CONDITION

Treacher collins-franceschetti syndrome 1

An inherited condition affecting development of the facial bones, ears, hearing, and airway

WHAT IT IS
Genetic condition affecting facial and ear development

BODY SYSTEM
Facial bones, ears, hearing, mouth, and airway

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

WHAT CAUSES IT
Inherited or new TCOF1 gene change

HOW IT IS DIAGNOSED
Clinical assessment and TCOF1 genetic testing

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

CAN IT BE CURED?
No - the underlying gene change remains

IS IT AN EMERGENCY?
Sometimes - severe breathing difficulty or blue lips

LONG-TERM OUTLOOK
Often normal lifespan with appropriate supportive care

Overview

Treacher Collins-Franceschetti syndrome 1 is a genetic condition that affects how the face, ears, and related structures develop before birth. It is often shortened to Treacher Collins syndrome type 1 or TCS1.

TCS1 is caused by a change in a gene called TCOF1 . This gene helps guide early development of facial bones and tissues. Other genetic changes can cause similar forms of Treacher Collins syndrome, but "type 1" specifically refers to the form linked to TCOF1.

The condition most often affects the cheekbones, lower jaw, outer ears, ear canals, eyelids, and roof of the mouth. These differences are usually present on both sides of the face, although one side may be affected more than the other.

The effects range from subtle facial differences to more complex breathing, feeding, hearing,

and eye problems. Intelligence is usually unaffected. However, untreated hearing loss can interfere with speech, language, learning, and social development.

The condition is present from birth and does not spread through the body. Care focuses on protecting breathing and hearing, supporting feeding and communication, and addressing facial or dental differences when useful.

Symptoms

The exact features differ considerably, including among relatives with the same TCOF1 change. Some people have mild signs that are not recognised until another family member is diagnosed.

Common facial and ear features include:

A small lower jaw can leave less space for the tongue. In some newborns, the tongue may fall backward and partly block the airway. Breathing may be especially difficult during sleep or while lying on the back.

Hearing loss is common. It is often conductive hearing loss , meaning that sound cannot move normally through the outer or middle ear. The inner ear and hearing nerve may still work. Hearing loss may affect one or both ears.

Some people have a cleft palate, which is an opening in the roof of the mouth. Others have a high or unusually shaped palate. These differences can contribute to feeding difficulty, nasal-sounding speech, or repeated middle-ear problems.

Dental crowding and an uneven bite can develop as the jaws and permanent teeth grow. Eyelid differences may also leave the eye surface exposed, causing dryness or irritation.

Features affecting the arms, hands, or legs are not typical of TCS1. If limb differences are present, the healthcare team may investigate another craniofacial condition with overlapping features.

- Underdeveloped cheekbones
- A small or set-back lower jaw
- Downward-slanting openings of the eyes
- Notches or missing tissue in the lower eyelids
- Small, unusually shaped, or absent outer ears
- Narrow, closed, or missing ear canals

Causes and risk factors

TCS1 results from a harmful change, also called a pathogenic variant, in TCOF1. The change disrupts early formation of the bones and soft tissues of the face.

It usually follows an autosomal dominant inheritance pattern. This means that one altered copy of TCOF1 is enough to cause the condition. A person with TCS1 has a 1 in 2 chance of passing the altered gene to each child. The chance is the same for every pregnancy.

Sometimes the gene change is inherited from a parent who has clear, mild, or previously unrecognised features. In other cases, it is a new change that began in the egg or sperm or soon after conception. A new genetic change is not caused by anything a parent did during pregnancy.

The severity in a child cannot be predicted reliably from the parent's features. A mildly affected parent can have a more affected child, and the reverse can also happen.

TCS1 affects people of any sex or ethnic background. Having an affected parent or a known familial TCOF1 variant is the main risk factor.

Diagnosis

Diagnosis starts with a detailed physical examination. The clinician looks at facial bone development, jaw size, eyelids, ears, palate, breathing, and feeding. A family history may reveal relatives with similar but less obvious features.

Genetic testing can confirm TCS1 by finding a disease-causing variant in TCOF1. Testing may begin with TCOF1 alone or use a panel that examines several genes linked to similar craniofacial conditions. Occasionally, a person with typical features has no identifiable variant with current testing.

Other assessments show how the condition affects health and development. These can include:

CT imaging can show the facial bones, middle ear, and airway in detail. It is generally used when the findings will help plan treatment or surgery rather than simply to label the condition.

During pregnancy, ultrasound may identify marked jaw or facial differences, but milder TCS1 can be missed. If a familial TCOF1 variant is known, prenatal genetic testing can determine whether a pregnancy has inherited that variant.

- Formal hearing tests, including tests suitable for newborns

- Airway and sleep assessment
- Feeding and swallowing evaluation
- Eye examination
- Dental and orthodontic assessment
- Speech and language evaluation

Treatment and management

There is no treatment that removes the TCOF1 change. Management is tailored to the parts of the body affected and usually involves a craniofacial team. This may include specialists in airway care, hearing, genetics, eye care, dentistry, speech, feeding, and reconstructive surgery.

Breathing is the first priority in an affected newborn. Some babies can breathe safely with positioning and monitoring. Others need breathing support or a procedure to keep the airway open. Sleep studies may be used later to check for obstructive sleep apnoea, in which breathing repeatedly narrows or stops during sleep.

Feeding support may include changes to feeding position, specialised bottles, or temporary tube feeding. A swallowing assessment can identify whether milk or food is entering the airway. A cleft palate may be repaired to support feeding and speech.

Hearing should be assessed early, even if the inner ear appears to work normally. Hearing aids or bone-conduction hearing devices can carry sound past a blocked or absent ear canal. Early access to sound and communication support is important for language development.

