CONDITION

TCS

TCS is a genetic condition that affects development of the face, ears, hearing, and airway.

WHAT IT IS
Genetic condition affecting facial and ear development

BODY SYSTEM
Face, ears, hearing, airway, and feeding

MAIN SYMPTOMS
Small jaw, underdeveloped cheekbones, ear differences, hearing loss

WHAT CAUSES IT
Gene changes affecting early facial development

HOW IT IS DIAGNOSED
Clinical examination, hearing tests, imaging, and genetic testing

MAIN TREATMENT
Airway, hearing, feeding, dental, and reconstructive care

CAN IT BE CURED?
No - developmental differences cannot be reversed

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

LONG-TERM OUTLOOK
Most people have typical intelligence and lifespan with appropriate care

Overview

TCS stands for Treacher Collins syndrome. It is a genetic condition that affects how the bones and soft tissues of the face develop before birth. It most often involves the cheekbones, jaw, ears, eyelids, and roof of the mouth.

The effects range from mild facial differences to significant breathing, feeding, or hearing problems. The two sides of the face are usually affected in a similar way, although they may not be exactly the same.

A small lower jaw can leave less room for the tongue and airway. This may make breathing difficult, particularly in a newborn or during sleep. Differences in the outer and middle ear can cause hearing loss. The inner ear is often able to detect sound, but sound may not travel through the outer and middle ear normally. This is called conductive hearing loss.

TCS does not usually affect intelligence. However, untreated hearing loss can interfere with speech, language development, learning, and social communication. Early hearing and communication support can make an important difference.

Care is tailored to the features a person has. It may involve specialists in breathing, hearing, speech, feeding, dentistry, eye care, genetics, and facial surgery.

Symptoms

TCS is present from birth, but mild cases may not be recognised until later. Features can include:

A very small jaw may allow the tongue to sit farther back than usual. This can narrow or block the airway. Signs may include noisy breathing, pauses in breathing, difficulty coordinating sucking and breathing, or poor weight gain.

Obstructive sleep apnoea can occur in children and adults. This means the airway repeatedly narrows or closes during sleep. Possible signs include loud snoring, gasping, restless sleep, morning headaches, daytime tiredness, or behaviour and concentration changes.

Eyelid differences may leave part of the eye less protected. This can cause dryness, irritation, or damage to the clear front surface of the eye.

The severity of one feature does not always predict another. Someone with mild facial differences may still have meaningful hearing loss, while another person with more visible differences may breathe and feed without major difficulty.

- Underdeveloped or flatter cheekbones
- A small lower jaw and chin
- Ears that are small, differently shaped, or absent
- Narrow or closed ear canals
- Conductive hearing loss
- Eyes that slant downward
- Notches or missing tissue in the lower eyelids
- Fewer lower eyelashes
- A cleft palate, meaning an opening in the roof of the mouth
- Dental crowding or a bite that does not line up
- Breathing or feeding difficulty

Causes and risk factors

TCS is caused by a change in a gene involved in early facial development. Genes linked with the condition include TCOF1 , POLR1B , POLR1C , and POLR1D .

These genes help make structures needed by developing cells. A harmful change can reduce the number of cells available to form facial bones and tissues during early pregnancy.

Many cases follow an autosomal dominant inheritance pattern. This means a person can have TCS after inheriting one altered copy of a gene from either parent. An affected parent may have very mild features, so the condition may not have been diagnosed previously.

Some cases result from a new gene change in the egg, sperm, or early embryo. In that situation, neither parent has the change in their blood cells. Less commonly, TCS caused by certain gene changes follows an autosomal recessive pattern, in which both parents carry one altered copy.

The features can differ greatly within the same family. A parent with mild TCS may have a child with more significant breathing or feeding needs, and the reverse can also occur.

Nothing a parent did during pregnancy causes the gene change responsible for TCS.

Diagnosis

TCS may be suspected from the pattern of facial, jaw, eyelid, and ear differences. There is no single examination finding that confirms every case.

A diagnostic assessment may include:

Genetic testing can confirm the diagnosis when a disease-causing change is found. A negative result does not always rule out TCS because current tests may not identify every possible genetic cause.

During pregnancy, ultrasound may show a small jaw or other facial differences, especially when they are pronounced. Mild TCS may not be visible on ultrasound. If a specific family gene change is known, prenatal diagnostic testing may be available. A genetics specialist can explain what a result can and cannot predict, including the uncertainty about severity.

- A physical examination of the face, mouth, ears, eyes, and airway
- Formal hearing tests
- Imaging, such as CT scans, to examine facial bones or plan surgery
- Sleep testing if sleep apnoea is suspected

- Feeding, speech, dental, and eye assessments
- Genetic testing for known TCS-related gene changes

Treatment and management

Treatment does not remove the underlying genetic change. Its goals are to protect breathing, support feeding and hearing, promote communication, and address facial, dental, or eye problems that affect health and daily life.

Breathing is the first priority in a newborn with a narrow airway. Depending on severity, hospital care may include airway support, treatment for sleep-related obstruction, or surgery. Some children need a temporary tracheostomy, which is an opening made in the neck to support breathing. Others may have surgery that gradually lengthens the lower jaw.

Feeding support may involve help with positioning, specialised feeding equipment, or care from a feeding and swallowing team. If eating by mouth is unsafe or does not provide enough nutrition, temporary or longer-term tube feeding may be considered.

