Rett Syndrome

Introduction

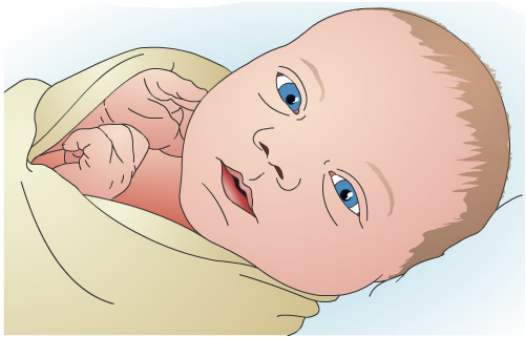
Rett syndrome is a rare genetic disorder. It affects the way the brain develops. It mostly affects girls.

Most babies with Rett syndrome seem to develop normally at first. Symptoms begin after 6 months of age. Over time, children with Rett syndrome have problems with movement, coordination and communication.

This reference summary explains Rett Syndrome. It discusses the symptoms, causes and treatment of the disorder.

Symptoms

Rett syndrome is a disorder of the nervous system. It causes developmental reversals. This means that a child loses some abilities as he or she ages. Communication skills and hand use are most often affected.



Babies with Rett syndrome tend to look healthy at birth. They often are born after a normal pregnancy and birth. Most seem to grow and behave normally for the first six months of life.

Symptoms of Rett syndrome begin to appear after the first six months. The most noticeable changes happen at 12 to 18 months of age. These changes develop over a period of weeks or months. Brain growth slows in babies with Rett syndrome. Smaller than normal head size often is the first sign that a child has the disorder. Delayed growth in other parts of the body also may happen.

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Other Rett syndrome symptoms include:

- Slowed growth.
- Loss of normal movement and coordination.
- Loss of communication and thinking abilities.
- Abnormal hand movements.
- Unusual eye movements, such as intense staring, blinking or winking.

At first, the loss of abilities happens quickly. The loss then continues more gradually. The most significant loss of movement skills usually starts between 12 and 18 months of age. The first signs often are a loss of hand control and the ability to crawl or walk normally.

Children with Rett syndrome typically begin to lose the ability to speak. Some children have sudden changes, such as an abrupt loss of speech. They may also lose the ability to communicate in other ways.

Children with Rett syndrome become more agitated and irritable as they get older. Periods of crying or screaming may start suddenly and last for hours. The child may:

- Make odd, sudden facial expressions.
- Laugh or scream for no reason.
- Lick his or her hands.
- Grasp at hair or clothing.

Most people who have Rett syndrome have seizures at some time during their lives. Seizures are sudden, uncontrolled body movements and changes in behavior. Seizures happen because of abnormal electrical activity in the brain. Symptoms include loss of awareness, changes in emotion, loss of muscle control and shaking.

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Symptoms vary from person to person. They can range from periodic muscle spasms to full epilepsy. A spasm is a sudden cramp or contraction of a muscle or group of muscles. Epilepsy is a group of disorders marked by problems in the normal functioning of the brain. These problems can produce seizures, unusual body movements or loss of consciousness. It can also cause mental problems or problems with the senses.

Dysrhythmia is a life threatening problem for many children and adults with Rett syndrome. It causes the heart to beat irregularly. The heart may beat too fast or too slow. Dysrhythmia can cause chest pain, dizziness, fainting and shortness of breath. The condition may increase a person's chances of heart failure, a blood clot or a stroke.

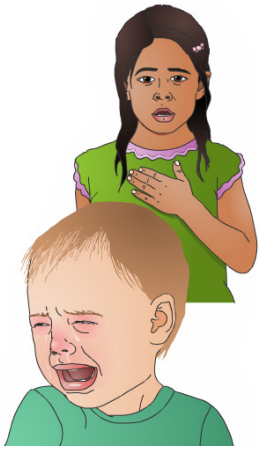
Other signs and symptoms of Rett syndrome may include:

- Sleep problems.
- A wide gait when walking.
- Walking on the toes.
- Teeth grinding and difficulty chewing.
- Constipation.
- Thin, fragile bones.

Stages of Rett Syndrome

Rett syndrome has four stages. Stage I is known as the early onset stage. During this stage, signs and symptoms are subtle. This means that they are hard to notice. Stage I starts between 6 and 18 months of age. Babies in stage I may make less eye contact and start to lose interest in toys. They may also have delays in sitting or crawling. They may move jerkily.