Treatment may also include:

Facial and jaw procedures are often staged across childhood, adolescence, and adulthood because the face continues to grow. The best timing and expected benefit differ for each procedure. Surgery is not required simply to make someone look typical; decisions should reflect breathing, eating, hearing, eye protection, comfort, and the person's own goals.

- Speech and language therapy
- Eye lubrication or eyelid procedures when the eye surface is exposed
- Dental care and orthodontic treatment
- Support for jaw growth, bite problems, and chewing
- Reconstructive procedures for the jaw, cheekbones, eyelids, or ears
- Psychological and school support when appearance or hearing affects wellbeing

Complications

A very small lower jaw can cause upper-airway blockage. This may be most important in infancy, during sleep, during respiratory infections, or around an operation.

Obstructive sleep apnoea can lead to poor sleep, morning headaches, daytime tiredness, behaviour changes, or difficulty concentrating. It may occur even when daytime breathing seems comfortable.

Untreated hearing loss can delay speech and language. A child may appear not to listen or may struggle in a noisy classroom because spoken words are unclear.

Feeding and swallowing problems can contribute to slow weight gain. If food or liquid repeatedly enters the lungs, it may cause coughing or chest infections.

Eyelid gaps may allow the clear front surface of the eye, called the cornea, to become dry or injured. Jaw and palate differences can also contribute to crowded teeth, tooth decay, gum problems, and difficulty biting or chewing.

Living with visible facial differences can expose someone to staring, intrusive questions, bullying, or social isolation. Emotional support and contact with others who have craniofacial differences can be useful at any age.

Prevention

TCS1 cannot be prevented through diet, medicines, supplements, or changes during pregnancy. It results from a genetic change that is already present during early development.

Genetic counselling can explain inheritance and testing options without directing reproductive decisions. If the family's TCOF1 variant is known, options may include testing during pregnancy or testing embryos created through in vitro fertilisation before pregnancy. These options provide information or help with reproductive planning; they do not change TCS1 in an existing pregnancy or person.

Relatives with subtle facial or hearing features may choose genetic evaluation, particularly when planning a pregnancy.

Living with it and what to expect

TCS1 is lifelong, but it is not usually progressive. The original facial and ear differences do not keep damaging the body. Their practical effects can change as the face grows, permanent

teeth appear, school demands increase, or sleep and airway needs change.

Infancy may involve the most intensive breathing and feeding care. During childhood, attention often shifts toward hearing, speech, dental development, school access, and planned reconstructive care. Adolescents and adults may consider further jaw, dental, ear, or facial treatment based on function and personal preference.

Many people attend mainstream school, work, form relationships, and live independently. Most have typical intellectual ability. Accessible communication, properly fitted hearing equipment, and suitable classroom seating can make a major difference when hearing is affected.

Follow-up is usually planned rather than urgent. Regular reviews may include hearing tests, dental and orthodontic care, eye checks, growth monitoring, and assessment for sleep apnoea. Devices and treatment plans may need adjustment as a child grows.

The facial structure and genetic change remain throughout life. Procedures can improve breathing, hearing access, chewing, eye protection, or appearance, but they may not remove every difference. Some people need several operations, while others need few or none.

When to get medical help

Call emergency services for:

Seek urgent medical assessment if a baby is working harder to breathe, repeatedly pauses during sleep, cannot take enough feeds, has fewer wet nappies, or coughs and turns dusky during feeding.

Prompt advice is also appropriate for eye pain, marked redness, light sensitivity, or a feeling that the eye surface has been scratched. These symptoms can indicate corneal irritation or injury.

Arrange routine follow-up for changes such as snoring, restless sleep, daytime tiredness, hearing equipment problems, worsening speech clarity, dental crowding, or concerns about growth and feeding. Emotional distress, bullying, or withdrawal also deserves timely support, even when there is no physical emergency.

- Severe trouble breathing, gasping, or an inability to cry or speak normally
- Blue, grey, or unusually pale lips or skin
- A prolonged pause in breathing or difficulty waking
- Choking with an inability to cough or clear the airway

Common questions

Why is it called Treacher Collins syndrome type 1?

The numbered type identifies the genetic cause. Type 1 is caused by a pathogenic variant in TCOF1. Similar facial features can result from changes in other genes, which may be given other type numbers.

Does Treacher Collins syndrome affect intelligence?

Intelligence is usually typical. Hearing loss, speech differences, disrupted sleep, or missed classroom communication can affect learning if they are not recognised and supported.

Does it get worse with age?

It is not usually progressive. However, its effects can change with facial growth, tooth development, sleep, and ageing. Hearing devices, dental treatment, or airway care may need adjustment over time.

Can someone with TCS1 pass it to their children?

Yes. A person with a TCOF1-related autosomal dominant condition has a 1 in 2 chance of passing the altered gene to each child. The severity cannot be predicted from the affected parent's appearance.

Can surgery cure it?

No. Surgery does not remove the gene change. It may improve breathing, chewing, eye protection, facial balance, or other specific concerns.

Can it be detected before birth?

Sometimes. Ultrasound may show pronounced facial or jaw differences, but it can miss mild features. If the family's TCOF1 variant is known, prenatal genetic testing can determine whether the variant is present.

Sources

- GeneReviews: Treacher Collins Syndrome , University of Washington, Seattle, published through the NCBI Bookshelf
- MedlinePlus Genetics: Treacher Collins syndrome , U.S. National Library of Medicine
- Orphanet: Treacher Collins syndrome , INSERM and the Orphanet network
- Chang CC, Steinbacher DM. Treacher Collins syndrome . Seminars in Plastic Surgery . 2012;26(2):83-90



- Trainor PA, Dixon J, Dixon MJ. Treacher Collins syndrome: etiology, pathogenesis and prevention . European Journal of Human Genetics . 2009;17(3):275-283