Hearing should be tested early. Hearing devices may send sound through bone rather than through a narrowed or absent ear canal. As a child grows, other hearing devices or reconstructive procedures may become options. Speech and language support may be useful, particularly when hearing loss or a cleft palate affects communication.

Cleft palate repair, eyelid procedures, ear reconstruction, cheek or jaw surgery, and dental treatment may be offered at different ages. Some operations are delayed until facial growth has reached a suitable stage. Decisions consider breathing, function, growth, appearance, and the person's preferences.

Regular dental and orthodontic care can help manage crowded teeth and bite problems. Eye care may be needed if the eyelids do not fully protect the eyes.

Emotional support also matters. Visible facial differences can lead to unwanted attention, questions, or bullying. Counselling, peer support, and practical help at school or work may be valuable.

Complications

The most serious complications are related to a narrowed airway. Severe obstruction can interfere with breathing while awake or asleep. Untreated sleep apnoea may affect energy, concentration, behaviour, heart health, and growth.

Hearing loss can delay speech and language development if it is not recognised and supported. Hearing can also change over time, so follow-up testing remains important even after a hearing device is fitted.

Feeding and swallowing difficulty may lead to slow growth or food and liquid entering the airway. Dental crowding and an uneven bite can make chewing and oral hygiene harder.

Eyelid differences can expose the surface of the eye, causing ongoing dryness or injury. Some people also experience distress related to appearance, repeated medical procedures, or social reactions.

Not everyone develops these complications. Monitoring is based on the person's facial structure, symptoms, age, and previous treatment.

Prevention

TCS cannot usually be prevented. It results from an inherited or newly occurring gene change during early development, not from behaviour, diet, or an avoidable exposure.

If TCS runs in your family, genetic counselling can help clarify the likely inheritance pattern and the chance of passing on a known gene change. Testing relatives may sometimes identify people with mild features who did not know they carried the change.

People planning a pregnancy may wish to discuss options such as testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices, and testing cannot reliably predict how mild or severe TCS would be in a child.

Living with it and what to expect

TCS is lifelong. The underlying facial and ear differences do not disappear, but they are not usually a steadily worsening process. Needs change as the face grows, adult teeth appear, communication develops, and personal priorities change.

Infancy may involve close attention to breathing, feeding, and weight gain. During childhood, hearing, speech, dental development, sleep, and school participation often become more prominent. Some reconstructive procedures are staged across childhood, adolescence, or adulthood.

Many people with TCS attend mainstream schools, work, form relationships, and live independently. Intelligence is usually unaffected. Individual experiences still differ, especially when airway, hearing, or feeding problems are substantial.

Adults may continue to need hearing care, dental treatment, sleep assessment, or follow-up after earlier surgery. Some choose additional reconstructive treatment, while others do not. Appearance-related surgery is a personal decision unless a procedure is needed to improve breathing, eye protection, eating, or another function.

Routine follow-up should focus on practical changes, such as new snoring, reduced hearing, difficulty chewing, eye irritation, or problems with a hearing device. Support needs may also change during school transitions, employment, or periods of increased social stress.

When to get medical help

Call emergency services for:

Seek urgent medical assessment for new or worsening noisy breathing, repeated breathing pauses during sleep, difficulty feeding, signs of dehydration, or unusual sleepiness. A baby who cannot feed without coughing, choking, or becoming breathless also needs prompt assessment.

Arrange routine follow-up if you notice reduced hearing, new loud snoring, daytime tiredness, speech difficulties, persistent eye irritation, dental problems, or poor fit or function of a hearing device. These concerns are not always emergencies, but they should not be left until the next distant review.

- Severe difficulty breathing or struggling for each breath
- Blue, grey, or unusually pale lips or skin
- Long pauses in breathing, collapse, or inability to wake normally
- Choking with an inability to breathe, cough, or make sound

Common questions

Is TCS the same as Treacher Collins syndrome?

Yes. TCS is the commonly used abbreviation for Treacher Collins syndrome.

Does TCS affect intelligence?

TCS does not usually affect intelligence. Hearing loss, missed schooling, sleep problems, or communication barriers can affect learning if they are not recognised and supported.

Can TCS get worse with age?

The condition is not usually progressive in the way a degenerative condition is. However, facial

growth, dental development, ageing, and sleep-related airway changes can reveal new needs. Hearing devices and previous surgical results may also require review.

Can someone with TCS have children?

Yes. Some forms can be passed from an affected parent to a child. The chance depends on the gene and inheritance pattern. Genetic testing and counselling can provide information specific to a family, but they cannot predict severity with certainty.

Can TCS be seen on an ultrasound?

Sometimes. Ultrasound may detect a small jaw or other pronounced facial differences, but mild TCS can be missed. Genetic testing may be possible when a specific family gene change is known.

Is there a cure for TCS?

There is no cure that reverses the genetic change or makes facial development typical. Treatment can improve breathing, hearing, feeding, speech, eye protection, dental function, and appearance.

Sources

- Katsanis SH, Jabs EW. "Treacher Collins Syndrome." GeneReviews , University of Washington, Seattle
- MedlinePlus Genetics. "Treacher Collins syndrome." U.S. National Library of Medicine
- National Organization for Rare Disorders. "Treacher Collins Syndrome." Rare Disease Database
- National Health Service. "Treacher Collins syndrome." NHS
- Trainor PA, Dixon J, Dixon MJ. "Treacher Collins syndrome: etiology, pathogenesis and prevention." European Journal of Human Genetics , 2009