CONDITION

Rubinstein-taybi syndrome

Rubinstein-Taybi syndrome is a lifelong genetic condition affecting development, growth, physical features, and several body systems.

WHAT IT IS
Lifelong genetic condition affecting growth and development

BODY SYSTEM
Brain, skeleton, heart, digestion, hearing, and vision

MAIN SYMPTOMS
Developmental delay, broad thumbs, short stature, distinctive facial features

WHAT CAUSES IT
Usually a new genetic change

HOW IT IS DIAGNOSED
Clinical assessment and genetic testing

MAIN TREATMENT
Developmental support and treatment of specific health problems

CAN IT BE CURED?
No - the genetic change is lifelong

IS IT AN EMERGENCY?
Sometimes - severe breathing difficulty needs emergency care

LONG-TERM OUTLOOK
Lifelong support needs, with many people reaching adulthood

Overview

Rubinstein-Taybi syndrome is a rare genetic condition present from birth. It affects development, learning, growth, and several parts of the body.

Common physical features include broad thumbs and first toes, short stature, and a recognizable pattern of facial features. Many people have intellectual disability, meaning they learn and understand information more slowly than expected for their age. Speech often develops later than other skills.

The condition can also affect feeding, hearing, vision, breathing during sleep, the heart, kidneys, bones, and skin. The combination and severity of these effects differ from person to person. Some people need substantial lifelong support, while others gain a greater level of independence.

Early recognition matters because hearing problems, heart differences, feeding difficulties, and other treatable concerns can otherwise interfere with health and development. Regular follow-up is usually coordinated across several specialties.

Symptoms

Features may be noticed during pregnancy, at birth, or during early childhood. A baby may have low muscle tone, feeding difficulties, slow growth, or unusual thumb and toe shape. In other children, developmental delay is the first concern.

Common features include:

Speech may be especially delayed. A person may understand more language than they can express. Gestures, pictures, signs, or electronic communication devices can help.

Some people have congenital differences, meaning changes that developed before birth. These may involve the heart, kidneys, genitals, spine, knees, or other joints.

Other possible concerns include hearing loss, crossed eyes, drooping eyelids, severe short-sightedness, dental crowding, frequent ear or respiratory infections, and obstructive sleep apnea. Sleep apnea happens when the airway repeatedly narrows or closes during sleep, causing snoring and pauses in breathing.

Not everyone has all these features. Physical appearance alone cannot show how someone will learn, communicate, or manage daily activities.

- Broad, sometimes angled thumbs and first toes
- Short height and a smaller-than-average head size
- Delayed movement, speech, learning, and self-care skills
- Distinctive facial features, such as arched eyebrows and a prominent nose
- Low muscle tone during infancy
- Feeding difficulties, reflux, or constipation
- A friendly or sociable manner, although behavior differs widely

Causes and risk factors

Rubinstein-Taybi syndrome is usually caused by a change in either the CREBBP or EP300 gene. These genes help control how many other genes are switched on and off during growth and development.

Most identified cases involve CREBBP. A smaller number involve EP300. Changes in these genes can produce overlapping features, although there may be differences between individuals.

The condition follows an autosomal dominant inheritance pattern. This means one altered copy of the gene is enough to cause the condition. In most people, the genetic change happened for the first time in the egg or sperm, or soon after conception. It was not caused by anything the parents did during pregnancy.

Less often, the genetic change is inherited from a parent who has the condition. An affected parent has a 1 in 2 chance of passing the altered gene to each child.

When neither parent has the genetic change in a blood sample, the chance of it happening again is usually low but not zero. Rarely, a parent can have the change in some egg or sperm cells without having it in their blood. A genetics professional can explain recurrence chances for a particular family.

Diagnosis

Diagnosis begins with a medical and developmental assessment. A clinician may look for the typical pattern of broad thumbs and first toes, facial features, growth differences, and developmental delay. Pregnancy, birth, feeding, learning, and family history may also be reviewed.

Genetic testing can confirm the diagnosis when it finds a disease-causing change in CREBBP or EP300. Testing may include:

A negative genetic test does not always rule out the condition. Current tests may not find every relevant genetic change. In some cases, the diagnosis remains based on clinical features.

After diagnosis, assessments are often used to identify related health needs. These may include heart and kidney imaging, hearing and eye examinations, growth and feeding reviews, developmental assessment, and evaluation for sleep apnea. The tests needed depend on the person's age, symptoms, and previous results.

- Sequencing, which looks for small changes within a gene
- Deletion and duplication testing, which checks for missing or extra genetic material
- A multigene panel, exome test, or genome test when the diagnosis is unclear

Treatment and management

There is no treatment that removes the underlying genetic change. Care focuses on development, communication, comfort, independence, and specific medical problems.

Early developmental support may include physical therapy, occupational therapy, and speech and language therapy. School-age children may benefit from an individualized education plan. Communication support should not wait for speech to develop; signs, picture systems, and communication devices can reduce frustration and help participation.

Feeding support may involve positioning, texture changes, or help from a feeding therapist and dietitian. Reflux and constipation can be treated. Some children need temporary or longer-term tube feeding when eating by mouth is not safe or does not provide enough nutrition.

Hearing aids, glasses, eye treatment, or ear procedures may be recommended when needed. Sleep apnea can require treatment from an ear, nose, and throat or sleep specialist. Heart, kidney, dental, spine, knee, or genital differences are managed according to their type and effect.

