

Rett Syndrome



What is Rett Syndrome?

Rett syndrome is a rare, severe neurological disorder that affects mostly girls. It's usually discovered in the first two years of life, and a child's diagnosis with Rett syndrome can feel overwhelming. Although there's no cure, early identification and treatment may help girls and families who are affected by Rett syndrome. In the past, it was felt to be part of the Autism Spectrum Disorder. We now know that it is mostly genetically based.

Symptoms

The age when symptoms appear varies, but most babies with Rett syndrome seem to grow normally for the first 6 months before any signs of the disorder are obvious. The most common changes usually show up when babies are between 12 and 18 months, and they can be sudden or progress slowly.

Symptoms of Rett syndrome:

- **Slowed growth.** The brain doesn't grow properly, and the head is usually small (doctors call this microcephaly). This stunted growth becomes clearer as the child gets older.
- **Problems with hand movements.** Most children with Rett syndrome lose the use of their hands. They tend to wring or rub their hands together.
- **No language skills.** Between ages 1 to 4, social and language skills start to decline. Children with Rett syndrome stop talking and can have extreme social anxiety. They may stay away from or not be interested in other people, toys, and their surroundings.
- **Problems with muscles and coordination.** This can make walking awkward.
- **Trouble with breathing.** A child with Rett may have uncoordinated breathing and seizures, including very fast breathing (hyperventilation), forceful exhaling of air or saliva, and swallowing air.

Children with Rett syndrome also tend to become tense and irritable as they get older. They may cry or scream for long periods of time, or have long fits of laughter.

Symptoms of Rett syndrome usually don't improve over time. It's a lifelong condition. Often, the symptoms worsen very slowly, or don't change. It's rare for people with Rett syndrome to be able to live independently.

Causes

Most children with Rett syndrome have a mutation on the X chromosome. Exactly what this gene does, or how its mutation leads to Rett syndrome, isn't clear. Researchers believe that the single gene may influence many other genes involved in development.

Although Rett syndrome is genetic, children almost never inherit the faulty gene from their parents. Rather, it's a chance mutation that happens in DNA.

When boys develop the Rett syndrome mutation, they rarely live past birth. Males have only one X chromosome (instead of the two girls have), so the effects of the disease are much more serious, and almost always fatal.

Diagnosis

A diagnosis of Rett syndrome is based on a girl's pattern of symptoms and behavior. Doctors can make the diagnosis based on these observations alone and by talking to the girl's parents about things like when the symptoms started.

Because Rett syndrome is rare, doctors will first rule out other conditions, including autism spectrum disorder, cerebral palsy, metabolic disorders, and prenatal brain disorders. Genetic testing can help confirm the diagnosis in 80% of girls with suspected Rett syndrome. These tests may also predict how severe it will be.

Treatments

Although there is no cure for Rett syndrome, there are treatments that can improve symptoms. And children should continue these treatments for their entire life.

The best options available to treat Rett syndrome include:

- Standard medical care and medication
- Physical therapy
- Speech therapy
- Occupational therapy
- Good nutrition
- Behavioral therapy
- Supportive services

Medicines can treat some of the problems with movement in Rett syndrome. Medication can also help control seizures. Many girls with Rett syndrome can live at least into middle age. Researchers are studying women with the disease, which was only widely recognized in the last 20 years.

For more information, visit:

- **RettSyndrome.org** – www.rettysyndrome.org
- **New Jersey Rett Syndrome Association** - www.njrja.org
- **Rett syndrome - Mayo Clinic** - www.mayoclinic.org/diseases-conditions/rett-syndrome/basics/definition/con-20028086