

## What is Rett syndrome?

Rett syndrome is a rare neurodevelopmental disorder that presents in childhood. It was first described in 1966 by Andreas Rett, an Austrian pediatrician and neurologist. The symptoms, progression, and severity can vary for each child. Rett syndrome is considered a very severe brain disorder that affects both the person with Rett and their family.

Rett syndrome has been divided into two main categories:

- Typical (classic Rett)
- Atypical (variant Rett)

## Who is affected by Rett syndrome?

Rett syndrome almost always occurs in girls. Boys may also be affected, but it is rare. Approximately 1 per 10,000 girls ages 2 through 18 may develop Rett syndrome. All ethnic and racial groups can be affected by Rett syndrome.

## How does typical Rett syndrome present?

Children with Rett syndrome will develop normally at first. In the early months of life, a child will not show signs of any difficulties. At some point, usually between 6 and 18 months, the child's development will stop. They will begin to show signs of losing skills (regression.) Head growth slows and the child will develop microcephaly (smaller head size than expected for age.)

Children with Rett syndrome typically have:

- Partial or complete loss of their ability to speak.
- Partial or complete loss of purposeful hand and finger movements
- Difficulty walking or inability to walk.
- Stereotyped (near constant, repetitive) hand movements that usually include hand wringing, but may also involve clapping, tapping, rubbing, or hand to mouth movements. Purposeful hand movement is lost.

A child impacted by Rett syndrome may also have:

- Poor brain growth and intellectual disability
- Nutritional problems, growth failure, muscle wasting, low bone density.
- Abnormal breathing patterns with periods of over breathing mixed with slower breathing. This can be alarming to watch but is not dangerous.
- Heart problems which may lead to abnormal heart rhythm.
- Drooling, sweating, cold hands and feet.
- Poor sleep patterns
- Bowel and bladder incontinence
- Orthopedic problems (scoliosis, kyphosis)
- Seizures

- Behaviors consistent with autism (inappropriate laughing and screaming)
- Grinding teeth (bruxism)

## How does atypical Rett syndrome present?

There are 3 types of atypical Rett syndrome, Rett syndrome with:

- Preserved speech (milder form)
- Early seizures (seizures start before fifth month of life)
- Testing that may show a congenital (during fetal development) variant that begins in first 6 months of life.

## What kinds of seizures do children with Rett syndrome have?

Seizures occur in the majority of children with Rett syndrome. Studies have shown that presence of seizures in children increases with age:

- About 1 in 3 children ages 2 to 5 will have seizures
- About 2 out of 3 children ages 5 to 10 will have seizures
- About 3 out of 4 children ages 10 to 15 will have seizures

How often a child has seizures will vary. Sometimes children have seizures during sleep which are not identified. The most common [type of seizures](#) seen in Rett syndrome are focal impaired awareness, tonic-clonic, tonic, and myoclonic seizures. About half of children with Rett syndrome and epilepsy have [drug-resistant seizures](#) (seizures that do not respond to medical therapy.) Behavioral symptoms can sometimes be mistaken for seizures. Other symptoms related to Rett syndrome are more severe in children who also have seizures.

## How is Rett syndrome diagnosed?

Rett syndrome is diagnosed by a healthcare provider and is based on a child's symptoms and developmental history. The healthcare provider will pay close attention to when a child reaches their developmental milestones (speech, hand and finger skills, walking). A loss of skills (clinical regression) occurs in all children diagnosed with Rett syndrome. The evaluation will include typical testing to [diagnose epilepsy](#) (medical and neurologic exam, blood tests, EEG and brain imaging) and will also include genetic testing.

The underlying cause of Rett syndrome can be identified by [genetic testing](#). Prior to genetic testing, discuss the potential implications of these results with your healthcare provider. Understanding what genetic test results can and cannot determine is important. Genetic counseling can also be helpful in sorting out what your test results mean, and for providing counseling on the risk of recurrence for other family members.

**Call our Helpline:** 1.800.332.1000 | 1.866.748.8008 (en español)

**Contact your local Epilepsy Foundation:** [epilepsy.com/local](http://epilepsy.com/local)

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## What is known about the genetics of Rett?

In most children, Rett syndrome is caused by a variant in the MECP2 gene. In 1999, the identification of variants in the MECP2 gene in children with Rett syndrome has made genetic testing and confirmation of disease possible for children who are affected. Rett syndrome is most often caused by a de novo (new, first time occurring in a family) variant in the MECP2 gene. Variants in the MECP2 gene can also be present in other neurologic disorders. Exactly how MECP2 gene variants lead to Rett syndrome is not known. Variants in the MECP2 gene have been found in about 95% of typical Rett syndrome cases. Variants in the MECP2 gene have been found in 3 out of 4 children with atypical Rett syndrome. Other gene variants seen in atypical Rett syndrome include variants in the [CDKL5](#) and FOXP1 genes.

## What EEG findings are seen with Rett syndrome?

In Rett syndrome, [EEG testing](#) is always abnormal. Changes in brain waves that are consistent with epilepsy are usually seen by about two years of age. Abnormal changes seen on an EEG test in a child with Rett syndrome include brain wave slowing, epileptiform discharges, seizures, and abnormal patterns. The changes seen on EEG get worse as the disease progresses.

## How is Rett syndrome treated?

No specific therapy exists for Rett syndrome. [Treatment](#) focuses on managing as best possible the different conditions that cause a person's Rett Syndrome symptoms:

- A multidisciplinary team made up of different specialist providers (neurologist, orthopedist, internist, cardiologist, gastroenterologist) is helpful when caring for a child with Rett syndrome.
- Physicians, nurses, physical and occupational therapist, social work, and nutrition teams all work together to help a child and family facing the challenges of Rett syndrome.
- Treatment of motor problems, nutritional problems, seizures, behavior, orthopedic, pulmonary, cardiac, and gastrointestinal problems is often necessary.
- Seizures can usually be treated with anti-seizure medications. However, sometimes medically prescribed diet therapy (ketogenic diet) and vagal nerve stimulator (VNS) are often used as add on treatments when seizures do not respond to anti-seizure medications.
- There is no single "best" anti-seizure medication, the choice of medication will typically depend on the type or types of seizures.
- If infantile spasms are present, treatment with hormonal therapy or vigabatrin can be helpful.

## What is the outlook Rett syndrome?

Children with typical Rett syndrome will develop normally for the first 6 to 18 months, then regress or lose skills. Loss of speech, purposeful hand movement, trouble walking, and stereotyped hand movements are seen and limit typical toddler or childhood function. The regression phase can sometimes be followed by a period of stability or some recovery of nonverbal communication. People with Rett syndrome are dependent on others for activities of daily living (ADLs) throughout their lifetime. The long-term course or outlook usually includes a slow loss of motor function.

Pneumonia may occur in people with swallowing problems who aspirate (food or fluid go into the lungs.) Complications of pneumonia and seizures are the most common causes of death among people with Rett syndrome.

About 2 out of 3 women over age 18 with Rett syndrome need anti-seizure medication. Most people affected by Rett syndrome will live into adulthood. The caregiver burden is high and support for caregivers' mental and physical health are needed. People with Rett syndrome may live beyond middle age. Families need to factor this into consideration for financial planning, guardianship, and long-term providers of care.

### For more Information

- [International Rett Syndrome Foundation](#)
- [Rettlund Foundation](#)
- [Rare Epilepsy Network \(REN\)](#)
- [MedlinePlus](#)

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