CONDITION

Rett syndrome

Rett syndrome is a rare genetic condition that affects brain development, movement, communication, breathing, and other body functions.

WHAT IT IS
Genetic condition affecting brain development and body control

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Lost hand skills, reduced speech, repetitive movements, walking problems

WHAT CAUSES IT
Genetic, usually a new MECP2 gene change

HOW IT IS DIAGNOSED
Clinical criteria and MECP2 genetic testing

MAIN TREATMENT
Supportive therapies and symptom-specific medicines

CAN IT BE CURED?
No - current treatments manage symptoms only

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or prolonged seizure

LONG-TERM OUTLOOK
Lifelong disability; many people live well into adulthood

Overview

Rett syndrome is a lifelong condition affecting brain development. It mainly occurs in girls, although boys and people of other sexes can have it. It is sometimes shortened to RTS.

Most children with Rett syndrome appear to develop as expected during early infancy. Development then slows, usually followed by a period when the child loses skills. This is called developmental regression. Purposeful hand use, speech, balance, and walking may be affected.

Rett syndrome can also affect breathing, digestion, sleep, heart rhythm, bone strength, and the body's automatic control systems. Intelligence is difficult to measure because limited speech and hand control can hide what a person understands.

The condition requires long-term support, but abilities and needs differ widely. Many people with Rett syndrome remain socially interested and learn to communicate through eye gaze, facial expression, pictures, or communication technology.

Symptoms

Symptoms often become noticeable between 6 and 18 months of age, although the timing is not the same for everyone. Early signs may include slower head growth, reduced interest in toys, delayed movement, or unusual hand movements.

The most characteristic changes include:

Other possible symptoms include poor coordination, low muscle tone early in life, later muscle stiffness, and difficulty planning movements. Some people develop tremors or episodes of sudden muscle tightening.

Breathing patterns may become irregular while the person is awake. They can include breath-holding, rapid breathing, air swallowing, or forceful breathing out. Breathing is often more regular during sleep.

Seizures are common, but staring, shaking, or breathing episodes are not always seizures. Recording an episode on video can sometimes help the clinical team understand what is happening.

Constipation, reflux, chewing or swallowing difficulties, sleep problems, teeth grinding, cold hands and feet, anxiety, and episodes of unexplained distress may also occur. Scoliosis, meaning a sideways curve of the spine, can develop over time.

- Partial or complete loss of purposeful hand skills
- Loss or major reduction of spoken language
- Repetitive hand movements, such as wringing, clapping, tapping, or bringing hands to the mouth
- Unsteady, stiff, or absent walking

Causes and risk factors

Rett syndrome is usually caused by a harmful change in the MECP2 gene. This gene helps control the activity of many other genes involved in brain development and function.

MECP2 is on the X chromosome. Most girls have two X chromosomes, while most boys have

one. Differences in how X chromosomes work help explain why symptoms can differ greatly between people. Boys with an MECP2 change may have severe symptoms, but milder and atypical forms are also possible.

In most cases, the gene change happens by chance in an egg or sperm or shortly after conception. Nothing a parent did during pregnancy caused it. Most cases are not passed down through a family, although inherited cases can occur rarely.

A small number of people with Rett-like symptoms have changes in other genes or do not have an identified genetic cause.

Diagnosis

Rett syndrome is diagnosed from the pattern of development and symptoms. The central feature is a period of regression followed by typical problems with hand use, communication, walking, and repetitive hand movements.

A genetic blood test can look for a harmful MECP2 change. Finding one supports the diagnosis and may help clarify the likely form of the condition. However, Rett syndrome remains a clinical diagnosis: not every person with typical features has an identifiable MECP2 change, and an MECP2 change can sometimes cause a different pattern of symptoms.

Tests may also be used to assess health needs or exclude other causes. These can include hearing and vision checks, an electroencephalogram to measure brain electrical activity, heart rhythm testing, sleep studies, and swallowing or nutrition assessments. Brain imaging is not usually enough to confirm Rett syndrome.

Genetic counselling can explain the test result, the small chance of recurrence in another pregnancy, and testing options for relatives when relevant.

Treatment and management

There is no cure for Rett syndrome. Treatment aims to preserve abilities, support communication and movement, improve comfort, and manage specific health problems. Care often involves a coordinated team with experience in developmental and physical disabilities.

Physical therapy can support mobility, posture, joint movement, and balance. Braces, standing equipment, walking aids, or wheelchairs may help a person move safely and take part in daily life. Regular assessment is important because stiffness, foot position, and scoliosis can change over time.

Occupational therapy focuses on hand use, seating, personal care, and practical activities. Speech and language therapy may address both communication and swallowing. Many people benefit from communication systems controlled by eye gaze because looking can be easier than pointing or speaking.

Nutrition support may include changing food texture, adjusting mealtime positioning, or using higher-calorie foods when growth is poor. If eating is unsafe or does not provide enough nutrition, tube feeding may be discussed. Constipation and reflux often need active management.

Medicines may be used for seizures, sleep problems, muscle stiffness, constipation, reflux, or other symptoms. Choosing a medicine requires attention to side effects, swallowing ability, breathing, and possible heart rhythm changes.