Stage II is known as the rapid destructive phase. It starts between 1 and 4 years of age. Children in this stage gradually lose the ability to speak and to use their hands. Children in stage II begin to move their hands in repetitive and purposeless movements. The child may hold his or her hands clasped behind the back or at the sides. The hands may randomly touch, grasp, clap, rub and release. The movements continue while the child is awake but go away during sleep.



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During stage II, some children with Rett syndrome begin to hold their breath or hyperventilate. Hyperventilation is rapid or deep breathing. It can be caused by anxiety or panic. It is also called over breathing. They may scream or cry for no reason. It is often hard for them to move on their own.

The third stage of Rett syndrome is called the plateau or pseudo-stationary stage. It usually happens between the ages of 2 and 10 years. Stage III can last for years.

Although problems with movement continue, behavior may improve. Children in stage III often cry less and become less irritable. Eye contact usually improves during this stage. A child in stage III may show more interest in his or her surroundings. Alertness, attention span and communication skills may improve. Many people remain in this stage for most of their lives.

During the last stage of Rett syndrome, it is hard for the person with the condition to move. They may feel weakness in their muscles and develop scoliosis. Scoliosis is a condition marked by a side-to-side curve of the backbone. The curve is usually shaped like an S or a C. In most cases, the cause of scoliosis is not known. An arm, leg or top part of the body may move into an abnormal pose.

In stage IV, a person who was able to walk may stop walking. Understanding, communication and hand skills usually do not decline during this stage. Repetitive hand movements may decrease. Eye contact also may improve.

Although sudden death can occur, the average life span of people with Rett syndrome is more than 50 years. They usually need care throughout their lives.

Causes

Rett syndrome is rare. It is estimated to affect one in every 10,000 to 15,000 female babies. It affects all racial and ethnic groups worldwide. Rett syndrome is a genetic disorder. But it is only inherited in a few cases, meaning it is passed from a parent to a child. Less than 1 percent of cases are inherited. Most cases of Rett syndrome are spontaneous, which means they happen randomly.

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Prenatal testing is available for families who already have a child with Rett syndrome. But the risk of a family having a second child with the disorder is less than 1 percent. Prenatal is the time when a woman is pregnant, before birth happens.

Most people with Rett syndrome are girls. Males have a different chromosome combination than females. Most boys who have the genetic mutation that causes Rett syndrome are affected in severe ways and die before birth or as babies.

A genetic mutation is any change in the DNA sequence of a cell. Mutations may be caused by mistakes during cell division. They may be caused by exposure to DNA-damaging agents in the environment. Mutations can be harmful, beneficial or have no effect.

A very small number of boys have a less destructive form of Rett syndrome. Similar to girls with Rett syndrome, these boys are likely to live to adulthood. But they are still at risk of a number of health and behavior problems.

Nearly all cases of Rett syndrome are caused by a mutation of a certain gene. As a result of the mutation, not enough protein is made by the body, or it is made incorrectly. This can cause other genes to function abnormally. Not everyone who has the mutation has Rett syndrome. Scientists have identified mutations in other genes in people who have Rett syndrome. But they are still learning how those mutations cause the disorder. Scientists believe the remaining cases may be caused by partial gene deletions. They also may be caused by other genes that have not yet been identified.

Diagnosis

Rett syndrome may be hard to notice at first. A health care provider may first become aware of it when a parent shares concerns about a child’s motor or language development. Diagnosing Rett syndrome requires careful observation of your child's growth and development. You may be asked questions about her or his medical and family history.

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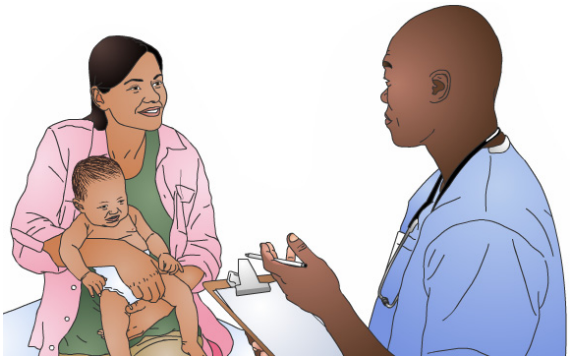
Your child may also have tests to identify conditions that can cause some of the same symptoms as Rett syndrome. Some of these conditions include:

- Autism.
- Cerebral palsy.
- Hearing or vision problems.
- Epilepsy.
- Disorders that cause the brain or body to break down.
- Brain disorders caused by trauma or infection.
- Prenatal brain damage.



There are several tests that may be used to diagnose Rett syndrome. The type of test your child needs depends on his or her particular symptoms.

