett syndrome, including the age of onset and the severity of symptoms, varies from child to child. Before the symptoms begin, however, the child appears to grow and develop normally. Then, gradually, mental and physical symptoms appear. Hypotonia (loss of muscle tone) is usually the first symptom. As the syndrome progresses, the child loses purposeful use of her hands and the ability to speak. Other early symptoms may include problems crawling or walking and diminished eye contact. The loss of functional use of the hands is followed by compulsive hand movements such as wringing and washing. The onset of this period of regression is sometimes sudden.

Another symptom, apraxia — the inability to perform motor functions — is perhaps the most severely disabling feature of Rett syndrome, interfering with every body movement, including eye gaze and speech. Individuals with Rett syndrome often exhibit autistic-like behaviors in the early stages. Other symptoms may include toe walking; sleep problems; wide-based gait; teeth grinding and difficulty chewing; slowed growth; seizures; cognitive disabilities; and breathing difficulties while awake such as hyperventilation, apnea (breath holding), and air swallowing.

### **What is the course of Rett Syndrome?**

***STAGE ONE, EARLY-ONSET (SIX–18 MONTHS OF AGE).*** The early symptoms of RS are not always noticeable in Stage 1. The infant may not make eye contact with family members and may not show much interest in toys. There may also be delays in gross motor skills such as sitting or crawling. There may be noticeable hand-wringing and slowing of head growth. This stage usually lasts for a few months but can persist for more than a year.

***STAGE TWO, RAPID DETERIORATION (ONE–FOUR YEARS).*** This stage may be either rapid or gradual in onset. The child loses her ability to speak and to make purposeful hand movements. Hand-to-mouth movements may appear, as well as hand-wringing or hand-clapping gestures. These movements may be nearly constant while the child is awake but disappear during sleep. There may be noticeable episodes of breath holding and hyperventilating (rapid shallow breathing). The child may have trouble sleeping, and may become irritable. If the child is able to walk, she will start to look unsteady on her feet and may have periods of trembling or shaking. Slowed growth of the head is usually most noticeable during this stage.

***STAGE THREE, PLATEAU (TWO–10 YEARS).*** Motor problems (i.e. Apraxia) and seizures often appear during this stage. The child's behavior, however, often shows some improvement, with less irritability and crying. She may show greater interest in her surroundings, and her attention span and communication skills often improve. Many patients with RS remain in stage 3 for most of their lives.

***STAGE FOUR, LATE DETERIORATION OF MOTOR SKILLS (USUALLY AFTER 10 YEARS OF AGE).*** In stage 4, patients with RS gradually lose their mobility; some stop walking while others have never learned to walk. There is, however, no loss of cognitive or communication skills, and the repetitive hand movements may decrease. The spine begins to develop an abnormal sideways curvature (scoliosis), and the patient may develop muscle rigidity. Puberty begins at the same age as in most girls.

### **What causes Rett Syndrome?**

Rett syndrome is caused by mutations (structural alterations or defects) in the MECP2 (pronounced *meck-pea-two*) gene, which is found on the X chromosome (*see* section on "Who gets Rett syndrome" for a discussion of the importance of the involvement of the X chromosome). Scientists identified the gene — which is believed to control the functions of several other genes — in 1999. The MECP2 gene contains instructions for the synthesis of a protein called methyl cytosine binding protein 2 (MeCP2), which acts as one of the many biochemical switches that tell other genes when to turn off and stop producing their own unique proteins. Because the MECP2 gene does not function properly in those with Rett syndrome, insufficient amounts or structurally abnormal forms of the protein are formed. The absence or malfunction of the protein is thought to cause other genes to be abnormally expressed, but this hypothesis has not yet been confirmed.

Seventy to 80 percent of girls given a diagnosis of Rett syndrome have the MECP2 genetic mutation detected by current diagnostic techniques. Scientists believe the remaining 20 to 30 percent of cases may be caused by partial gene deletions, by mutations in other parts of the gene, or by genes that have not yet been identified; thus, they continue to search for other mutations.

Incidence:

### **What treatment is available?**

There is no cure for Rett syndrome. Treatment for the disorder is symptomatic — focusing on the management of symptoms — and supportive, requiring a multidisciplinary approach. Medication may be needed for breathing irregularities and motor difficulties, and antiepileptic drugs may be used to control seizures. There should be regular monitoring for scoliosis and possible heart abnormalities. Occupational therapy (in which therapists help children develop skills needed for performing self-directed activities — *occupations* — such as dressing, feeding, and practicing arts and crafts), physiotherapy, and hydrotherapy may prolong mobility. Some children may require special equipment and aids such as braces to arrest scoliosis, splints to modify hand movements, and nutritional programs to help them maintain adequate weight. Special academic, social, vocational, and support services may also be required in some cases.

### **Diagnostic Category:**

*IDEA*: Other Health Impaired

means having limited strength, vitality or alertness, including a heightened alertness to environmental stimuli, that results in limited alertness with respect to the educational environment, that—(i) Is due to chronic or acute health problems such as asthma, attention deficit disorder or attention deficit hyperactivity disorder, diabetes, epilepsy, a heart condition, hemophilia, lead poisoning, leukemia, nephritis, rheumatic fever, and sickle cell anemia; and (ii) Adversely affects a child's educational performance

*DSM-IV*: Pervasive Developmental Disorder

### **299.80 Rett's Disorder**

(A) All of the following:

1. Apparently normal prenatal and perinatal development
2. Apparently normal psychomotor development through the first 5 months after birth
3. Normal head circumference at birth

(B) Onset of all of the following after the period of normal development:

1. Deceleration of head growth between ages 5 and 48 months
2. Loss of previously acquired purposeful hand skills between ages 5 and 30 months with the subsequent development of stereotyped hand movements (e.g., hand-wringing or hand washing)
3. Loss of social engagement early in the course (although often social interaction develops later)
4. Appearance of poorly coordinated gait or trunk movements
5. Severely impaired expressive and receptive language development with severe psychomotor retardation

**Deficits and Long-Term Developmental Outcomes:**

Despite the difficulties with symptoms, most individuals with Rett syndrome continue to live well into middle age and beyond. Because the disorder is rare, very little is known about long-term prognosis and life expectancy. While it is estimated that there are many middle-aged women (in their 40s and 50s) with the disorder, not enough women have been studied to make reliable estimates about life expectancy beyond age 40.

**Assessment Approaches:**

Generally these assessments are multidisciplinary. Developmental clinics may be the best place to complete an evaluation. Often the battery of assessment devices includes a cognitive assessment, an adaptive functioning assessment, and often pediatricians or geneticists may complete the evaluation to assess for genetic or chromosomal contributions.

**Contributions of the School Psychologist:**

Interventions and treatment will vary from child to child. Some considerations for school psychologists are: maintain communication between parents and school, provide consultation to teachers and parents. The child may benefit from occupational, speech, and physical therapy as well as, academic, social, vocational, and support services.

**Resources**

International Rett Syndrome Foundation- <http://www.rettsyndrome.org/>

National Institute of Neurological Disorder and Stroke -  
[http://www.ninds.nih.gov/disorders/rett/detail\\_rett.htm](http://www.ninds.nih.gov/disorders/rett/detail_rett.htm)

Rett Syndrome Research Foundation - [http://www.rsrp.org/parent\\_resources/9.2.html](http://www.rsrp.org/parent_resources/9.2.html)