Trofinetide is approved in the United States for Rett syndrome in adults and children aged 2 years and older. It may reduce some core symptoms but does not cure the condition or restore lost development. Diarrhoea, vomiting, and weight loss are important possible side effects. Availability and approval differ between countries.

Regular care may include monitoring of growth, nutrition, spine shape, hip position, bone health, walking, seizures, breathing, sleep, and heart rhythm. Dental care is also important because teeth grinding and difficulty with mouth care can cause problems.

Complications

Swallowing difficulties can allow food or liquid to enter the airway. This is called aspiration and can contribute to chest infections. Poor intake, reflux, air swallowing, and constipation may affect comfort and weight.

Reduced movement and nutrition problems can weaken bones, increasing fracture risk. Scoliosis may eventually affect sitting, comfort, or breathing. Some people develop significant muscle stiffness or lose the ability to walk.

Epilepsy can require long-term treatment. Irregular breathing and changes in the electrical recovery of the heart may also need monitoring. Not everyone develops these complications, and their severity differs considerably.

Pain can be difficult to recognise when someone cannot describe it. Changes in sleep, expression, movement, appetite, or behaviour may be signs of discomfort rather than part of Rett syndrome itself.

Prevention

Rett syndrome usually cannot be prevented. Most MECP2 changes happen by chance and are not caused by lifestyle, medicines, infection, or anything a parent did.

After a diagnosis, genetic counselling can help a family understand why the condition occurred and the chance of it happening again. That chance is usually low but is not always zero because a parent can rarely carry the gene change in some reproductive cells.

Early therapy and health monitoring do not prevent Rett syndrome, but they may reduce avoidable complications and help maintain comfort, movement, nutrition, and communication.

Living with it and what to expect

The course of Rett syndrome is often described in stages, but real life does not always follow a fixed sequence. After early development, there is usually a period of regression. This may be followed by a more stable period in which social engagement, attention, and communication can improve even when speech and hand control remain limited.

The rapid loss of skills does not usually continue at the same rate throughout life. Some people regain limited hand use or walking ability, while others do not. Later in childhood or adulthood, stiffness, weakness, scoliosis, and reduced mobility may become more important.

Communication can continue to develop for years. A person who cannot speak may still understand language, make choices, enjoy humour, form close relationships, and express clear preferences. Consistent access to a suitable communication system can make a major difference at home, in education, and in the community.

Most people need help with daily activities throughout life. Some walk independently, some walk with support, and others use wheelchairs. Educational and adult services should focus on participation, communication, comfort, and meaningful activities rather than assuming limited understanding.

Many people with Rett syndrome live into adulthood, including middle and later adulthood. Lifespan can be shortened by complications such as severe epilepsy, swallowing problems, chest infections, reduced mobility, or heart rhythm abnormalities. An individual lifespan cannot be predicted from the diagnosis alone.

When to get medical help

Arrange routine follow-up with the clinical team for changes in walking, posture, spine shape,

sleep, constipation, feeding, weight, communication, or seizure control. New staring, shaking, or stiffening episodes should be assessed rather than assumed to be part of Rett syndrome.

Seek prompt medical advice for:

Call emergency services if the person has severe trouble breathing, turns blue or grey, cannot be awakened, or has a seizure lasting more than five minutes unless their emergency seizure plan gives different instructions. Emergency help is also needed for repeated seizures without recovery, serious choking, or a severe injury.

Breath-holding can occur in Rett syndrome, but a new, prolonged, or severe breathing episode still needs urgent assessment.

- A first possible seizure or a major change in known seizures
- Frequent coughing, choking, or wet-sounding breathing during meals
- Ongoing vomiting, diarrhoea, poor drinking, or signs of dehydration
- Unexplained weight loss, persistent pain, or a sudden loss of function
- A possible fracture, especially after a minor injury

Common questions

Is Rett syndrome a type of autism?

No. Rett syndrome is a distinct genetic condition. During regression, a child may lose social interaction and communication skills in a way that resembles autism. Social interest often improves later, while movement and communication difficulties remain.

Can boys have Rett syndrome?

Yes. Rett syndrome is much less often recognised in boys, but it can occur. Symptoms may be severe because boys usually have only one X chromosome. Some boys have a less severe course because of a particular MECP2 change, a mixture of affected and unaffected cells, or an extra X chromosome.

Is Rett syndrome inherited from a parent?

Usually not. Most cases result from a new MECP2 change that occurred by chance. Rarely, a parent carries the change or has it in some egg or sperm cells. Genetic counselling provides advice based on the family's actual test results.

Can someone with Rett syndrome learn to talk or walk again?

Some people regain limited skills or make progress after the regression period. Walking may be maintained or relearned with support, but others never walk or lose this ability later. Spoken

language usually remains very limited. Eye-gaze systems and other communication methods can reveal abilities that speech cannot show.

Does Rett syndrome keep getting worse?

The condition changes over time, but it is not usually a continuous rapid decline. Regression is often followed by a long period of relative stability. Mobility, muscle stiffness, scoliosis, and other physical problems may worsen later, while attention and communication may continue to develop.

What is the life expectancy with Rett syndrome?

Many people live well into adulthood. It is not possible to predict one person's lifespan from the diagnosis alone. Mobility, nutrition, swallowing, seizures, breathing, heart rhythm, and access to ongoing care all influence long-term health.

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