Tests used to diagnose Rett syndrome may include:

- Blood tests.
- Urine tests.
- Tests to measure the speed of impulses through a nerve.
- Imaging tests.
- Hearing tests.
- Eye and vision exams.
- Electroencephalograms.
- Genetic testing.

Imaging tests, such as magnetic resonance imaging (MRI) or computerized tomography (CT) scans, take pictures of areas inside the body. Electroencephalograms, or EEGs, are brain activity tests. They test electrical activity in the brain. Electrodes are placed on the skin covering the top of the head. Impulses are recorded by a machine.

Your health care provider may recommend a genetic test known as DNA analysis. This test can confirm a diagnosis of Rett syndrome. DNA analysis requires a small amount of blood to be taken from a vein in your child's arm. The blood is then sent to a lab. Technicians will examine your child's DNA. They will look for clues to the cause and severity of your child's disorder.

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If your child's health care provider still suspects Rett syndrome, he or she will use specific guidelines for diagnosis. These guidelines are based on the signs and symptoms your child has shown. In some cases, children may have many of the signs and symptoms of Rett syndrome but not all of them. This is known as variant or atypical Rett syndrome.

Treatment

Although there is no cure for Rett syndrome, potential treatments are being studied. Current Rett syndrome treatment focuses on improving movement and communication skills. Treatment also focuses on providing care and support for affected children and their families.

Treating Rett syndrome requires a team approach, including:

- Regular medical care.
- Medications to control symptoms, such as seizures and muscle stiffness.
- Physical, occupational and speech therapy.
- Academic, social and job training services.
- Nutritional support.

Physical therapy and the use of braces or casts can help children who have scoliosis. It can also help build walking skills, balance and flexibility. If repetitive arm and hand movement is a problem, splints that restrict motion may help.

Occupational therapy is used to teach life skills. It can help children develop skills such as dressing, feeding and making arts and crafts. Speech therapy can help improve a child's life by teaching nonverbal ways of communicating. A child may learn to use his or her eyes and hands to communicate.

Some children with Rett syndrome may need a high-calorie, well-balanced diet. Others may need to be fed through a tube placed in the nose or directly in the stomach. Children with Rett syndrome need help with most daily tasks, such as eating, walking and using the bathroom. This constant care can be exhausting and stressful for families.

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You may arrange for outside help. If you care for your child at home, seek the help of outside caregivers. They can give you a break from time to time. You may consider residential care at some point, especially when your child becomes an adult.

Getting to know other families facing problems similar to yours also can help you feel less alone.

Alternative Treatment

Because Rett syndrome is a severe and incurable disorder, parents may search for complementary and alternative treatments. The goal of these treatments is to improve a child's quality of life.

Complementary and alternative medicine (CAM) are used in addition to or instead of standard care. These practices generally are not considered standard medical approaches. Standard treatments go through a long and careful research process to prove they are safe and effective. But less is known about most types of CAM.

Examples of therapies that have been tried in children with Rett syndrome include:

- Acupuncture.
- Chiropractic treatment.
- Myofascial release, a massage therapy that helps loosen stiff muscles and joints.
- Yoga, a practice that uses exercise and meditation to balance mind and body.

Acupuncture is used to control pain and other symptoms. Thin needles are inserted through the skin at specific points on the body.

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Chiropractic treatment is a type of therapy that uses hands to manipulate the spine or other parts of the body. Heat and ice, relaxation techniques and exercise may also be used.

- Animal-assisted therapy.
- Auditory integration training, which uses sound frequencies to treat speech and language problems.
- Music therapy.
- Hydrotherapy, which involves swimming or moving in water.

Animal-assisted therapy is a type of therapy that uses dogs or other pets to improve the physical and mental health of patients with certain acute or chronic diseases.

There is not much evidence that these approaches are effective. But some parents who have used them report good results. If you think these therapies might help your child, ask your health care provider about the possible benefits.

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Most babies with Rett syndrome seem to develop normally at first. Symptoms begin after 6 months of age. Over time, children with Rett syndrome have problems with movement, coordination and communication.

Smaller than normal head size often is the first sign that a child has Rett syndrome. Delayed growth in other parts of the body also may happen. Rett syndrome is rare. It is estimated to affect one in every 10,000 to 15,000 female babies. It affects all racial and ethnic groups worldwide.

Although there is no cure for Rett syndrome, potential treatments are being studied. Current Rett syndrome treatment focuses on improving movement and communication skills. Treatment also focuses on providing care and support for affected children and their families.

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