Behavioral support can help with anxiety, attention problems, repetitive behavior, or sensitivity to noise and crowds. Changes in behavior should not automatically be assumed to be part of the syndrome. Pain, constipation, sleep loss, hearing problems, or another medical issue may be contributing.

Genetic counselling can help a person or family understand test results, inheritance, family planning options, and what results may mean for relatives.

Any planned operation or sedation requires careful preparation. Rubinstein-Taybi syndrome can be associated with airway differences, sleep apnea, reflux, and heart conditions. The anesthesia team should know about the diagnosis and previous anesthesia experiences.

Complications

Possible complications reflect the different body systems that may be affected. They do not occur in everyone.

Feeding and swallowing problems can lead to poor growth or food entering the airway. Later in childhood or adulthood, weight gain may become a concern, especially when mobility is limited.

Untreated hearing or vision problems can further affect communication and learning. Sleep apnea may lead to poor sleep, daytime behavior changes, headaches, or strain on the heart and lungs.

Joint looseness, kneecap instability, curvature of the spine, or other bone differences may

cause pain and limit movement. Some people develop thick raised scars called keloids, sometimes after minor skin injury or surgery.

Benign growths, including certain skin and brain-covering tumors, have been reported more often in people with the condition. Benign means non-cancerous. Current evidence does not support assuming that every person will develop a tumor. New lumps, unexplained neurological symptoms, or persistent changes should be assessed rather than waiting for a routine visit.

Emotional health and family wellbeing can also be affected by communication barriers, repeated appointments, and long-term care needs. Respite services, social support, and practical planning may help.

Prevention

Rubinstein-Taybi syndrome usually cannot be prevented. In most families, it results from a new genetic change that could not have been predicted or avoided.

If the genetic change in a family is known, genetic counselling can explain reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilization. These are personal choices, not required steps.

Good medical care does not prevent the genetic condition, but it may prevent or reduce some complications. Examples include treating hearing loss, addressing unsafe swallowing, managing constipation, and identifying sleep apnea or heart problems.

Living with it and what to expect

Rubinstein-Taybi syndrome is lifelong. Development continues, but usually at a slower pace. Skills may emerge over a longer period than expected, and progress can continue during adolescence and adulthood.

Speech often remains more limited than understanding. Finding an effective way to communicate can improve learning, relationships, and choice-making. Some people learn to read, work with support, take part in community activities, and manage parts of daily life. Others need close help with communication, personal care, safety, and decision-making.

Growth is often slow in childhood. Weight may rise more quickly later, so growth patterns are usually followed over time rather than judged from one measurement.

Health needs also change with age. Feeding and infections may be more prominent in early

childhood. Joint problems, sleep apnea, weight, mental health, and support for adult living may become more important later. Adult care should continue to include hearing, vision, dental, sleep, mobility, and general health needs.

Routine follow-up is useful even when things are stable. It can identify gradual changes that may be hard to notice, such as hearing loss, worsening snoring, spinal curvature, or reduced mobility.

Many people reach adulthood. Life expectancy is strongly influenced by the severity of heart, breathing, feeding, and other medical complications. Long-term planning may include education, work or day services, supported living, finances, legal decision-making, and transition from children's to adult healthcare.

When to get medical help

Arrange routine medical review for new concerns about feeding, growth, hearing, vision, sleep, constipation, pain, mobility, or behavior. Developmental loss-losing a skill that was previously established-also needs assessment.

Seek prompt medical advice for:

Call emergency services for severe breathing difficulty, blue or grey lips, unresponsiveness, or a seizure that lasts several minutes or repeats without recovery. Follow an existing emergency seizure plan if one has been provided.

These emergency signs are not expected simply because someone has Rubinstein-Taybi syndrome, but they require the same urgent response as they would in anyone else.

- Repeated choking, coughing, or breathing changes during meals
- Loud snoring with pauses, gasping, or marked daytime sleepiness
- A new lump, persistent unexplained pain, or a major change in movement
- Signs of dehydration, such as very little urine or unusual drowsiness
- A first suspected seizure or a clear change in known seizures

Common questions

Is Rubinstein-Taybi syndrome inherited?

Usually, it is caused by a new genetic change and neither parent has the condition. Less often, it is inherited from an affected parent. Genetic testing of the person and, when appropriate,

their parents can clarify the pattern.

Can someone with Rubinstein-Taybi syndrome talk?

Many people develop spoken language, but speech is often delayed and may remain limited. Understanding may be stronger than spoken expression. Signs, pictures, or communication devices can support communication at any age.

Can people with Rubinstein-Taybi syndrome live independently?

Some adults manage parts of daily life with limited support. Others require lifelong help with personal care, communication, safety, money, or medical decisions. Childhood test scores alone cannot predict a person's exact adult abilities.

Does Rubinstein-Taybi syndrome get worse over time?

It is not usually a progressive condition that steadily destroys skills. Development generally continues. However, health needs can change, and untreated hearing loss, sleep apnea, joint problems, or other complications may affect function. Loss of established skills should be assessed.

What is the life expectancy?

Many people live into adulthood. There is no single life expectancy that applies to everyone. Outlook is influenced by congenital heart problems, breathing and swallowing difficulties, serious infections, and access to appropriate care.

Is there a cure for Rubinstein-Taybi syndrome?

No. The genetic change remains throughout life. Treatment can still make a meaningful difference by supporting communication and development and by managing hearing, feeding, sleep, heart, mobility, and other health concerns.

Sources

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